

Prenatal Diagnosis Center Manual

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California Department of Public Health
Genetic Disease Screening Program

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Chapter 1: Program Overview

1.1. Introduction to the California Prenatal Screening Program

The California Code of Regulations, Title 17, Sections 6523–6532, was developed to implement laws governing the testing of pregnant women for birth defects. The Genetic Disease Screening Program (GDSP) of the California Department of Public Health (CDPH) administers the California Prenatal Screening (PNS) Program to conduct this testing. A guide to GDSP personnel can be found in Appendix A.

The original California Alpha-Fetoprotein (AFP) Screening Program began in April 1986. Among women screened, the Program was successful in detecting 80% of fetuses with open neural tube defects, 97% of fetuses with anencephaly, and 85% of fetuses with abdominal wall defects. However, maternal serum screening for AFP enabled the Program to identify only about 20% of fetuses with Down syndrome. In 1995, the Program added two analytes, human chorionic gonadotropin (hCG) and unconjugated estriol (uE3). Using a 1:190 at mid-trimester cutoff, the Program identified approximately 70% of fetuses with Down syndrome and 60% of fetuses with Trisomy 18. In 2007, the Program added a fourth analyte, Inhibin, to improve the detection of Down syndrome and Trisomy 18 and to lower the *Screen Positive* rates.

In March 2009, GDSP expanded the PNS Program to include First Trimester Screening and Integrated Screening. These expansions provide women a risk assessment for Down syndrome and Trisomy 18 earlier in pregnancy and provide more accurate risk assessments in the second trimester of pregnancy. The PNS expansion

- Captures and utilizes the first trimester analytes hCG and Pregnancy Associated Plasma Protein A (PAPP-A) and Nuchal Translucency (NT) measurements to calculate a risk assessment for Down syndrome and Trisomy 18.
- Incorporates first trimester and integrated screening algorithms and follow-up activities into the Program.

Under the expanded Program, women will be offered five screening options:

1.1.1 First Trimester Combined Screening

Women may undergo first trimester screening and an NT exam to provide the earliest risk assessment for Down syndrome and Trisomy 18. If the preliminary risk assessment is *Screen Negative*, the woman may participate in second trimester screening (Full Integrated Screening) for a refined risk assessment for Down syndrome and Trisomy 18 and to screen for neural tube defects, abdominal wall defects, and Smith-Lemli-Opitz syndrome.

1.1.2 Full Integrated Screening

Women may undergo first trimester screening and an NT exam followed by second trimester screening to provide the most accurate risk assessment for Down syndrome and Trisomy 18.

1.1.3 Serum Integrated Screening

Women who submit blood specimens in the first and second trimester, but who do not undergo an NT exam, will have Serum Integrated Screening. No risk assessment will be provided in the first trimester.

1.1.4 Quad/Quad-NT Screening

For women who do not submit a blood specimen in the first trimester, the Program will still offer second trimester Quad Screening, based on four analytes. The combination of Quad Screening with first trimester NT information increases the accuracy of Down syndrome and Trisomy 18 screening.

1.1.5 NTD/SCD Screening

Women who have had chorionic villus sampling (CVS) and submit a blood specimen in the second trimester will receive a risk assessment for neural tube defects (NTDs) and Smith-Lemli-Opitz syndrome (SCD) only.

1.2. Prenatal Screening Process

Women who choose first trimester screening must have their blood drawn between 10 weeks 0 days and 13 weeks 6 days. The NT exam for risk assessment is performed when the crown-rump-length (CRL) is between 45–84 mm (11 weeks 2 days and 14 weeks 2 days). Second trimester screening must be done between 15 weeks 0 days and 20 weeks 0 days.

A Test Request Form (TRF) is completed by the prenatal care provider and mailed or sent by courier, along with the blood specimen, to one of seven state-approved laboratories, where it is analyzed. The results are then transmitted to the Genetic Disease Laboratory, where they are reviewed for quality control. The demographic data and analytical results are interpreted by the Program, and result mailers are sent to the prenatal care provider.

A Case Coordination Center (CCC) is assigned to all cases with *Screen Positive* results, inadequate blood specimens, cases with missing or inconsistent information, and specimens drawn *Too Early* or *Too Late*. Thirteen CCCs are located throughout California, and prenatal care providers are assigned a CCC based on their ZIP code. A Case Coordinator at the CCC calls the prenatal care provider to confirm or correct all information available in the case. (See List of Case Coordinators in Appendix A.) If the case remains *Screen Positive*, the woman is offered follow-up services at a state-approved Prenatal Diagnosis Center (PDC). These follow-up services are authorized up to 24 weeks' gestation. See Flowchart in Appendix A.

When the prenatal care provider or patient requests information about state-approved PDCs, the CCC is required to list at least three PDCs in the patient's geographic area. (See List of PDCs in Appendix A.) CCCs do not refer patients to a specific center; they respond to requests from the prenatal care provider or the patient.

1.3. Prenatal Care Provider Responsibilities

Prenatal care providers are required to discuss and offer prenatal screening to all pregnant women in California who present for prenatal care by the 20th week of gestation. A patient education booklet is provided by GDSP and is distributed free of charge to all prenatal care providers. The booklet describes the five screening options and contains an informed consent/refusal form. If the patient chooses one of the Prenatal Screening Program options, the current fee (2009) is \$162 for analyte testing and any authorized follow-up. Either MediCal or the patient is billed. In the latter case, the patient must then submit the bill to her own insurance company.

1.3.1 Women with High-Risk Indications

Prenatal care providers should offer a referral to a state-approved PDC to women who are at high risk of a condition detected by prenatal diagnosis, as part of current standard of care. These high-risk indications can include the following:

- History of a previous fetus with a chromosomal defect.
- History of multiple miscarriages.
- Known carriers of chromosome or single-gene disorder.
- Ultrasound findings that suggest a chromosome defect.

1.3.2 Women Age 35 or Older

Women who are 35 years of age or older should be offered a referral to a PDC for genetic counseling to discuss the implications of screening versus prenatal diagnosis. The cost of this counseling is not covered by the Program. Women who decline referral to a PDC should be offered prenatal screening.

1.3.3 Women Who Select CVS

If a woman selects chorionic villus sampling (CVS), she must be offered second trimester screening for the detection of neural tube defects and Smith-Lemli-Opitz syndrome. No risk assessment will be given for Down syndrome or Trisomy 18. Screening is performed between 15 and 20 weeks of gestation, preferably having the blood drawn two weeks or more after the CVS.

1.4. Manual Purpose and Organization

This manual was developed by GDSP and is meant to assist staff at state-approved Prenatal Diagnosis Centers (PDCs) when providing authorized follow-up services to women identified as *Screen Positive* by the California Prenatal Screening (PNS) Program. The manual includes an introduction to PNS results and interpretations, requirements for providing follow-up services, instructions for billing and invoicing, and chart documentation and reporting requirements. In addition, the manual includes PNS Program data and other reference documents.

For more information about the PNS Program, PDC guidelines, or Screening Information System (SIS) protocols, please contact

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Chapter 2: PNS Interpretation Factors and Results

2.1. PNS Interpretation/Risk Assessment

2.1.1 Screening Options and Cut-offs

The California Prenatal Screen Program offers five screening options, depending on what information is available (See Table 2.1.). The cut-offs are different depending on the available information. Genetic Counselors, Ph.D. Medical Geneticists, and Clinical Geneticists should pay close attention to the cut-offs when providing genetic counseling about the prenatal screening results.

Table 2.1: Screening Options and Cut-Offs

Screening Options	T21 cut-off	T18 cut-off	SCD cut-off	NTD cut-off
Option 1: First Trimester Combined Screening for Preliminary Risk Assessment (Follow with Option 2 below) (1 st T blood specimen + NT)	1 in 100	1 in 50		
Option 2: Full Integrated Screening (1 st T blood specimen + NT + 2 nd T blood specimen)	1 in 200	1 in 100	1 in 250	≥ 2.50 MoM for singleton ≥ 4.50 MoM for twin
Option 3: Serum Integrated Screening (1 st T blood specimen + 2 nd T blood specimen)	1 in 200	1 in 100	1 in 250	≥ 2.50 MoM for singleton ≥ 4.50 MoM for twin
Option 4: Quad Screening (2 nd T blood specimen) OR	1 in 150	1 in 100	1 in 250	≥ 2.50 MoM for singleton ≥ 4.50 MoM for twin
Option 4: Quad + NT Screening (2 nd T blood specimen + NT)	1 in 200	1 in 100	1 in 250	≥ 2.50 MoM for singleton ≥ 4.50 MoM for twin
Option 5: NTD/SCD Screening only (2 nd T blood specimen with CVS)			1 in 250	≥ 2.50 MoM for singleton ≥ 4.50 MoM for twin

2.1.2 Risk Assessment and Analytes

Table 2.2 identifies which analytes are required for risk assessment, if maternal age and NT are used, and if a risk assessment is possible for twins.

Table 2.2: Analytes and Risk Assessment

Risk Assessment	Maternal Age	NT	First Trimester Analytes		Second Trimester Analytes				Twins	
			hCG1	PAPP-A	AFP	hCG2	uE3	INH		
T21	Yes	Yes If missing, T21 risk assessment is still possible in 2 nd Trimester	Yes	Yes	Yes	Yes	Yes	Yes If missing, T21 risk assessment is still possible	Yes If missing, T21 risk assessment is still possible	Yes
T18	Yes	Yes	Yes If missing, then hCG2 has to be available	Yes	Yes	Yes	Yes If missing, then hCG1 has to be available	Yes If missing, no risk assessment unless PAPP-A or NT is present		Yes, only if NT is available
NTD					Yes					Yes
SLOS					Yes	Yes	Yes If missing, no risk assessment			
		1st Trimester Analytes and NT PAPP-A = Pregnancy-Associated Plasma Protein A hCG1 = Human Chorionic Gonadotropin NT = Nuchal Translucency				2nd Trimester Analytes AFP = Alpha Fetoprotein hCG2 = Human Chorionic Gonadotropin uE3 = Unconjugated Estriol INH = Inhibin (Dimeric Inhibin A)				

Screen Positive with Values Inconsistent with a Viable Pregnancy

Occasionally, patients will reach the PDC with laboratory results indicating very low or undetectable levels of analytes, especially in the second trimester. These values inconsistent with a viable pregnancy can usually be attributed to (1) non-pregnancy, (2) fetal demise, (3) a blood

specimen mix-up, (4) a male specimen, (5) a very early pregnancy. Before referring the patient to the PDC, the CCC will confirm the status of the pregnancy and identity of the blood specimen with the prenatal care provider. In some situations, the prenatal care provider will be given the option to redraw the blood specimen.

Partial Screening Results for 2nd Trimester

Unconjugated estriol is not a very stable second trimester analyte. If a blood specimen is delayed in transit by 10 days or more, then analysis for unconjugated estriol will not be used in the risk assessment.

2.1.3 Expected Positive Rates for Prenatal Screening Program

Table 2.3 displays the expected positive rates for each screening option, based on each screening indication.

Table 2.3: Expected Positive Rates

Screening Options	Screening Indication			
	T21	T18	SCD	NTD
Option 1: First Trimester Combined Screening (Follow with Option 2 below) (1 st T blood specimen + NT)	2.5%	0.20%		
Option 2: Full Integrated Screening (1 st T blood specimen + NT + 2 nd T blood specimen)	4.50%	0.30%	0.20%	1.00%
Option 3: Serum Integrated Screening (1 st T blood specimen + 2 nd T blood specimen)	4.50%	0.30%	0.20%	1.00%
Option 4: Quad Screening (2 nd T blood specimen) OR	4.50%	0.30%	0.20%	1.00%
Option 4: Quad + NT Screening (2 nd T blood specimen + NT)	3.40%	0.20%	0.20%	1.00%
Option 5: NTD/SCD Screening only (2 nd T blood specimen with CVS)			0.20%	1.00%

2.1.4 Expected Detection Rates

Table 2.4 displays the expected detection rates for each birth defect screened for under the Prenatal Screening Program.

Table 2.4: Expected Detection Rates

Screening Options	Screened Birth Defects					
	T21	T18	Anencephaly	Open Spina Bifida	AWD*	SCD**
Option 1: First Trimester Combined Screening (Follow with Option 2 below) (1 st T blood specimen + NT)	75%	69%				
Option 2: Full Integrated Screening (1 st T blood specimen + NT + 2 nd T blood specimen)	90%	81%	97%	80%	85%	60%
Option 3: Serum Integrated Screening (1 st T blood specimen + 2 nd T blood specimen)	85%	79%	97%	80%	85%	60%
Option 4: Quad Screening (2 nd T blood specimen) OR	80%	67%	97%	80%	85%	60%
Option 4: Quad + NT Screening (2 nd T blood specimen + NT)	89%	72%	97%	80%	85%	60%
Option 5: NTD/SCD Screening only (2 nd T blood specimen with CVS)			97%	80%	85%	60%

*AWD = Abdominal wall defect

**SCD = Smith-Lemli-Opitz, congenital abnormalities, and fetal demise

2.1.5 Screening Results

Table 2.5 displays the screening results as they will appear on the **Case Summary** screen under the Risk/Assessment Interpretation.

Table 2.5: Screening Results

Screening Options	Results/Risk Assessment
Option 1: First Trimester Combined Screening (Follow with Option 2 below) (1 st T blood specimen + NT)	1 st T Combined: Screen Positive ; increased risk for T21, T18, or T21 and T18 <p style="text-align: center;">OR</p> 1 st T Combined: Preliminary Risk Assessment (Acronym: PRA)
Option 2: Full Integrated Screening (1 st T blood specimen + NT + 2 nd T blood specimen)	Full Integrated: Screen Positive ; increased risk for any of the following: T21, T18, NTD, or SCD <p style="text-align: center;">OR</p> Full Integrated: Screen Negative
Option 3: Serum Integrated Screening (1 st T blood specimen + 2 nd T blood specimen)	Serum Integrated: Screen Positive ; increased risk for any of the following: T21, T18, NTD, or SCD <p style="text-align: center;">OR</p> Serum Integrated: Screen Negative
Option 4: Quad Screening (2 nd T blood specimen) OR	2 nd T Quad: Screen Positive ; increased risk for any of the following: T21, T18, NTD, or SCD <p style="text-align: center;">OR</p>
Option 4: Quad + NT Screening (2 nd T blood specimen + NT)	2 nd T Quad: Screen Negative 2 nd T Quad + NT: Screen Positive ; increased risk for any of the following: T21, T18, NTD, or SCD <p style="text-align: center;">OR</p> 2 nd T Quad + NT: Screen Negative
Option 5: NTD/SCD Screening only (2 nd T blood specimen with CVS)	NTD/SCD: Screen Positive ; increased risk for NTD, SCD, or NTD and SCD <p style="text-align: center;">OR</p> NTD/SCD: Screen Negative

NOTE: All first trimester specimens with only one adequate analyte are called “Inadequate specimens” under the Prenatal Screening Program. There are no partial screening results in the first trimester, but there may be partial screening results in the second trimester.

2.2. PNS Interpretation Factor Adjustments

2.2.1 The Prenatal Screening Program makes adjustments for several Interpretation Factors such as maternal weight, status of insulin-dependent diabetes, race/ethnicity, smoking, and number of fetuses. At the beginning of Prenatal Screening expansion in March 2009, no maternal weight or race/ethnicity adjustments will be made for first

trimester specimens. Once the Program establishes the maternal weight and race/ethnicity adjustment factors, they will be provided and updated in this *PDC Manual*.

2.2.2 The concentration of the analytes used for Prenatal Screening is variable depending on the weight of the pregnant woman.

2.2.3 Adjustments must be made for insulin-dependent diabetes, since most of the analytes in the maternal serum tend to be lower. These adjustments are shown in Table 2.6.

Table 2.6: Adjustments for Insulin-Dependent Diabetes for Interpretation Version 14.0

Analyte	Weight-Adjusted MoM is used and then divided by
PAPP-A	1.02
hCG1	0.90
AFP	0.75
hCG2	0.94
uE3	0.92
INH	0.88

2.2.4 Race/Ethnicity: Multiple ethnicities are established by a priority level.

2.2.5 Adjustments for smoking will occur when the smoking question is answered “Yes.” A “Yes” answer for the smoking question means that the patient has smoked cigarettes within the 7 days prior to having her blood drawn for Prenatal Screening. These adjustments are shown in Table 2.7.

Table 2.7: Adjustments for Smoking for Interpretation Version 14.0

Analyte	Race-Adjusted MoM is used and then divided by
PAPP-A	0.827
hCG1	1.03
AFP	1.0
hCG2	0.77
uE3	1.0
INH	1.56

2.2.6 When two fetuses are in a pregnancy, the analyte concentrations are affected; therefore, adjustments must be made. Adjustments for twins are shown in Table 2.8.

Table 2.8: Adjustments for Twins for Interpretation Version 14.0

Analyte	Race-Adjusted MoM is used and then divided by
PAPP-A	1.90
hCG1	2.10
AFP	2.04
hCG2	1.93
uE3	1.64
INH	2.05

NOTE: No twins adjustment is made for T18 or SLOS screening. **However, when an NT/CRL dating method exists for the specimen or case, a T18 risk assessment will be made.**

For Down syndrome, the algorithm used to determine a risk divides each final MoM by approximately two and then gives a risk as if there were a single fetus. This adjustment for twins will not appear on the computer screen or mailer but is simply part of the calculation for determining a risk.

For twins with NT, the risk for a monochorionic pregnancy is the maximum of the two risk calculations. For dichorionic or “unable to determine” chorionicity pregnancies, the risk for the pregnancy is the sum of the two risks (add risks) (e.g., 1:200 + 1:300 is the same as 3:600 + 2:600 = 5:600 or 1:120).

2.3. PEII Case Summary Screen Overview

The **Case Summary** screen in the Screening Information System (SIS) has changed with the Prenatal Screening Program expansion. The **Case Summary** screen now contains three tabs: Case Overview, Interpretation Factors, and Test Results.

2.3.1 Case Overview Tab

This tab displays the valid first trimester and second trimester specimens with their accession numbers, risk assessment interpretations, ovum donor interpretations (if applicable), date when the specimen becomes 24 weeks, and gestational age of the specimen on the date the SIS user is logged on to the Screening Information System (SIS). The label “valid” on a specimen means the specimen contains all the necessary information to obtain a risk assessment.

The **Case Overview** tab first appears when the user navigates to the **Case Summary** screen (i.e., it is the “default” tab). Information on this tab is divided into five sections:

- Valid specimens and associated specimens are included in one section. Valid specimens are used for risk assessment. The term “associated specimen” refers to any specimen received for a particular patient in the first or second trimester after a valid specimen was received (e.g., partial panel result; another first or second trimester blood specimen is received after a valid first or second trimester specimen has been processed). The data displayed for both valid and associated specimens is the Test Requisition Form (TRF) number, blood collection date (BCD), risk assessment interpretation, and tracking status. The information displayed under case notes, alerts, and open HC (Headline Case) is specifically used by the CCC to keep track of activity associated with the accession number. Under the CCC column is the number for the Case Coordination Center to which the accession number is assigned. PDC users should not have access to any information displayed under the column headers for the CCC (e.g., case notes, alerts, and open HC). If the PDC user clicks on the header, a red text message will be displayed.
- Hyperlinks to “Search for Mailers,” “View Case Alerts,” and “Faxing Options” such as sending fax results or fax result mailer PDF are for use of the CCC. **PDCs should not use the hyperlinks and faxing options.**
- Clinician and Client Information sections are readable to the PDC user. If any changes are necessary, the CCC should be contacted.
- Additional information may include the patient’s medical record number for each valid specimen as well as a Medi-Cal number, a Social Security Number (SSN), an alternate date of birth for the patient, and information where the patient had her blood specimen drawn. If any corrections to this section are necessary, the CCC should be contacted.
- The Scanned Document section is for CCC use, and **the PDC should not use any hyperlinks in this section.**

2.3.2 Interpretation Factors Tab

This tab will display only the valid first trimester and second trimester specimens. For each valid specimen, the Interpretation Factors are separated into four sections: Specimen-Specific Interpretation Factors, Gestational Age at Collection, Case-Wide Interpretation Factors, and NT Information.

- Specimen-Specific Interpretation Factors section includes the blood collection date, patient weight in pounds or kilograms, whether the patient had CVS, and whether the patient smokes.
- Gestational Age at Collection is the calculation of the gestational age of the specimen based on the dating method being used.

- Case-Wide Interpretation Factors is information about the patient or pregnancy that is common to the first trimester and second trimester specimens. This data that is common to the pregnancy or pregnant woman, regardless of the trimester of the blood specimen received, is known as Master Data. Master Data includes the patient's date of birth, ovum donor/ovum donor age (if applicable), race/ethnicity, insulin-dependent diabetic status, dating method, number of fetuses, fetal reduction status, and fetal demise greater than 8 weeks' status.
- NT Information includes information about who entered the NT data (e.g., the CCC or the NT Practitioner) and when this data was entered into SIS. This section also displays details about the NT Practitioner (i.e., NT site code and name and NT practitioner name and phone number). The NT data used to calculate the NT information for the risk assessment are NT exam date, CRL GA on NT exam date, twins and chorionicity (if applicable), NT CRL, and NT measurement for fetus A and fetus B (if applicable). The NT MoM per fetus will be displayed.

The NT/CRL refers to the crown-rump length that was acquired during the visit to obtain the nuchal translucency measurement. The NT/CRL used for interpretation should be between 45 mm and 84 mm (which correspond to 11 weeks 2 days and 14 weeks 2 days). The NT/CRL used for gestational dating can be between 10 mm and 84 mm (which correspond to 7 weeks 1 day and 14 weeks 2 days). If the NT/CRL is within the expected range and is available for a first or second trimester specimen, it will be used for the risk assessment calculation.

The Prenatal Screening Program will use any NT value between 0.1 mm and 10.0 mm for risk assessment. An NT measurement of 3.5 mm or greater is usually considered a very high risk for chromosomal abnormalities. The NT Practitioner will make a recommendation to the patient regarding diagnostic testing versus screening. However, if a patient opts for prenatal screening, the Program will still use the NT measurement for risk assessment.

2.3.3 Test Results Tab

This tab on the **Case Summary** screen will provide the first trimester risk assessment, along with the accession number, analyte values and their unadjusted and adjusted multiple of the medians (MoMs) for weight, insulin-dependent diabetic status, race, and smoking.

This tab also displays the interpretation, ovum donor interpretation (if applicable), and the tracking status. Tracking status indicates what is presently occurring with the accession number (e.g., Case/Specimen closed—PDC Appt Kept; 1st T Positive Closed—Clinician Told of Superseded Results).

When a first trimester specimen is superseded by a second trimester specimen, the second trimester risk assessment will appear in a black font, while the first trimester risk assessment for the first trimester specimen will change to a gray font. The second trimester risk assessment will display the similar information as the first trimester risk assessment with regard to analyte values and their unadjusted and adjusted MoMs; however, it will display additional second trimester analytes (i.e., hCG, uE3, AFP, and INH) with their values as well as MoMs.

2.4. Confirming PNS Interpretation Factors

The Prenatal Screening Coordinator at the Case Coordination Center (CCC) confirms the Interpretation Factors for a *Screen Positive* result with the NT Practitioner or prenatal care provider's office. If the Prenatal Screening (PNS) result is still *Screen Positive*, then the prenatal care provider needs to indicate to the CCC if the patient would like follow-up services at a state-approved PDC. The Prenatal Screening Coordinator then refers the patient to the PDC that the prenatal care provider or patient has requested. This referral will appear on the **Cases Referred** screen in SIS under the Follow-Up Center tab for the PDC as a "Not Scheduled" referral. An alert to the PDC Scheduler will also appear on the **View Alert** screen under the **Monitor** tab, indicating the new referral. PDCs should pay close attention to the **Cases Referred** screen for which patients are authorized or still authorized for follow-up services. PDC schedulers and Genetic Counselors should pay close attention to what accession number a patient is authorized under and what service(s) are authorized.

At the state-approved PDC, the patient's name, her demographic information, and her clinician's demographic information from the **Case Summary** screen should be confirmed with the patient. The Genetic Counselor should click the **Interpretation Factors** tab on the **Case Summary** screen to view the Interpretation Factors to be confirmed with the patient. Each Interpretation Factor from the **Case Summary** screen, not from the Prenatal Screening result mailer, **must** be confirmed with the patient and this confirmation **must** be documented in the patient's chart prior to any other follow-up services being provided. Checkboxes for the PDC's use are located next to the Interpretation Factors. Documentation of the Interpretation Factor confirmation **must** be noted by the signature of the person confirming Interpretation Factors, along with the date.

NOTE: Any change(s) to Interpretation Factors **must** be reported to the CCC prior to any other follow-up services being provided (CVS or ultrasound and/or amniocentesis).

The Interpretation Factors from the **Case Summary** screen to be confirmed with the patient include the following:

- **Blood Collection Date:** Confirm the date of blood collection with the patient. She may not specifically know when her blood was drawn for prenatal screening; however, she should be able to remember having her blood drawn for prenatal labs or other blood work on this day.
- **Weight:** Confirm with the patient the number of pounds (lbs) or kilograms (kilos) on the date the blood was collected for prenatal screening.
- **Had CVS:** Confirm with the patient if she had chorionic villus sampling (CVS).
- **Patient Smokes:** Confirm with the patient that she smoked cigarettes within the 7 days prior to having her blood collected for prenatal screening.
- **Date of Birth:** Confirm with the patient her actual date of birth (sometimes patients have an incorrect date of birth on their insurance cards; the date of birth on the

insurance card should not be used for prenatal screening risk assessment. The CCC can be contacted to enter this information.)

- **Ovum Donor:** Confirm with the patient if an ovum donor egg was used for the pregnancy. If the answer is “yes,” then obtain the ovum donor age at donation. The CCC should be contacted with this information. If there is an ovum donor, then SIS will provide a prenatal screening result/risk assessment based on the ovum donor information. If prenatal screening result is *Screen Positive* with either the patient’s date of birth or the age of the ovum donor, then the patient is authorized for follow-up at a state-approved PDC.
- **Ovum Donor Age at Donation:** Confirm with the patient the age of the ovum donor at the time of donation, if applicable.
- **Race/Ethnicity:** Confirm with the patient her race/ethnicity. Typically, if she is 1/2 of a particular race, then include it for risk assessment; however, if she is a 1/4 or 1/8 of a particular race, do not include it for risk assessment. If she has a Hispanic last name because of marriage and she does not have a Latina heritage, she is not Hispanic. A maximum of four races/ethnicities are captured on the **Case Summary** screen.
NOTE: Specific calculations in the prenatal screening algorithm are made for Hispanics, Blacks, Chinese, Koreans, Japanese, Southeast Asians, and Filipinos.
- **Insulin-Dependent Diabetic:** Confirm with the patient if she was on insulin prior to pregnancy. A woman who has gestational diabetes or did not take insulin prior to pregnancy should not be considered an insulin-dependent diabetic for prenatal screening purposes.
- **Dating Method:** Confirm with the patient the gestational dating method
- **Ultrasound:** Confirm with the patient the date and gestational age of the ultrasound. She may not recall the actual date or gestational weeks; however, she should be able to remember if she had an ultrasound and if the date and gestational weeks sound correct. A call to the prenatal care provider is not necessary to confirm this information; however, if an ultrasound report is readily available to the Genetic Counselor, then it can be shown to the patient to confirm if it appears to be a report for this pregnancy.
- **Last Menstrual Period (LMP):** Confirm with the patient if the date is correct.
- **Physical Exam:** Confirm with the patient if the date and gestational weeks are correct. She may not know the exam date or gestational weeks; however, she should recall if she had an OB visit around the date and approximately how far along in the pregnancy the prenatal care provider told her she was. A call to the prenatal care provider is not necessary to confirm this information; however, if a letter from the prenatal care provider is readily available to the Genetic Counselor, then it can be

shown to the patient to confirm if it appears to contain the physical exam information for this pregnancy.

- **Nuchal Translucency (NT):** Confirm with the patient if the date, the NT measurement and crown-rump length (CRL) for each fetus are correct. She may not know the specific measurement or CRL; however, she should recall if she had a NT ultrasound exam and the date she had it. A call to the NT Practitioner or prenatal care provider is not necessary to confirm this information; however, if a NT report is available to the Genetic Counselor, then it can be confirmed with the patient if the report pertains to this pregnancy. If an NT measurement or CRL data entry error exists, the CCC should be contacted. The CRL obtained with an NT measurement will be the dating method used (i.e., you should verify the dating method field has NT-CRL as the dating method) for risk assessment, even if other dating methods are listed on the **Case Summary** screen.
- **Twins:** Confirm with the patient if she is carrying twins. If NT data is included as part of her prenatal screening results, then it is important to report the viability of each fetus in the pregnancy to the CCC.
- **Chorionicity of twins:** Confirm with the patient carrying twins if she knows the chorionicity (only if NT data is used for the prenatal screening risk assessment). If a change in the chorionicity occurs, then this information **must** be reported to the CCC.
- **Number of fetuses:** Confirm with the patient. If LMP or physical exam was the dating method used, then the number of fetuses will typically be “unknown.” If the patient has had an ultrasound or nuchal translucency measurement, she should know the number of fetuses in this pregnancy. If the fetus number changes from “unknown” to “2,” or “1” to “2,” then the fetus number **must** be reported to the CCC. If there are 3 or more fetuses in this pregnancy, then this information **must** be reported to the CCC. Under the Prenatal Screening Program, this patient would no longer be authorized for any other follow-up services (CVS or ultrasound and/or amniocentesis). The prenatal screening results will become *Pregnancy Not Screenable*.
- **Fetal Reduction:** Confirm with the patient if there has been a fetal reduction for this pregnancy. If the answer is “yes,” this information **must** be reported to the CCC. Under the Prenatal Screening Program, this patient would no longer be authorized for any other follow-up services (CVS or ultrasound and/or amniocentesis). The prenatal screening results will become *Pregnancy Not Screenable*.
- **Fetal Demise >8 weeks:** Confirm with the patient that, to her knowledge, she has not experienced a fetal demise at 8 weeks or greater during this pregnancy for any fetus. If the answer is “yes,” this information **must** be reported to the CCC. Under the Prenatal Screening Program, this patient would no longer be authorized for any other follow-up services (CVS or ultrasound and/or amniocentesis). The prenatal screening results will become *Pregnancy Not Screenable*.

NOTE: If a first trimester specimen becomes *Pregnancy Not Screenable*, a second trimester specimen should not be obtained from the patient. If a second trimester specimen is obtained from the patient with first trimester risk assessment *Pregnancy Not Screenable*, then SIS will automatically change the second trimester specimen to *Pregnancy Not Screenable* when the first trimester and second trimester specimens for the patient are matched.

2.5. Changes in PNS Interpretation Factors

Changes to any Interpretation Factor(s) **must** be reported to the CCC responsible for the referral to the PDC before any other follow-up services are provided. Reporting of these change(s) to the CCC **must** occur on the day the change is noted.

If extenuating circumstances occur, then the PDC **must** leave a voicemail message for the CCC responsible for the referral on the day the change occurred and **must** contact the CCC the following day. Detailed documentation (date, time, message left or discussion with CCC, and signature) of these calls to the CCC **must** be in the patient's chart. If change(s) called to the CCC results in a change in the overall prenatal screening result to *Screen Negative*, *Too Early*, *Too Late*, or *Pregnancy Not Screenable*, then only genetic counseling will be reimbursed. The patient is not authorized for further follow-up services (CVS or ultrasound and/or amniocentesis).

2.6. View the Test Results

The **Case Summary** screen has the cut-off values and risk assessments for the patient and the ovum donor (if applicable) under the **Test Results** tab. If any changes are made to the Interpretation Factor(s), the first trimester and second trimester modified risk assessment will be located on the **Test Results** tab. The Genetic Counselor, Ph.D. Medical Geneticist, or Clinical Geneticist who provided genetic counseling should review the Interpretation Factor(s) reported to the CCC to make certain the changes were correctly made. Review of the modified risk assessment, along with Interpretation Factor(s) changed, **must** be documented in the patient's chart, with the date and signature of the person reviewing it.

Chapter 3: Referral and Scheduling at the Prenatal Diagnosis Center (PDC)

3.1. PDC Referrals

For the PDC to access patient results, the Case Coordination Center (CCC) must refer all *Screen Positive* patients for Prenatal Screening (PNS) Program follow-up services to the PDC via the Screening Information System (SIS). Once the patient is referred to the PDC for follow-up services, PDC schedulers will receive an alert in SIS. PDC staff with SIS rights will have access to the results and interpretations on the **Cases Referred** screen.

- If a patient calls to schedule an appointment and indicates it is for a *Screen Positive* result and the PDC has not received access to the results, the PDC must call the CCC to inform them that the patient called the PDC for an appointment.
- If the patient is assigned to one of the PDC sites and then changes the site assignment (even within the same PDC), the CCC must be notified by phone to give the new site access to the patient's result.

The PDC is responsible for making certain that the patient is still referred to them as a PNS patient up to the day when the patient is actually seen. If the patient comes to an appointment and that patient is no longer referred to your PDC, the patient may already have been seen at a different PDC or her referral is no longer indicated due to a refined risk (Integrated Screening). The appointment scheduler will receive a SIS alert indicating that the referral has been cancelled. If the PDC has not received the alert, the PDC is responsible for contacting the CCC to check on the status of the patient before the patient is seen.

Patients who are First Trimester Combined *Screen Positive* are authorized for first trimester services from 11 weeks 2 days to 14 weeks 6 days gestation. Patients who have opted to wait to have services in the second trimester are authorized from 15 to 24 gestational weeks. If a patient has opted to have her blood drawn in the second trimester for Integrated Screening, she will be considered to be *Screen Positive* until the Program receives the patient's second trimester blood specimen. Once an Integrated Screening result is released, the patient would no longer be considered First Trimester Combined *Screen Positive* and services would no longer be authorized based on that result. If the same patient is *Screen Positive* based on her Integrated Screening test result, all services would be authorized under the Second Trimester Integrated Screening test result.

Patients who are *Screen Positive* based on Quad Marker, QUAD + NT, Serum Integrated or Full Integrated Screening are authorized for services from 15 to 24 gestational weeks. The date the patient becomes 24 gestational weeks is provided on the **Case Summary** screen. Services provided after that date will not be covered unless a Special Authorization was approved by GDSP staff.

If a First Trimester Combined patient is referred to a PDC for genetic counseling and the PDC does not have CVS services available, but the patient requests a CVS, a second referral to a second PDC can be obtained by calling a CCC. A Special Authorization is required from GDSP staff if the patient declines genetic counseling at the second PDC and wants only the CVS procedure.

A Special Authorization is also required for split services (genetic counseling and ultrasound at one PDC and ultrasound and amniocentesis at another PDC) for second trimester referrals (Full Integrated, Serum Integrated, Quad Marker, Quad plus NT, NTD/SCD screening).

3.2. Appointment Scheduling

The PDC must schedule all women who have been referred for authorized follow-up services within seven calendar days from the date the patient or clinician contacts the PDC. If the PDC is unable to accomplish this goal, the CCC should be contacted. If the patient chooses to delay her appointment by more than seven calendar days, a note should be put in the patient's chart regarding the reason for a delay in scheduling.

The PDC must update the patient's appointment status with one of the following: "Scheduled," "GC appt kept-DX not scheduled," "Kept," "Error-Appt not Kept," "No Show," or "Cancelled."

The PDC must update the appointment status in SIS with "Scheduled" and the date and time of the appointment. It is preferred that this be done on the same day the patient or doctor has called to make the appointment; however, it must be done within three business days. Additionally, all other changes in the appointment status must be made within three business days. If the appointment status has not been updated within three days, an alert is generated to the appointment scheduler.

If the patient is a First Trimester Combined *Screen Positive* referral and she has a CVS in the first trimester, the patient's appointment status should be updated to "Kept." However, if the patient has not had diagnostic testing for any of the following reasons, the patient's appointment status should be updated to "GC appt kept-DX not scheduled."

- CVS is not available to the patient at the PDC,
- The patient has decided to have amniocentesis in the second trimester,
- The patient has opted to have her blood drawn in the second trimester for Integrated Screening, or
- The patient is undecided between screening or diagnosis.

By updating the status to "GC appt kept-DX not scheduled," the patient will remain on the **Cases Referred** screen. If the patient has opted to have amniocentesis in the second trimester, the patient's appointment should be updated in SIS with the appointment date and a status of "Scheduled." Once the patient completes her follow-up services, the appointment status should be updated in SIS to "Kept".

If the patient has decided to decline any further follow-up services, including having a second trimester specimen, the CCC should be notified to close the case accordingly.

For second trimester referrals, once the patient comes into the PDC, the patient's appointment status should be updated to "Kept." The PDC must update the SIS appointment screen within three (3) business days of the patient being seen.

Patients who either cancel or do not show for their appointment should have their appointment status updated in SIS within three business days. The CCC tracks the patient's status using the information entered on the **Cases Referred** screen, so the CCC can inform the patient's doctor that the patient did not keep her appointment.

3.3. PDC Staff Requirements

The minimum state-approved staff required at each PDC when providing PNS follow-up shall consist of persons qualified to provide genetic counseling, ultrasound, and amniocentesis. If the patient is in her first trimester and chooses a CVS procedure, she must be referred to a PDC that has an approved transabdominal (TA) and/or transcervical (TC) CVS practitioner. The Clinical Geneticist must be available to provide consultation, in person, to all families with abnormal or questionable results. (See PDC Standards 2009 in Appendix A.)

The PDC Director is required to inform GDSP within ten working days regarding any changes in personnel or locations where services are provided. This notification must include a plan to meet the PDC Standards on an interim and permanent basis. Changes in PDC staff must be submitted in writing by the PDC Director or PDC contact to GDSP for approval. (See Entity Protocol Table in Appendix A.) These staff include new Genetic Counselors, Consultative Sonologists, Amniocentesis Practitioners, CVS Practitioners, Appointment Schedulers, PSR Contacts, Invoice Liaisons, and Quarterly Report Contacts. This notification will allow PDC staff to gain access to SIS. It will also provide the appropriate drop-down selections on the Patient Service Report (PSR). GDSP requires new Genetic Counselors, Appointment Schedulers, and Invoice Liaisons to go through a SIS eLearning course and to sign an Oath of Confidentiality, which is kept on file at GDSP. The Oath of Confidentiality is available on the SIS portal page at <http://sis.dhs.ca.gov>.

Confidentiality: All personnel providing PNS follow-up services must keep all PNS information confidential, and information cannot be shared with persons outside the Program. Confidential and privileged information includes, but is not limited to, patient files, patient results, videos, permanent film records, and computer databases. This confidentiality requirement includes all information, software, computer disks, and any other materials provided by GDSP.

When utilizing SIS, a password is issued by GDSP for the use of that designated person only. Only those persons registered with GDSP are allowed access to the GDSP computer. The confidential data, which include Interpretation Factors, are to be accessed and used by the designated user for the sole purpose of completing a result or an interpretation of the result.

If a patient has made an appointment for genetic services that are not PNS Program related, but there is reason to believe that the patient did have a Prenatal Screening test, the PDC should

obtain the results from the patient's prenatal care provider. The PNS result is needed to provide complete and appropriate genetic counseling, just as blood type, Rh, and MCV are needed for diagnostic testing. If results cannot be obtained from the prenatal care provider, the patient must sign a consent for release of her PNS results. This consent must be filed in the patient's chart. Only then can the PNS CCC provide the PDC with a faxed copy of the results.

3.4. Genetic Counseling Requirements

Individualized, face-to-face genetic counseling must be provided to all women referred by the California Prenatal Screening Program. **Telephone counseling alone is NOT acceptable.**

3.4.1 Genetic Counselor Qualifications

All genetic counseling for PNS patients must be provided by qualified persons who are certified or are active candidates for certification in genetic counseling, medical genetics, or clinical genetics by the American Board of Medical Genetics or the American Board of Genetic Counseling. Genetic assistants may not provide genetic counseling to PNS patients, patients with a complicated family history, or patients identified as being at high risk. Genetic counseling students may provide counseling to PNS patients, under the direct supervision of an approved Genetic Counselor; this means that the approved counselor must be in the counseling room with the student at all times.

3.4.2 Charting

In each patient's medical record, an entry documenting the time and place of face-to-face genetic counseling must be made in the medical record of each PNS patient seen. The fact that the patient was offered an opportunity to ask questions about any aspect of the screening and follow-up services, and all questions were satisfactorily answered, should be documented. The risk(s) quoted to the patient are to be clearly stated in the notes. Counseling notes must be signed and dated by the person providing the genetic counseling. In addition, the following should be part of the medical record:

- A review of the **Genetic Risk Assessment** must be documented, signed, and dated by the person providing genetic counseling. (See Genetic Screening Questionnaires [in English and Spanish] in Appendix A).
- A printout of the **Interpretation Factors** screen and the **Test Results** screen, with documentation that the Interpretation Factors have been confirmed with the patient, must be in the chart. The printout must be signed and dated by the person verifying the information with the patient.
- A signed **State-approved informed consent form** must be present in the medical record for each woman who has CVS or amniocentesis. If a woman is undecided or declines the CVS or amniocentesis procedure, she can sign the refusal section of the consent form or a notation can be made in the patient's chart. The prenatal care provider must be notified of the patient's decision. If, at a later date, she decides to proceed with CVS or

amniocentesis, she must then sign a consent form and the prenatal care provider must be notified. Copies of all state-approved consent forms, in various languages, can be found in Appendix A.

- A **genetics summary letter** of the PDC visit is to be sent to the prenatal care provider and a copy retained in the patient's medical record. This letter is to include a summary of the genetic counseling session, the reason(s) for referral, reinterpretations(s), and procedures offered and performed, if applicable.

3.4.3 Counselor Availability

Qualified persons providing genetic counseling must be available on site following ultrasound exams or other procedures to explain the findings and implications to the woman.

3.4.4 Modified Genetic Counseling Requirements

First Trimester Combined patients who decline CVS and choose to come back to the same PDC for diagnostic services in the second trimester are authorized for a Modified Genetic Counseling session to explain the risks and benefits of amniocentesis. The PDC is required to offer the service to the patient, but the patient has the option to decline the service.

3.5. Chorionic Villus Sampling (CVS) Requirements

All CVS procedures must be performed by State-approved Transabdominal CVS (TA CVS) or Transcervical CVS (TC CVS) practitioners, using ultrasound guidance. CVS on PNS Program patients shall be performed at greater than or equal to 11 weeks gestation and not after 14 weeks 6 days gestation, unless expressly approved by GDSP. A Special Authorization may be requested for a placental biopsy performed after 15 weeks by a state-approved TA or TC CVS practitioner.

Any State-approved site that does not have a State-approved practitioner to provide chorionic villus sampling must provide a referral to a site that is authorized to provide CVS. In addition, at centers offering CVS, if either TA or TC CVS is clinically contraindicated or unsuccessful, either an appropriate alternative prenatal diagnosis procedure must be available at that Prenatal Diagnosis Center or a referral must be made to another State-approved Prenatal Diagnosis Center where the alternative procedure is available.

With pregnancies in which there are twins, a State-approved TA or TC CVS practitioner must verify the chorionicity status of the pregnancy and verify it is the same chorionicity on the PNS Interpretation Factor sheet. If the information is different, the CCC must be contacted to recalculate the case in the following situations:

- If the CVS practitioner determines the chorionicity is “monochorionic” and the information on the Interpretation Factors is “dichorionic” or “unable to determine”
- If the CVS practitioner determines the chorionicity is “dichorionic” or “unable to determine” and the information on the Interpretation Factors is “monochorionic”

If the CVS practitioner determines the chorionicity is “dichorionic” and the Interpretation Factors indicate the chorionicity was “unable to determine,” the CCC does not need to be contacted.

3.5.1 CVS Prior to Prenatal Screening

If a patient had a CVS with a successful karyotype prior to having her blood drawn for first trimester screening or prior to having a risk assessment generated through the PNS Program and the patient then has a Combined First Trimester *Screen Positive* result for Down syndrome or Trisomy 18, no follow-up services are authorized.

If the patient has screening in the second trimester and had a CVS with a successful karyotype, the PNS Program will not report screening results for Down syndrome or Trisomy 18. The patient will only be authorized for follow-up services if she is *Screen Positive* for neural tube defects or Smith-Lemli-Opitz syndrome. Follow-up services include genetic counseling, ultrasound, amniocentesis, AF-AFP, or SLOS diagnostic testing on amniotic fluid. Call GDSP to discuss special circumstances, if they exist.

3.6. Fetal Demise Identified During CVS Ultrasound Guidance

A fetal demise identified during CVS ultrasound guidance is **not authorized** for the CVS procedure. The PDC may obtain a Special Authorization for the ultrasound examination from GDSP. If the fetus has abnormalities identified (independent of the demise), a CVS may be performed after the PDC has obtained a Special Authorization from GDSP.

For patients who are First Trimester Combined *Screen Positive* based on a twin gestation, when a fetal demise is noted for one fetus after eight weeks’ gestation, the CCC must be called to report the fetal demise and the first trimester result should be considered *Pregnancy Not Screenable*. Further services are no longer authorized, and the patient should be advised that she should not have her blood drawn in the second trimester of pregnancy to get a refined risk.

For patients who are First Trimester Combined *Screen Positive* based on a twin gestation, when a fetal demise is noted for one fetus before eight weeks’ gestation, the number of fetuses is considered “one,” and the patient can have prenatal screening in the second trimester for a Full Integrated Screening result.

3.7. Ultrasound Requirements

A State-approved Consultative Sonologist must perform a hands-on ultrasound examination on all patients with *Screen Positive* PNS results who are seen in the second trimester, regardless of their referral indication.

The ultrasound examination shall meet the requirements of the American College of Obstetrics and Gynecology, the American College of Radiology, and the American Institute of Ultrasound in Medicine. The following must be determined and documented in the report of the examination: fetal number, fetal presentation, documentation of fetal life, placental localization,

amniotic fluid volume, gestational dating, detection and evaluation of maternal pelvic mass, and a survey of fetal anatomy for malformations.

In addition

- A BPD must be obtained and documented on the PSR for all second trimester referral patients. The only exception is when anencephaly is noted or when the patient is found to be *Too Early*.
- If the entire fetal anatomy cannot be evaluated due to fetal position or maternal conditions, another ultrasound must be offered as part of the follow-up (at no additional charge).
- If the initial ultrasound examination reveals a questionable abnormality, further review by another Consultative Sonologist is required. Also, if after further review of an ambiguous finding, a question remains as to the existence of an abnormality, GDSP may be contacted regarding approval for a second opinion ultrasound exam.
- All ultrasound exams must be reported and interpreted to the woman before she leaves the PDC and to the prenatal care provider within one week of the procedure. The ultrasound report must include the signature of the Consultative Sonologist who performed the exam.
- If the patient is First Trimester Combined *Screen Positive*, Full Integrated *Screen Positive*, or is *Screen Positive* based on Quad Marker and had an NT examination, the gestational dating for the pregnancy will be based on the crown-rump-length (CRL) measurement. Recalculations based on any type of gestational dating by ultrasound performed during the second trimester will not be allowed.

3.7.1 Gestational Dating

For PNS patients who have Serum Integrated Screening or Quad Marker Screening only, biparietal diameter (BPD) must be used for gestational dating, whenever possible. Composite dating is allowed only if the BPD is not available. A reinterpretation must be obtained if there is a greater than or equal to 14 days difference in dating or a multiple gestation is identified before proceeding to amniocentesis. If there is a significant discrepancy of fetal age via BPD versus other parameters, further services may be approved via a Special Authorization from GDSP. See the Redating Policy at State-Approved Prenatal Diagnosis Centers for more details.

3.7.2 Using Biparietal Diameter (BPD) Measurements When Determining Gestational Age (When No NT Exam Information Is Available)

The California Prenatal Screening Program requires Prenatal Diagnosis Centers to use biparietal diameter measurements as the sole indicator of gestational age when dating the pregnancy for screening purposes. The use of BPD increases the rate of detection of open spina bifida at 16–18

weeks by 10% to 12% and reduces false positives by 0.5%. Observations in the 1980's have shown that fetuses with spina bifida have smaller BPDs, resulting in higher multiples of the median (MoMs) of maternal serum AFP.^{1, 2, 3} Therefore, the use of BPD will include more borderline cases in the group called "*Screen Positive for NTD*" and allow women with these results to receive follow-up diagnostic services.

Likewise, the use of BPD can increase the detection of Down syndrome. Fetuses with Down syndrome often have shorter femurs.^{4, 5} If a pregnancy is dated using femur length, the pregnancy may be dated earlier than it should be, giving a *Screen Negative* result that might be *Screen Positive* by BPD. Using BPD eliminates the dating bias in pregnancies with a Down syndrome fetus.

Therefore, for the best screening results, prenatal care providers should use a gestational age based on biparietal diameter. The preference for the use of BPD for prenatal screening purposes does not negate the advantages of using composite or other measurements for other obstetrical purposes should clinicians or consultative sonologists wish to do so.

3.7.3 Redating Policy at State-Approved Prenatal Diagnosis Centers (PDC) for PNS Quad Marker Screening and Serum Integrated Screening Patients

All reinterpretation of PNS Quad Marker Screening and Serum Integrated Screening results must be called to the Case Coordination Center (CCC) before proceeding to amniocentesis or before the patient leaves the center. The following are the redating rules that should apply for women with *Screen Positive* results:

Screen Positive for NTD/AWD; Too Early with High AFP, and Too Late with High AFP

If a PDC ultrasound gestational age differs from the gestational age used to calculate the PNS Quad Marker Screening or Serum Integrated Screening result

- By 14 days or more, a recalculation and new interpretation must be obtained from the CCC. For those cases remaining *Screen Positive* or that become *Too Early with High AFP* or *Too Late with High AFP*, amniocentesis is authorized when the patient is between 15–24 weeks gestation. Amniocentesis before 15 weeks requires a Special Authorization. For those cases converting to *Screen Negative* or *Too Late*, no further services are authorized.
- Between 8–13 days, a recalculation can be obtained at the discretion of the PDC. The new interpretation will be used and saved in the computer as the final result.
- By 7 days or less, a recalculation is not allowed.

¹ Wald, N. et al., British J. of Ob/Gyn, 87:219-21, March 1980.

² Wald, N. et al., British J. of Ob/Gyn, 89:1050-53, Dec. 1982.

³ Cuckle, H.S. and Wald, N., British J. of Ob/Gyn, 94:274-76, March 1987.

⁴ Cuckle, H., Wald N., Quinn J., and Royston, P., British J. of Ob/Gyn, 96:1373-1378, 1989.

⁵ Wald, N., Smith, D., Kennard, A., Palomaki, G.E., Salonen, R. et al., British J. of Ob/Gyn, 100:430-435, 1993.

Screen Positive for Down Syndrome

If a PDC ultrasound gestational age differs from the gestational age used to calculate the PNS Quad Marker Screening or Serum Integrated Screening result

- By 14 days or more, a recalculation and new interpretation must be obtained from the CCC. For those cases remaining *Screen Positive*, amniocentesis is authorized when the patient is between 15–24 weeks gestation. For those cases converting to *Screen Negative* or *Too Late*, no further services are authorized. If the result changes to *Too Early*, the patient should be redrawn between 15–20 weeks gestation. This *Too Early* result must be communicated to the woman’s prenatal clinician in the genetic counseling summary letter. The new gestational age information will also be sent to the prenatal clinician by GDSP.
- Between 0–13 days, a recalculation can be obtained at the discretion of the PDC. If the patient wants an amniocentesis, there is no benefit to recalculating her risk using the PDC ultrasound dating information. If the decision is made to have a recalculation, the new interpretation will be used and saved in the computer as the final result. For those cases remaining *Screen Positive*, amniocentesis is authorized when the patient is between 15–24 weeks gestation. For those cases converting to *Screen Negative* or *Too Late*, no further services, such as amniocentesis, are authorized. If the result changes to *Too Early*, the patient should be redrawn between 15–20 weeks gestation. This *Too Early* result must be communicated to the woman’s prenatal clinician in the genetic counseling summary letter. The new gestational age information will be sent to the prenatal clinician by GDSP.

Screen Positive for Trisomy 18 and/or SLOS

The PNS Program does not recommend redating for Trisomy 18 and/or SLOS risk assessment. However, GDSP will permit a recalculation, at the PDC’s discretion, if the PDC ultrasound gestational age differs from the gestational age used to calculate the PNS Quad Marker Screening or Serum Integrated Screening result by greater than or equal to 14 days and the patient becomes a *Too Early* and requests a redraw. Data entry errors that change the interpretation may be entered.

Screen Positive for More than One Indication

- *Screen Positive for Trisomy 18 and/or SLOS* in combination with any other *Screen Positive* result: Follow the *Screen Positive for Trisomy 18 and/or SLOS* redating rules.
- *Screen Positive for NTD and Down syndrome*: Follow the *Screen Positive for Down syndrome* redating rules. However, note that making the gestational dating earlier only makes the AFP MoM higher, whereas making the gestational dating later makes the Down syndrome risk higher. Even with redating, these patients will usually remain authorized for amniocentesis.

Other Considerations

- While verifying the Interpretation Factors with the patient, PDC counselors are not required to check with prenatal care providers to validate unreported ultrasound exams. However, change(s) to any other Interpretation Factors revealed by the patient must be reported to the CCC before proceeding with follow-up services.
- A Special Authorization for services can be requested from GDSP for any unusual circumstances.

We will continue to evaluate the effectiveness of our PDC ultrasound redating policy, and we encourage your input.

3.7.4 Changes in Gestational Age and Recalculations

If the PDC staff determines that a change in gestational age should be made in accordance with the policies described, the CCC must be contacted for a reinterpretation of the result. If the CCC assigned to this case is unavailable, any CCC listed in Appendix A may be called. Also, GDSP may be called at (510) 412-1502 to perform a recalculation of the results.

Patients should not be seen for PNS follow-up services when the CCC or GDSP is closed, in the event that a recalculation is needed. If the CCC office or GDSP is closed, e.g., after normal business hours, leave a message with the CCC and submit the request for a modification via SIS.

The patient's appointment status must be updated to "Appointment Kept" prior to obtaining a reinterpretation. If any Interpretation Factors change, the PDC must contact the CCC with the changes before proceeding to any additional services.

Whenever the CCC is contacted by the PDC to reinterpret a PNS result and changes are made to the case, a modified mailer is sent to the PDC as well as to the prenatal care provider. This mailer should be reviewed for accuracy and filed in the patient's chart.

3.7.5 Reinterpretation to Too Early/Negative and Ultrasound Findings

In the event a woman's screening results are redated by the PDC ultrasound and the CCC recalculation indicates the patient is *Too Early* or *Screen Negative* but an abnormality is seen, amniocentesis or fetal tissue karyotype may be authorized when, after reasonable effort, no other source of payment is identified. GDSP must be called for a Special Authorization for the service prior to the service being performed. The Patient Service Report (PSR) form must document the abnormal ultrasound finding (see Table 3.1). A notation should also be made in the patient's chart as to the absence of resources for payment.

Table 3.1: Ultrasound Findings to Call for Special Authorization

lemon sign	banana sign
choroid plexus cysts	holoprosencephaly
spina bifida	encephalocele
ventriculomegaly	cerebellar hypoplasia
cystic hygroma	nuchal skin fold thickening >6 mm
congenital heart defect	pleural effusion
omphalocele	gastroschisis
diaphragmatic hernia	duodenal atresia
marked oligo or polyhydramnios	non-immune hydrops fetalis
echogenic bowel	

This list does not constitute a recommendation that amniocentesis be performed in all of these circumstances. PDC personnel should use their clinical judgment regarding the medical appropriateness of amniocentesis. This list reflects GDSP's commitment to pay for services that could not otherwise be obtained.

3.7.6 Isolated Anencephaly or Fetal Demise Noted During Ultrasound

Isolated anencephaly or fetal demise without abnormalities (independent of the demise) is not authorized for amniocentesis or cytogenetic follow-up. However, if other abnormalities are identified, prenatal diagnostic services may be performed only with a Special Authorization from GDSP. The only exception is if a fetal demise is found in the fetus of a woman who is *Screen Positive for Trisomy 18 or SLOS*.

3.7.7 Fetal Demise of a Twin

For patients who are *Screen Positive* based on a twin gestation, when a fetal demise is noted for one or both fetuses after eight weeks gestation, the fetal demise should be documented on the PSR. If this information is discovered in the genetic counseling session, only the genetic counseling will be reimbursed. If the fetal demise is discovered during the second trimester ultrasound examination, only the genetic counseling and ultrasound will be reimbursed. If the patient is *Screen Positive for Trisomy 18 and/or SLOS* and fetal abnormalities are seen on the ultrasound, contact GDSP for a Special Authorization if the patient wants to pursue prenatal diagnostic services.

3.7.8 Second Opinion Ultrasound

If the AFAFP is ≥ 2.0 MoM and/or AChE is positive, then another ultrasound exam for fetal anomalies must be performed at the same PDC by a Consultative Sonologist, at no additional charge to the patient.

If no abnormality is identified, or if there is a question about the ultrasound results, a second opinion ultrasound by an approved Consultative Sonologist must be offered to the patient. She must first be offered a second opinion ultrasound at a different PDC. If she prefers to go to an

approved Consultative Sonologist within the same PDC, but whose practice is financially independent of the provider(s) that performed the initial ultrasound examination, this preference must be documented in the patient's chart. Once the PDC determines where the patient wants to be seen, a Special Authorization must be requested from GDSP. Once the authorization is given, the referral will appear in SIS to the second PDC.

On the rare occasion that a patient disbelieves the findings on the initial ultrasound exam or declines amniocentesis, a Special Authorization for a second opinion ultrasound may be obtained by GDSP.

Consultative Sonologists who perform second-opinion ultrasound exams must confirm that requirements for reimbursement are met as per the Vendor Agreement and that a Special Authorization has been approved by GDSP.

The PDC where the patient was originally seen may contact GDSP if there is a question regarding authorization for the second-opinion ultrasound to be performed at another approved center. The patient's Genetic Counselor is encouraged to contact a Genetic Counselor at the PDC where the second opinion is to occur, to facilitate the process for the patient and to provide the appropriate records/history. The PDC providing the second-opinion ultrasound should ensure that the patient is referred to the PDC in SIS, in order to be reimbursed for services.

3.8. Guidelines for Reporting a BPD or an NT Examination from a PDC during a Non-PNS Appointment

These guidelines are to be used when a patient has gone to the PDC for a non-PNS appointment and

- Has had her blood drawn for first trimester screening by her referring clinician and does not have an NT examination prior to going the PDC; or
- Has already had Serum Integrated or Quad Marker Screening with a *Screen Negative* result, and the PDC performs an ultrasound examination and found that it was not used for PNS screening; or
- Goes to the PDC for a non-PNS referral indication.

These guidelines were developed to facilitate communications between the PDC, referring clinician, and Case Coordination Center (CCC) if a patient has an ultrasound at the PDC and the PDC wishes to obtain a recalculation of the PNS result using the new NT or ultrasound information.

At the time the PDC receives a referral from a clinician, the PDC may want to ask the clinician's office if a prenatal screening test was performed. The PDC staff should first try to obtain the Test Requisition Form (TRF) number or accession number from the referring clinician in advance of the appointment.

Using either the TRF or accession number, the SIS NT Practitioner can enter NT examination information directly into SIS to obtain a preliminary risk assessment. This can be done within a week of the NT ultrasound exam, otherwise the information will need to be called to a CCC.

If the PDC is not provided the TRF or accession number from the referring clinician, the PDC should verify with the patient if a PNS test was performed. If the interpretation is based on an LMP or physical exam dating method, it is advantageous to use ultrasound dating, using crown-rump-length and the NT exam information to obtain a First Trimester Combined, Full Integrated, or Quad plus NT result. BPD is the preferred method of gestational dating for Serum Integrated or Quad Marker Screening if the ultrasound is performed in the second trimester. All calls for recalculation to a CCC are entered into SIS and saved in the case.

If the recalculation indicates that the new interpretation is *Too Early* or *Too Late* or *Screen Negative*, a modified mailer will be sent to the clinician by the PNS Program and it will reflect the PDC's name and phone number as the "contact information." The PDC should also send a summary letter to the clinician indicating the PDC services provided, as well as the new PNS interpretation. The CCC will call the clinician if the new interpretation is *Too Early* or *Too Late*.

If the recalculation indicates that the new interpretation is *Screen Positive for NTD/AWD, Down syndrome, Trisomy 18, or SLOS*, the PDC must call the clinician and verify that the clinician wants the PDC to continue to provide the necessary follow-up services related to the *Screen Positive* result. Some PDCs may have arrangements or an agreement with their referring clinicians, which make phoning the referring clinician each time unnecessary. The PDC should communicate to the CCC that the clinician has agreed to the authorized follow-up services, releasing the PNS results via SIS to the PDC. The PDC should send a summary letter to the clinician indicating the services provided related to the original reason for referral and the PNS follow-up services provided.

NOTE: The PDC should not call the CCC to recalculate a PNS result if the patient is currently past 24 weeks 0 days' gestation, by ultrasound, on the day of the PDC visit.

3.9. Ultrasound Dating Changes after 24 Weeks' Gestation

Occasionally, PNS patients will be seen at the PDC after 24 weeks gestation, and new ultrasound information could modify the result. The date when a patient becomes 24 weeks gestational age is indicated on both the **Cases Referred** screen and on the **Case Summary** screen for the patient.

Patients seen after 24 weeks should have services billed to their insurance unless a Special Authorization was requested from GDSP. If a recalculation is requested or performed, those services are to be billed to the patient's insurance unless the patient recalculates to *Screen Positive* and is seen for follow-up at less than 24 weeks. The patient must be informed that the services provided will be billed to her insurance.

Table 3.2 provides guidelines to use when recalculating the original result:

Table 3.2: Guide to Ultrasound Dating Changes after 24 Weeks Gestation

PDC PATIENTS WHOSE PNS RESULT AT REFERRAL INDICATES A GESTATIONAL AGE (G.A.) \leq24 WEEKS	
A. Current PNS result is <i>Screen Positive</i>	<p>PDC Ultrasound: GA >24 weeks 0 days (assuming the new dating falls within the PDC redating rules)</p> <ul style="list-style-type: none"> • If the patient’s result was <i>Screen Positive for NTD</i>, her result will probably change to <i>Screen Negative</i> or <i>Too Late</i>, or perhaps stay <i>Screen Positive</i> with a lower MoM. Further PNS-funded services are not authorized since the G.A. is past 24 weeks. • If the patient’s result was <i>Screen Positive for Down syndrome</i>, her risk becomes higher, but further PNS funded services are not authorized since the G.A. is past 24 weeks.
B. Current PNS result is <i>Screen Negative, Too Early</i> , or <i>Too Late</i> (The patient is at the PDC for an indication other than a positive PNS result.)	<p>PDC Ultrasound : GA >24 weeks 0 days</p> <p>Do not make any changes to the case. PNS-funded services are not authorized since the G.A. is past 24 weeks.</p>
PDC PATIENTS WHOSE PNS RESULT AT REFERRAL INDICATES A GESTATIONAL AGE \geq24 WEEKS 0 DAYS	
A. Current PNS result is <i>Screen Positive</i> . (Usually the patient is at the PDC because the PDC is performing an ultrasound that is being billed to her insurance.) The CCC may have already closed the case as “Pregnancy Too Advanced for Follow-up” and the patient no longer appears as a referral in SIS – Cases Referred.	<ol style="list-style-type: none"> 1. PDC Ultrasound = \leq 24 weeks 0 days (assuming the new dating falls within the PDC redating rules). <ul style="list-style-type: none"> • If the patient is still Screen Positive by the new ultrasound dating, she may receive PNS-authorized services, as long as they are provided at or prior to 24 weeks 0 days G.A. • If the patient’s result changes to <i>Screen Negative, Too Early</i> or <i>Too Late</i>, no PNS-funded services are authorized. Follow-up services are not billable to PNS unless a Special Authorization was obtained prior to the patient being seen. 2. PDC Ultrasound = REMAINS > 24 weeks 0 days (assuming the new dating falls within the PDC redating rules). <p>The patient’s result can be reinterpreted since the result may change to <i>Screen Negative, Too Early</i> or <i>Too Late</i>. This policy gives the patient the opportunity to possibly modify her PNS result, thus alleviating anxiety. Follow-up services are not billable to PNS unless a Special Authorization was obtained prior to the patient being seen.</p>

<p>B. Current PNS result is Screen Negative, Too Early, or Too Late. (The patient is at the PDC for a non-PNS indication.)</p>	<p>1. PDC Ultrasound = ≤ 24 weeks 0 days (assuming the new dating falls within the redating rules).</p> <p style="padding-left: 40px;">The PDC should call the CCC to enter and save the new ultrasound information in the case. Proceed according to the new interpretation. Further PNS follow-up services may be authorized based on the new <i>Screen Positive</i> result. The PDC should notify the clinician and the PDC, and clinician will receive a modified result mailer from GDSP.</p> <p>2. PDC Ultrasound = REMAINS > 24 weeks 0 days (assuming the new dating falls within the redating rules).</p> <p style="padding-left: 40px;">Do not make any changes to the case. Follow-up services are not authorized by or billable to PNS.</p>
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3.10. Amniocentesis Requirements

All amniocentesis procedures must be performed by State-approved Amniocentesis Practitioners, using ultrasound guidance. Amniocentesis on PNS Program patients shall be performed at greater than or equal to 15 weeks gestation and not after 24 weeks gestation, unless specifically approved by GDSP. In special circumstance, a Special Authorization may be requested for amniocentesis performed between 14 and 15 weeks.

Authorization for analysis of amniotic fluid, including AFP/AChE and fetal chromosome analysis, is restricted to laboratories approved by GDSP. Approved laboratories must comply with applicable GDSP standards and agree to maintain a program of quality control and successfully participate in proficiency testing programs required by GDSP.

All AF-AFP and AChE results must be completed and reported to the PDC within ten days of performing the amniocentesis. Ninety-five percent of all chromosome analyses must be completed and reported to the prenatal care provider within three weeks of performing the amniocentesis. If the AF-AFP result is ≥ 2.0 MoM, it shall be followed by AChE analysis.

3.10.1 Prenatal Diagnosis Prior to PNS Screening

If a patient has CVS or early amniocentesis with a successful karyotype and her PNS result is *Screen Positive for NTD/AWD or SLOS*, then an amniocentesis for AF-AFP and AChE only is authorized. However, if cytogenetic studies were not completed, another fetal karyotype is authorized.

If a patient had CVS or early amniocentesis with a successful karyotype, the PNS Program will not report screening results for Down syndrome or Trisomy 18. Call GDSP to discuss special circumstances, if they exist.

If a woman had amniocentesis at 14 weeks gestation or greater, and then she has PNS Screening with a positive result, no follow-up services are authorized as long as a successful karyotype and AF-AFP results were obtained.

3.11. Fetal Demise or Anencephaly

Isolated anencephaly or fetal demise, without other fetal abnormalities noted, is not authorized for amniocentesis or cytogenetic follow-up. However, if other abnormalities are identified, services may continue with a Special Authorization from GDSP. Other abnormalities do not include conditions associated with fetal demise.

3.12. Special Authorizations

Special Authorizations are approved on an individual case-by-case basis. The PDC must contact GDSP, not the CCC, prior to performing any special services for which they are seeking authorization. PDCs must call for a Special Authorization for the following :

3.12.1 Special Services

Second Opinion Ultrasound:

- When there is an unexplained AF-AFP MoM ≥ 2.0 and/or AChE is positive and the patient has had a second ultrasound from the same PDC.
- If a question remains as to the existence of an abnormality after the consultative ultrasound examination.

Placental Biopsy (procedure must be performed by State-approved CVS practitioners only):

If a PNS patient is indicated for a placental biopsy, the patient must be given appropriate procedure-related risks and limitations. GDSP will reimburse at the amniocentesis rate for authorized procedures.

Fetal Tissue Karyotype:

A fetal tissue karyotype may be authorized in lieu of amniocentesis whenever a woman is authorized for amniocentesis but chooses to terminate the pregnancy.

3.12.2 Special Circumstances:

Patient being seen at ≥ 24 weeks gestation: The request must be made prior to the patient being seen at the PDC.

Patient to be seen at second PDC for split services

Fetal demise on ultrasound with other anomalies

Anencephaly on ultrasound with other anomalies

Patient redates to *Screen Negative, Too Early, or Too Late*, but anomalies present on ultrasound and patient has no insurance

3.13. Services Not Covered by the PNS Screening Program

Parental Karyotypes

GDSP does not authorize payment for parental karyotypes (even if a variant/translocation is found in a fetal karyotype). Third-party payers can be approached for these reimbursements.

FISH

At this time, the PNS Screening Program does not reimburse for FISH studies.

Rhogam

At this time, the PNS Screening Program does not reimburse for the administration of Rhogam.

Other Abnormalities and Case Management

Services relating to the management of the pregnancy (i.e., serial follow-up ultrasounds or fetal echocardiography), termination of the pregnancy, and diagnosis or management for other abnormalities unrelated to PNS results are not reimbursed through the PNS Program. Other resources may be available for these studies. Third-party payers can be approached for these reimbursements. However, prior to the delivery of services, women must be informed of the charges associated with these services.

Unauthorized Services

Services performed on women referred for prenatal diagnostic services other than those authorized by the PNS Program may be billed to the woman and/or third-party payers; however, prior to the delivery of services, women must be informed of the charges associated with the delivery of services. Patients should be provided the Services Covered by the PNS Screening Program consent form. (See Appendix A.)

3.13.1 Guidelines for Drawing PNS at the PDC

These guidelines were developed to facilitate communications between the PDC, referring clinician, and Case Coordination Center (CCC) when a patient needs to have her blood drawn for PNS Screening.

The PDC should develop an arrangement or agreement with its referring clinicians for patients referred to the PDC for the following:

- “Choices” counseling (discussion regarding screening versus prenatal diagnosis) OR
- PNS follow-up counseling and the patient recalculates to *Too Early*

The PDC can choose from three options if the patient opts to have PNS screening or needs to have her blood redrawn. These options are listed below. The PDC needs to communicate to the referring clinician which option was used, both verbally and in the counseling summary letter.

For patients who were *Screen Positive*, had a recalculation to *Too Early*, and are having their blood redrawn, the PDC will also need to verbally communicate with the CCC which option was used. For all options, the ultrasound gestational age by biparietal diameter (BPD) must be used to date the pregnancy for the redrawn specimen.

The PDC should have the patient sign a release of PNS records in the event the PDC will need to obtain results from the CCC. If a patient returns for a non-PNS visit, the PDC should first request the results from the referring clinician. However, if that is not possible, the PDC may fax a signed release of medical records to the CCC a day in advance of the patient's appointment.

- **Option 1: Send patient back to referring clinician to have her blood drawn.**

Help the patient and clinician by giving the patient, in writing, her time window for having her blood drawn and her gestational age by BPD on the day of her PDC ultrasound. **OR**

- **Option 2: Draw her blood at the PDC and put the referring clinician's name, license number, and address on the Test Requisition Form (TRF).**

Do this only if the clinician agrees to this arrangement. If the PDC performed an ultrasound, use the gestational age by BPD for dating on the TRF. The PDC should send out the counseling summary letter quickly, along with the pink copy of the TRF, so the documentation gets to the clinician before the PNS result does. The clinician will get a phone call to verify the Interpretation Factors if the redraw is assigned to a CCC for any reason. Many clinicians are annoyed at having to verify information or provide missing information if the clinician was not involved. **OR**

- **Option 3: Draw her blood at the PDC and put a PDC clinician's name, license number, and address on the TRF.**

The PDC should obtain the information for the TRF from either the patient or the clinician and optimally use the gestational age by BPD to date the pregnancy. The PDC should send the referring clinician the pink copy of the TRF, along with the summary letter regarding the genetic counseling issues and ultrasound results.

Use an address and phone number that is accessible to the CCC on a daily basis. The PDC should set up a file for pending PNS results. Results usually arrive in 1 to 2 weeks. Even if the patient is not returning to the PDC, the CCC should be contacted to obtain the results if they have not been received within 10 days.

- If the result is *Screen Negative*, after the PDC receives the result in the mail, the PDC should send the referring clinician a copy of the result for the patient's prenatal chart.
- If the result is *Screen Positive*, the CCC will call the PDC for verification of all Interpretation Factors and facilitate the referral for follow-up. The CCC will then refer the patient to the PDC in SIS. The PDC will also receive the initial result in the mail. The PDC should contact the patient to schedule the PNS follow-up visit. As

usual, the PDC should reverify the Interpretation Factors with the patient, discuss the results and risk interpretations, and make an offer of ultrasound and amniocentesis. All PNS results, follow-up tests, and results should be communicated back to the referring clinician in the final summary letter. An Outcome of Pregnancy Request will be sent to the PDC on all initial *Screen Positive* cases. The PDC should put the referring clinician's name in the Alternate Provider section of the Outcome of Pregnancy Request and return it to the PNS Program at GDSP.

Chapter 4: Documentation of Follow-Up Services with Patient Service Reports (PSRs)

4.1. Two Ways to Locate/Track the PSR for Completion

Table 4.1 provides instructions for two alternative methods of locating/tracking Patient Service Reports (PSRs) for completion.

Table 4.1: Method of Locating/Tracking PSRs

Follow Up Center Module tab on the **Cases Referred** screen (See Figure 4.1.)

- Click **PSR Status** (second-level link on the left side of the screen). User will be taken to the **View PSR Status** screen.

NOTE: A patient’s name and accession number will stay on the **Cases Referred** screen for only seven days after the appointment has been kept. Then it can be found on the **View PSR Status** screen.

Data Intake Module tab on the **Client/Case Search** screen (See Figure 4.2.)

- Enter the accession number. User will be taken to the **Case Summary** screen.
- Click **Enter PSR** (third-level link located at the top right corner of the screen).
- If you type in the wrong number, click the **Data Intake Module** tab and SIS will clear the accession number boxes.

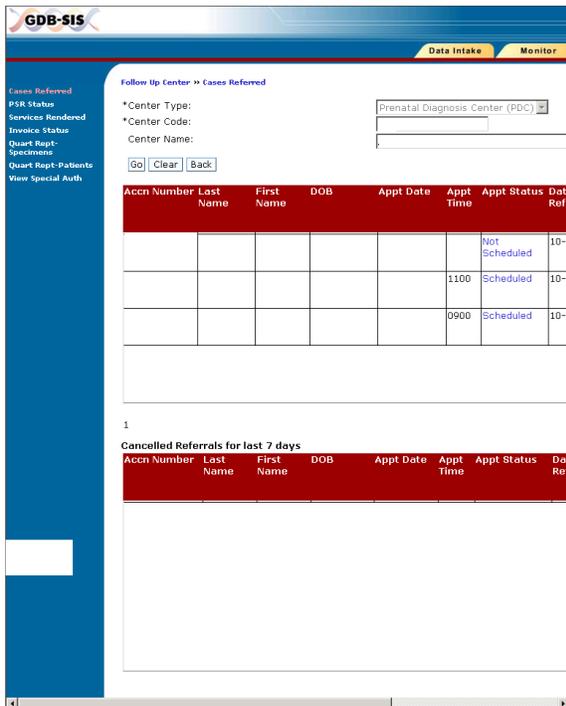


Figure 4.1: Cases Referred Screen

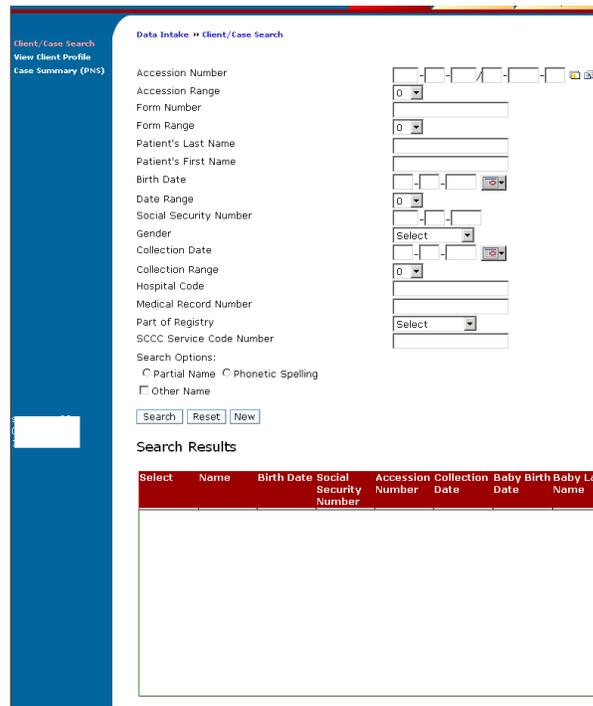


Figure 4.2: Client/Case Search Screen

4.2. Details about the View PSR Status Screen

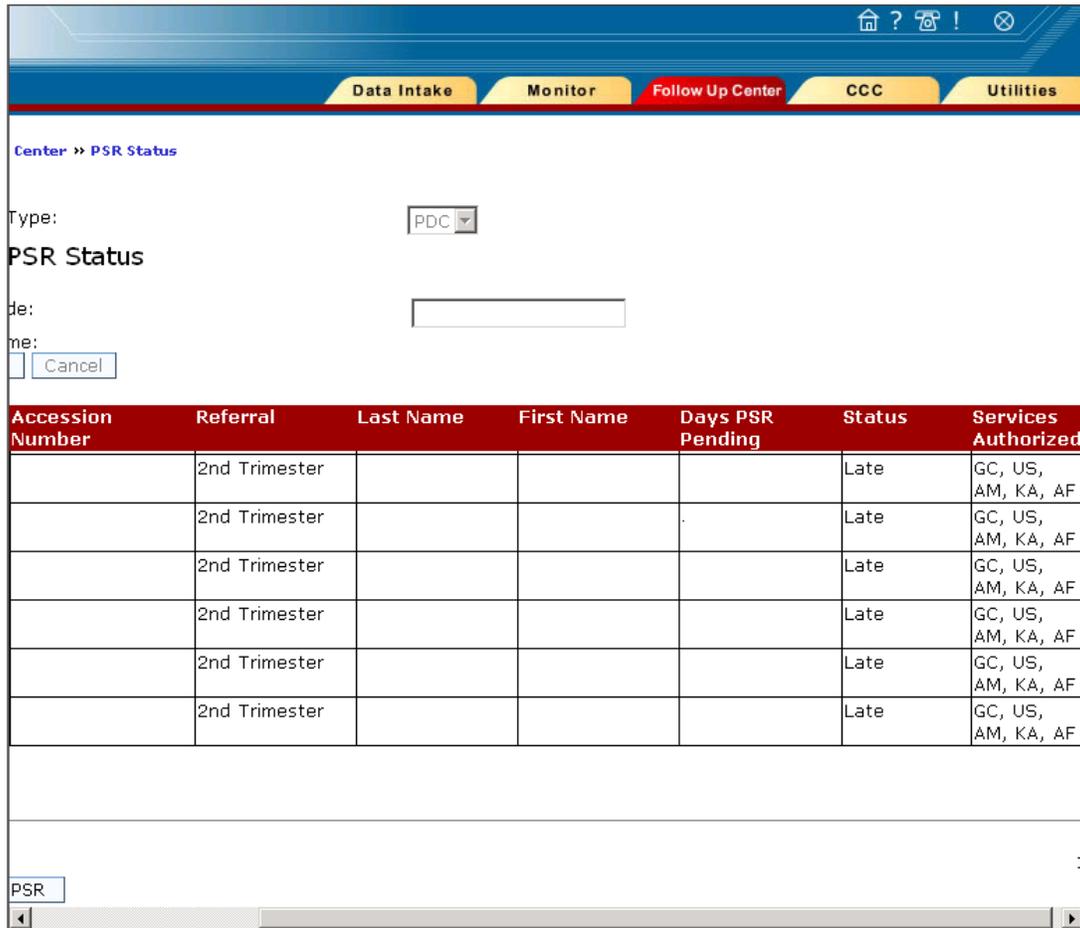


Figure 4.3: PSR Status Screen

PSRs are arranged according to the number of the days the PSR has been pending submission. Names of patients who were seen earlier are toward the top of the screen, and patients seen recently are toward the bottom of the screen. (e.g., 80 days PSR pending means it has been 80 days since the patient’s appointment status was kept in SIS, and a PSR has not been completed and submitted to GDSP.)

A scroll bar is located on the right side of the gridbar for PSRs pending. Use this scroll bar to see all pending PSRs for the PDC. If numbers appear in the bottom right corner of the gridbar, these numbers signify that additional pages of pending PSRs follow. Click these numbers and use the scroll bar to view the patient names.

The **PSR Status** screen also contains a column titled **Accession Number**. Clicking the words “Accession Number” will sort this column by ascending or descending accession numbers.

PSRs with incomplete information should stay on the **PSR Status** screen until all the information has been entered on the PSR. **Do not** click the **Submit** button for PSRs with pending information.

If PSRs are still on this screen, review these PSRs for completeness, individually, by clicking the **Save** button on each screen to make certain no “red text inconsistency messages” appear. Click the **Submit** button to send the PSR to GDSP and remove the PSR from this screen.

4.3. Data Entry Instructions for Online Patient Service Reports (PSRs)

The Patient Service Report (PSR) is used to perform data entry for follow-up diagnostic services provided to a Prenatal Screening patient. Since a patient may have a first trimester and a second trimester specimen during the same pregnancy, more than one accession number may be issued for each patient seen at the PDC. If more than one accession number is associated with the case, a data grid is displayed as an intermediate screen for the user to select the specific PSR to work on. The user will not be able to navigate to a PSR for an accession number that was not referred to that PDC.

The PSR can be viewed once the appointment information has been entered as “GC Appt Kept-Dx Not Scheduled” or “Kept.” The appointment status will also affect the PSR screen(s) that is displayed.

4.3.1 First Trimester Referrals

GC Appt Kept-Dx Not Scheduled: The “GC Appt Kept-Dx Not Scheduled” appointment status can be used only with patients who are first trimester referrals. It should be selected when genetic counseling only has been provided prior to 14 weeks 6 days gestational age. No other follow-up diagnostic services can be entered as “Provided” on the PSR when the “GC Appt Kept-Dx Not Scheduled” has been entered. The only PSR screens that will be displayed when this status has been entered are the **Genetic Counseling, Chorionic Villus Services, and Pregnancy Status** screens.

The “GC Appt Kept-Dx Not Scheduled” appointment status can be updated to “Scheduled” and followed by “Kept” to indicate when a patient returns to the PDC for additional services, either in the first or second trimester.

Kept: The “Kept” appointment status must be selected if a patient has genetic counseling and CVS on the same day. Selecting this status will allow the PDC to enter genetic counseling and CVS as “Provided.” In these situations, “GC Appt Kept-Dx Not Scheduled” should not be selected at all.

The “Kept” appointment status should also be selected if a patient has returned to the PDC for additional services after the appointment that had the appointment status of “GC Appt Kept-Dx Not Scheduled.” This situation may occur if the patient has returned on a different day for CVS or if the patient has returned in the second trimester of pregnancy for additional follow-up

services. Once “Kept” is selected, the CVS service status may be entered as “Provided” or the user can access to the PSR screens for services provided in the second trimester of pregnancy.

4.3.2 First Trimester Referrals Where the Result is Superseded by a Second Trimester Risk Assessment (Full Integrated Screening Risk Assessment)

When a second trimester specimen is received for a patient who has a first trimester *Screen Positive* result, and the specimen is used to generate a second trimester Risk Assessment, the first trimester result becomes superseded. Once the result is superseded, the first trimester referral is cancelled and only the services provided before receipt of the second trimester specimen are considered to be authorized.

The on-time entry of the “GC Appt Kept-Dx Not Scheduled” or “Kept” appointment statuses will allow PDCs to track these statuses appropriately.

4.3.3 Second Trimester Referrals (Quad, Quad +NT, Full Integrated, Serum Integrated, NTD and SLOS Only)

The “Kept” appointment status is the only status that can be selected once the patient has been seen at the PDC, regardless of the services provided.

Screens displayed due to appointment status are listed below.

- With appointment status of “GC Appt Kept–DX Not Scheduled” or “Kept” in the first trimester (prior to 14 weeks 6 days), the following PSR screens will be displayed:

Enter PSR-Genetic Counseling Screen

Enter PSR-Chorionic Villus Sampling Screen (including Karyotype by CVS)

Enter PSR-Pregnancy Status Screen

- With appointment status of “GC Appt Kept–DX Not Scheduled” that is updated to “Kept” after a gestational age of 15 weeks, the following PSR screens will be displayed (in addition to Genetic Counseling, Chorionic Villus Sampling, and Pregnancy Status screens, unless CVS Service Status has NOT been indicated as “Provided” or “Not Authorized”):

Enter PSR - Modified Genetic Counseling Screen

- This screen will be available only to first trimester referrals. Modified genetic counseling must be offered to the patient, but the service can be declined. There should be no changes to the Interpretation Factor(s) during modified genetic counseling, since

they should have been reviewed with the patient in the first genetic counseling session. If there are changes, contact GDSP.

Enter PSR - Modified Ultrasound Screen

- This screen will not display any Interpretation Factors for recalculation from the Ultrasound.

Enter PSR – Amniocentesis Screen (including AF-AFP results)

Enter PSR - Other/Karyotype Screen (for entering the karyotype results due to amniocentesis)

- With appointment status of “Kept” after gestational age of 15 weeks, the following PSR screens will be displayed:

Enter PSR - Genetic Counseling Screen

Enter PSR - Ultrasound Screen

Enter PSR - Amniocentesis Screen (including AF-AFP results)

Enter PSR - Other/Karyotype Screen (for entering the karyotype results due to amniocentesis)

Enter PSR - Pregnancy Status Screen

NOTE : Required fields on the PSR are denoted by an asterisk (*) on the left side of the label for the field name (e.g., *Last Name of Person Completing Form).

4.4. Information for Completing Various Enter PSR Screens

Specific information for completing each of the **Enter PSR** screens is provided below. Please contact GDSP if you need further clarification on how to enter any information in the Screening Information System (SIS).

4.4.1 Enter PSR (Main Screen)

Data Intake >> Case Summary (PNS) >> Enter PSR

Appointments Services History Enter PSR GA Calculator

Back Next

*Last Name of Person Completing Form:

*First Name of Person Completing Form:

*Date Form Completed: - -

Patient Information - 1st Trimester Referral

Genetic Disease Screening Program Accession Number: - - / - -

*Last Name:

First Name:

Maiden Name:

Birthdate: 08 - 27 - 1956

Social Security Number: - -

PDC Medical Record Number:

PDC Code - Center Name:

PNS Referral due to Screen Positive For

Down syndrome

Trisomy 18

Other

*Number of Fetuses in this Pregnancy (including fetal demises): Select

Authorization Details

*Were special exceptions and/or special services authorized by staff at the Genetic Disease Screening Program?: No

Last Name of person at the GDSP who authorized services:

Services Authorized by Prenatal Screening Program and Provided at your center.

Date	Service	Status	Provider

Review Results Modify Service

View or Add CVS Karyotype

View Subsequent Information Acquired and Pregnancy Status

Save Back Next Submit

Figure 4.4: Enter PSR (Main Screen)

The fields on the **Enter PSR (Main Screen)** are as follows:

- *Last name of person completing PSR
- *First name of person completing PSR
- *Date person is completing PSR
- Verify patient's last and first name—You can make the changes
- Verify patient's date of birth—Contact CCC with changes
- Verify patient's Social Security Number—You can make the change
- *Number of fetuses (including fetal demises)
- *Were special exceptions and/or special services authorized by staff at the Genetic Disease Screening Program? Currently, this field is defaulted to the answer "No." If "Yes" is answered, then the last name of the GDSP staff member who gave a Special Authorization becomes a required field.
- Click the **Save** button.
- Click the **Next** button.

Review all the services under "Services Authorized By Prenatal Screening Program and Provided at Your Center."

- Click the **Save** button.
- Review services in the gridbars.
 - Genetic counseling provided, then date of service and provider name, should be in gridbar.
 - Ultrasound provided, then date of service and provider name, should be in gridbar.
 - Amniocentesis provided, then date of service and provider name, should be in gridbar.
 - SLOS
 - **Not Indicated** if screening indication is not *SLOS Positive*.
 - **Provided** only if amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS was performed and if screening indication is *SLOS Positive*.
 - **Declined** is option to be selected when the screening indication is *SLOS Positive* and patient declines the diagnostic testing.
 - AF-AFP/AF-AChE Analysis if amniocentesis provided.
 - Analysis of Amniotic Fluid with Karyotype if amniocentesis provided.
- Click the **Submit** button when all services are correct and the inconsistencies are resolved.

NOTE : The **Submit** button will not be available unless Genetic Counseling, Ultrasound, Amniocentesis, and SLOS have a service status of either Provided, Declined, Not Indicated or Not Authorized.

NOTE: If the **Submit** button has been clicked on the **Enter PSR** main screen, services with the status of “Provided” will appear on the screen. If the **Submit** button has not been clicked, the red text message “No records found” will appear. See Figure 4.5.

Data Intake » Case Summary (PNS) » Services History

Appointments Services History Enter PSR GA Calculator

Client Name: _____ Date of Birth: _____

Accession Number: _____

Accession Number --/____-____

Patient Name _____

Services Provided

Line #	Date	Accession Number	Referral	Service Provided	Site Code	2nd Srvc Auth	Provider Last Name, First Name	Inv No
1	2/24/2009		1st Trimester	Chorionic Villus Sampling				
2	2/24/2009		1st Trimester	Karyotype by CVS				
3	2/24/2009		1st Trimester	Genetic Counseling				

Figure 4.5: Service History

4.4.2 Enter PSR – GC (Genetic Counseling) Screen

Figure 4.6: Enter PSR GC (Genetic Counseling) Screen

Genetic Counseling - 1st Trimester Referral

NOTE: Although Genetic Counseling Services status does not have an asterisk (*) next to it, SIS requires an answer in order to move to the next screen.

If the Genetic Counseling (GC) Services status is being entered as

- **Provided**
 - Click the Provided radio button.
 - Enter the date of service.
 - Select the genetic counseling provider from the drop-down list. (If the name is not present in the list, contact GDSP.)
 - Click the **Add to Grid** button.
- **Declined**
 - If you are entering Genetic Counseling Services as **Declined**, do not complete a PSR.

- Contact the CCC to inform them that the patient declined genetic counseling and, as a result, the patient has declined all follow-up services under the State's Prenatal Screening Program.
- **Not Indicated**
 - If you are entering Genetic Counseling Services as **Not Indicated**, do not complete a PSR.
 - However, if you are entering information on the PSR as the result of a Special Authorization for ultrasound, second-opinion ultrasound, amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS, then select **Not Indicated** for the Genetic Counseling Service.
- **Not Authorized**
 - If you are entering Genetic Counseling Services as **Not Authorized**, do not complete a PSR.
 - However, if the patient had genetic counseling that you are billing to her insurance, and a Special Authorization for another service such as ultrasound, second-opinion ultrasound, amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS was given by GDSP then:
 - Click the **Not Authorized** radio button.
 - Enter the date of service.
 - Select the genetic counseling provider from the drop-down list. (If the name is not in the list, contact GDSP.
 - Click the **Add to Grid** button.

Changes in Interpretation Factors During Genetic Counseling

If there were no changes to the Interpretation Factor(s) during genetic counseling, skip this section on the PSR. If Interpretation Factor(s) changed during genetic counseling, check which factor(s) changed. This information must be reported to the CCC.

- Number of Fetuses
- Weight
- Smoking
- Insulin-dependent diabetes
- Ovum Donor
- Ovum Donor Age
- Fetal Reduction
- Fetal Demise
- Maternal age
- Race/Ethnicity
- Blood Collection Date
- NT Exam Date
- NT Measurement

- CRL
- Chorionicity
- Gestational age (2nd T referrals only)

If a change to one or more Interpretation Factors occurred during genetic counseling and results in a change to the final (overall) interpretation, then select from the drop-down list either

- Negative
- Too Early
- Too Late
- Other *Screen Positive* (**DO NOT select this option if the case stays positive for the same indication, e.g., original indication was *Screen Positive* for Down syndrome and screening indication stays *Screen Positive* for Down syndrome with a different risk.**)
- Too Early High AFP (2nd T referrals only)
- Too Late High AFP (2nd T referrals only)
- If the screening result becomes *Pregnancy Not Screenable*, contact the CCC or GDSP.

4.4.3 Enter PSR – CVS (Chorionic Villus Sampling) Screen

Chorionic Villus Sampling - 1st Trimester Referral

Chorionicity: Monochorionic Dichorionic Unable to Determine

Fetus (Letter): A

Chorionic Villus Sampling Services: Provided Declined Not Indicated

Not Authorized Not Available Not Able to be Performed

Date CVS Provided: 07/02/2008

CVS Type: TransABDOMINAL(TA) TransCERVICAL(TC)

Name of Provider:

Service	Status	Date	Provider	Fetus (Letter)
<input type="checkbox"/> TransABDOMINAL CVS	Provided	07/02/2008		A

Results Determined From Services

CVS Karyotype Result

Fetus Letter: A

CVS Karyotype diagnosis: Not Performed Abnormal

Normal Male (Include normal variants) Normal Female (Include normal variants)

Normal (Gender not revealed) Culture Failed

Lab completing study:

Cytogenetic lab specimen number:

If CVS Karyotype abnormal, designate abnormality: Down syndrome Trisomy 18 Other Abnormal Karyotype

Mosaic

Abnormal CVS Karyotype result/cytogenetic diagnosis (ISCN short form):

Select:	Fetus	Karyotype Diagnosis	Abnormal Karyotype (ISCN)
<input type="checkbox"/>	A	Abnormal	47,XY,+21

Figure 4.7: Enter PSR – CVS (Chorionic Villus Sampling) Screen

Chorionic Villus Sampling - 1st Trimester Referral

NOTE: Although CVS service status does not have an asterisk (*) next to it, SIS requires an answer in order to move to the next screen.

For twin gestations, select Chorionicity as

- Monochorionic
- Dichorionic
- Unknown

For singleton cases (1 fetus) the fetus letter “A” checkbox will be pre-checked and grayed out.

If there is more than one fetus, you must enter CVS service for each fetus. If the twins are monochorionic, the fetus number will be defaulted to “A” and you must enter one CVS service.

If the CVS service status is being entered as

- **Provided**
 - Click the **Provided** radio button.
 - Enter the date of service.
 - Select the CVS types as either Transabdominal or Transcervical
 - Select the CVS provider from the drop-down list. (If the name is not in the drop-down list, contact GDSP.)
 - Click the **Add to Grid** button.
- **Declined**
 - Click the **Declined** radio button.
 - Click the **Add to Grid** button.
- **Not Indicated**
 - CVS service status will not be indicated if final (overall) screening indication is anything other than *Screen Positive*.
 - However, if you are entering a Special Authorization for CVS, then select **Not Indicated**.
- **Not Authorized**

Select this radio button when you know GDSP is not going to pay for this service, and it is billed to the patient’s insurance.
- **Not Available**

This radio button will be pre-selected and grayed out if a CVS provider is not available at this PDC, but the patient was referred to your PDC for genetic counseling in the first trimester.

- **Not Able to be Performed**

Select this radio button if a CVS was attempted but was not able to be performed.

- Click the **Add to Grid** button.

Results Determined from Services

If there is more than one fetus, you must enter karyotype result for each fetus. If the twins are monozygotic, you only need to enter one karyotype result; therefore, select and enter all results for fetus A.

- **Normal karyotype**

- Select one of the normal karyotype diagnoses.
 - Normal male (include normal variants)
 - Normal female (include normal variants)
 - Normal (gender not revealed)
- Select the cytogenetic lab that performed the karyotype (if the lab name is not in the drop-down list, contact GDSP.)
- Enter the cytogenetic lab specimen number.
- Skip if karyotype abnormal, designate abnormality.
- Skip abnormal karyotype result/cytogenetic diagnosis (ISCN short form).
- Click the **Add to Grid** button.
- Click the **Save** button.
- Click the **Next** button.

- **Abnormal karyotype**

- Select Abnormal in the karyotype diagnosis field.
- Select the cytogenetic lab that performed the karyotype. (If the lab name is not in the drop-down list, contact GDSP.)
- Enter the cytogenetic lab specimen number.
- Select one of the options under “If Karyotype abnormal, designate abnormality.”
 - Down syndrome
 - Trisomy 18
 - Other Chromosomal Abnormality
- Enter the abnormal karyotype result/cytogenetic diagnosis (ISCN short form). Use the commas, slashes, minus signs, plus signs, and spaces in the appropriate places. Double-check that you have typed in the correct nomenclature for the karyotype.
- Click the **Add to Grid** button.

- Click the **Save** button.
- Click the **Next** button.
- **No Karyotype result:** Select only if amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS was performed.
 - Select one of these options from the karyotype diagnosis field.
 - **Not Performed:** If this option is selected, enter an explanation on the next screen. (Enter PSR PregStatus) in the Comments box.
 - **Culture Failed:** If this option is selected, enter the cytogenetic lab and explanation, if applicable, on the next screen. (Enter PSR PregStatus) in the **Comments** box.
 - Click the **Add to Grid** button.
 - Click the **Save** button.
 - Click the **Next** button.

Modified Genetic Counseling - 1st Trimester Referral

If the Genetic Counseling (GC) service status is being entered as

- **Provided**
 - Click the **Provided** radio button.
 - Enter the date of service.
 - Select the genetic counseling provider from the drop-down list. (If the name is not in the list, contact GDSP.)

- **Declined**
 - Click the **Declined** radio button.
 - Click the **Add to Grid** button.

- **Not Indicated**

If you are entering information on the PSR as the result of a Special Authorization for ultrasound, second-opinion ultrasound, amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS, then select **Not Indicated** for the Modified Genetic Counseling Service.

- **Not Authorized**

If the patient had Modified Genetic Counseling that you are billing to her insurance, and a Special Authorization for another service such as ultrasound, second-opinion ultrasound, amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS was given by GDSP then

- Click the **Not Authorized** radio button.
- Enter the date of service.
- Select the genetic counseling provider from the drop-down list. (If the name is not in the list, contact GDSP.)

NOTE: There should be no changes to the Interpretation Factor(s) during Modified Genetic Counseling since they should have been reviewed with the patient in the first genetic counseling session. If there are changes, please contact GDSP.

4.4.5 Enter PSR – US (Ultrasonography) Screen

Ultrasonography - 1st Trimester Referral

Ultrasound Services: Provided Declined Not Indicated
 Not Authorized

Date Ultrasound Provided: []-[]-[] []

Name of Provider: [Select]

2nd opinion ultrasound: [No]

PDC code of referring PDC for 2nd opinion ultrasound: [Select]

Name of referring PDC: []

Service	Status	Date	Provider
<input type="radio"/> Ultrasound	Provided	05/26/2008	

Ultrasonography

Fetus (Letter): A

Bi-Parietal Diameter (BPD): [] mm

Ultrasound Result: [Select]

Fetal Abnormality by Anatomical Category: [Select]

Specific Fetal Abnormality: [Select]

Other Abnormality- Please Specify: []

Fetus (Letter)	BPD (mm)	Result	Abnormality

Figure 4.9: Enter PSR – US (Ultrasonography) Screen Modified for 1st Trimester

Changes in Interpretation Factor During Ultrasound

Interpretation factor which changed during ultrasound: Number of Fetuses Gestational Age
 If Interpretation changed during ultrasound, indicate final interpretation:

Results Determined From Services

Ultrasonography

Fetus (Letter): A

Bi-Parietal Diameter (BPD): mm

Ultrasound Result:

Fetal Abnormality by Anatomical Category:

Specific Fetal Abnormality:

Select Fetus (Letter)	BPD (mm)	Result	Abnormality
C	A	19	No Abnormality on Ultrasound for the Fetus/Pregnancy

3350::pdc001
11/20/2003
00:00:00.000

Figure 4.10: Enter PSR – US (Ultrasonography) “old” version (for 2nd Trimester) has a different bottom half

Ultrasonography - 1st Trimester Referral

NOTE: Although ultrasound service status does not have an asterisk (*) next to it, SIS requires an answer in order to move to the next screen.

If the ultrasound service status is

- **Provided**
 - Click the **Provided** radio button
 - Enter the date of service.
 - Select the Consultative Sonologist provider from the drop-down list. (If the name is not in the list, contact GDSP.)
 - Skip the second-opinion ultrasound question; otherwise, select “Yes” to second-opinion ultrasound if a Special Authorization was provided by GDSP.
 - Skip PDC code of referring PDC for second-opinion ultrasound unless a second-opinion ultrasound was provided.
 - Skip Name of Referring PDC
 - Click the **Add to Grid** button.

- **Declined**

- Click the **Declined** radio button.
- Skip the date of service.
- Skip the Consultative Sonologist provider from the drop-down list.
- Skip the second-opinion ultrasound.
- Skip PDC code of referring PDC for second-opinion ultrasound.
- Skip Name of Referring PDC.
- Click the **Add to Grid** button.

- **Not Indicated**

Ultrasound service status will not be indicated if final (overall screening) indication was *Negative, Too Early, Too Late, Pregnancy Not Screenable* or if you are entering information on the PSR as the result of a Special Authorization for genetic counseling, second-opinion ultrasound, amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS. In these instances, select **Not Indicated** for ultrasound service.

- Click the **Not Indicated** radio button.
- Skip the date of service.
- Skip the Consultative Sonologist provider from the drop-down list.
- Skip the second-opinion ultrasound.
- Skip PDC Code of Referring PDC for second-opinion ultrasound.
- Skip Name of Referring PDC.
- Click the **Add to Grid** button.

- **Not Authorized**

Select this radio button when you know GDSP is not going to pay for this service and it is billed to the patient's insurance.

- Click the **Not Authorized** radio button.
- Enter the date of service.
- Select the Consultative Sonologist provider from the drop-down list. (If the name is not in the list, contact GDSP.)
- Skip the second-opinion ultrasound.
- Skip PDC Code of Referring PDC for second-opinion ultrasound.
- Skip Name of Referring PDC.
- Click the **Add to Grid** button.

Results Determined from Services

Ultrasonography Findings: If there is more than one fetus, you must enter ultrasound results for each fetus.

- **Normal ultrasound**

- Select Fetus (Letter). If there is more than one fetus, you will have to select a letter (i.e., A, B, C, D, E) to designate the ultrasound findings for each fetus A–E.
 - Enter Biparietal Diameter (BPD) in mm: Enter two-digit number between 16 and 78.
 - Select **No abnormality** on ultrasound for the fetus/pregnancy under ultrasound result (**NOTE:** If there are monoamniotic/monochorionic twins, contact GDSP about how to enter **No Abnormality** on ultrasound for fetus/pregnancy)
 - Skip Fetal Abnormality by Anatomical Category
 - Skip Specific Fetal Abnormality
 - Click the **Add to Grid** button
 - Click the **Save** button
 - Click the **Next** button
- **Abnormal ultrasound**
 - Select Fetus letter. If there is more than one fetus, you will have to select a letter.
 - Enter Biparietal Diameter (BPD) in mm: Enter two-digit number between 16 and 78.

Do not enter a BPD for the following ultrasound findings:

- Anencephaly
- Fetal demise < 20 weeks
- Fetal demise > or = 20 weeks
- Acardiac twin
- Acrania
- Craniorachischisis
- Iniencephaly
- Molar pregnancy
- Pregnancy not detected

NOTE: The following inconsistency will appear at the top of the screen “Ultrasound provided or Not authorized, but no biparietal diameter (BPD) result for any fetus A-E.” Do not resolve it by entering a BPD. Submit the PSR to GDSP without resolving the inconsistency. See Ultrasound Abnormality PSR in Appendix A.

Select one of these options under ultrasound result:

- Molar Pregnancy
- Pregnancy Not Detected
- Fetal Demise < 20 weeks
- Fetal Demise > or = 20 weeks
- Conjoined Twins
- Twin-Twin Transfusion

- Acardiac Twin
- Monoamniotic/Monochorionic Twins
- Other Ultrasound Abnormality

Select Fetal Abnormality by Anatomical Category: If there is more than one abnormality, repeat the steps above for each abnormality

- Central Nervous System
- Face
- Neck
- Heart/Lung
- Abdomen
- Skeletal System
- Kidney/Urinary Bladder/Pelvis
- Size/Growth/Overall Appearance
- Amniotic Fluid Volume
- Umbilical Cord
- Placenta

Select Specific Fetal Abnormality under the Anatomical Category. If there is more than one abnormality, then repeat the steps above for each abnormality (You can enter up to four abnormal ultrasound findings. If an ultrasound finding cannot be located, contact GDSP)

- Click the **Add to Grid** button.
- Click the **Save** button.
- Click the **Next** button.

Changes in Interpretation Factors During Ultrasound

NOTE: If there were no changes to the Interpretation Factor(s) during the ultrasound, skip this section on the PSR.

If Interpretation Factor(s) changed during ultrasound, check the factor(s). This information must be reported to the CCC.

- **Number of fetuses**
If “unknown” is changed to “1,” this is considered a change; therefore, the number of fetuses checkbox should be checked.
- **Gestational age**

If a change to one or more Interpretation Factors occurred during ultrasound and this results in a change to the final (overall) interpretation, then select from the drop-down either:

- *Negative*
- *Too Early*
- *Too Late*

- *Other Screen Positive* (DO NOT select this drop-down if the case stays positive for the same indication, e.g., original indication *Screen Positive* for Down syndrome and stays *Screen Positive* for Down syndrome after the Interpretation Factor(s) but with a different risk number.
- *Too Early-High AFP*
- *Too Late-High AFP*

If the screening result becomes *Pregnancy Not Screenable*, contact CCC or GDSP.

4.4.6 Enter PSR - AM (Amniocentesis) Screen

Figure 4.11: Enter PSR – AM (Amniocentesis) Screen

Amniocentesis - 1st Trimester Referral

NOTE: Although amniocentesis service status does not have an asterisk (*) next to it, SIS requires an answer in order to move to the next screen.

If there is more than one fetus, you must enter amniocentesis service for each fetus. If the twins are monoamniotic/monochorionic, then select fetus A and enter one amniocentesis service.

If the Amniocentesis (AM) service status is being entered as

- **Provided**
 - Click the **Provided** radio button.
 - Enter the date of service.
 - Select the Amniocentesis Provider from the drop-down list (if the name is not in the list, contact GDSP).
 - Click the **Add to Grid** button.
- **Declined**
 - Click the **Declined** radio button.
 - Click the **Add to Grid** button.

- **Not Indicated**

Status of amniocentesis services will not be indicated if final (overall screening) indication was *Negative, Too Early, Too Late, or Pregnancy Not Screenable*. However, if you are entering a Special Authorization for genetic counseling, ultrasound, second-opinion ultrasound, fetal tissue karyotype, placental biopsy, or PUBS, then select **Not Indicated** for Amniocentesis Services.

- Click the **Not Indicated** radio button.
- Click the **Add to Grid** button.

- **Not Authorized**

Select this radio button when you know GDSP is not going to pay for this service and it is billed to the patient's insurance.

- Click the **Not Authorized** radio button.
- Enter the date of service.
- Select the Amniocentesis Provider from the drop-down list. If the name is not in the list, contact GDSP.
- Click the **Add to Grid** button.

Results Determined From Services

NOTE : If there is more than one fetus, you must enter AF-AFP and AF-AChE result for each fetus. If the twins are monoamniotic/monochorionic, you need to enter only one AF-AFP and AChE result; therefore, select and enter all results for fetus A.

Enter **Yes** or **No** to the field titled "Was there adequate Amniotic fluid collected?"

AF-AFP value (in MoM) available and less than 2.0 MoM, and AChE is negative, not performed, or uninterpretable. The entire number plus decimal of AF-AFP value must be entered (i.e., 0.70, 1.00, 1.95).

- Enter in AF-AFP value (in MoM).
- Skip “If AF-AFP MoM is not available.”
- Skip “If AF-AFP is over 2.0 MoM and/or positive AF-AChE, did patient receive another Ultrasound?”
- Select an option for AF-AChE
 - Negative
 - Not Performed
 - Uninterpretable
- Click the **Add to Grid** button.
- Click the **Save** button.
- Click the **Next** button.

AF-AFP value (in MoM) available and greater than 2.0 MoM, patient had another ultrasound and AChE is positive. The entire number plus decimal of AF-AFP value must be entered (e.g., 2.25, 6.00, 25.45).

- Enter in AF-AFP value (in MoM).
- Skip “If AF-AFP MoM is not available, give reason.”
- Select an option under the field titled “If AF-AFP is over 2.0 MoM and/or positive AF-AChE, did patient receive another Ultrasound?”
 - Yes,” then when you get to the Enter PSR-PregStatus screen, you must select “Later ultrasound” under the field titled “Information Acquired by” and must check as many boxes under the field titled “List Additional Diagnoses” that apply to the ultrasound findings discovered on the “Later ultrasound.”
 - “No,” then go to the next field titled AF-AChE and select Positive for AF-AChE.
- Click the **Add to Grid** button.
- Click the **Save** button.
- Click the **Next** button.

AF-AFP value (in MoM) available and greater than 2.0 MoM, patient had another ultrasound, and AChE is negative, not performed, or uninterpretable. The entire number plus decimal of AF-AFP value must be entered (e.g., 2.25, 6.00, 25.45).

- Enter AF-AFP value (in MoM).
- Skip “If AF-AFP MoM is not available, give reason.”
- Select an option under the field titled “If AF-AFP is over 2.0 MoM and/or positive AF-AChE, did patient receive another Ultrasound?”
 - “Yes,” then when you get to the Enter PSR-PregStatus screen, you must select “Later ultrasound” under the field titled “Information Acquired by” and must check as many boxes under the field titled “List Additional

Diagnoses” that apply to the ultrasound findings discovered on the “Later ultrasound.”

- “No,” then go to the next field titled AF-AChE and select an option in the drop-down list.
- Select an option for AF-AChE.
 - Negative
 - Not Performed
 - Uninterpretable
- Click then **Add to Grid** button.
- Click the **Save** button.
- Click the **Next** button.

No AF-AFP value, and AChE is negative, not performed, or uninterpretable:

- Skip AF-AFP value (in MoM).
- Select a reason why there is no AF-AFP value.
 - Uninterpretable
 - Not performed
- Skip “If AF-AFP is over 2.0 MoM and/or positive AF-AChE, did patient receive another Ultrasound?”
- Select an option for AF-AChE.
 - Negative
 - Not Performed
 - Uninterpretable
- Click the **Add to Grid** button.
- Click the **Save** button.
- Click the **Next** button.

No AF-AFP value and AChE is positive:

- Skip AF-AFP value (in MoM).
- Select a reason why there is no AF-AFP value.
 - Uninterpretable
 - Not performed
- Select an option under the field titled “If AF-AFP is over 2.0 MoM and/or positive AF-AChE, did patient receive another Ultrasound?”
 - “Yes,” then when you get to the Enter PSR-PregStatus screen, you must select “Later ultrasound” under the field titled “Information Acquired by” and must check as many boxes under the field titled “List Additional

Diagnoses” that apply to the ultrasound findings discovered on the “Later ultrasound.”

- “No,” then go to the next field titled AF-AChE.
- Select **Positive** for AF-AChE.
- Click the **Add to Grid** button.
 - Click the **Save** button.
 - Click the **Next** button.

4.4.7 Enter PSR - SLOS (Smith-Lemli-Opitz syndrome) Screen

Data Intake » Case Summary (PNS) » Enter PSR - SLOS

Update Case (PNS) Case Notes Link PNS Cases Re-assign CCC Tracking Events
 Appointments Services History Enter PSR Link Client to Entity Unlink Specimens
 GA Calculator

Back Next

Client Name: Date of Birth: 7/2/1987
 Accession Number:

Smith-Lemli-Opitz syndrome Services - 1st Trimester Referral

Smith-Lemli-Opitz syndrome Testing Provided Declined Not Indicated
 Not Authorized

Add to Grid Update Cancel

SLOS Services

Select:	Service	Status
<input type="radio"/>	SLOS Diagnostic Testing	Not Indicated

Edit Delete

Smith-Lemli-Opitz syndrome Results

Fetus Letter: A
 Smith-Lemli-Opitz syndrome Results:
 7-Dehydrocholesterol ng level
 Can 8-Dehydrocholesterol ng level be detected?
 8-Dehydrocholesterol ng level
 Laboratory where test performed

Add to Grid Update Cancel

Results

Select:	Fetus	SLOS Results	7-DHC ng level	8-DHC ng level
<input type="radio"/>	A	Fetus Not Affected		

Edit Delete

Save Back Next

3350:;pd=001
 11/20/2003
 00:00:00.000

Figure 4.12: Enter PSR - SLOS (Smith-Lemli-Opitz syndrome) Screen

Smith-Lemli-Opitz Syndrome Services - 1st Trimester Referral

NOTE: Although SLOS service status does not have an asterisk (*) next to it, SIS requires an answer in order to move to the next screen.

If the Smith-Lemli-Opitz syndrome (SLOS) service status is being entered as

- **Provided** (Should only be selected if patient is *Screen Positive* for SLOS, and patient wants SLOS diagnostic testing)
 - Click the **Provided** radio button.
 - Click the **Add to Grid** button.
- **Declined** (Should only be selected if patient is *Screen Positive* for SLOS, and patient does not want SLOS diagnostic testing and has declined amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS)
 - Click the **Declined** radio button.
 - Click the **Add to Grid** button.
- **Not Indicated** (Should be selected if patient is **not Screen Positive for SLOS**. However, if you are entering information on the PSR as the result of a Special Authorization for genetic counseling, ultrasound, second-opinion ultrasound, fetal tissue karyotype, placental biopsy, PUBS, then select **Not Indicated** for SLOS testing services.)
 - Click the **Not Indicated** radio button.
 - Click the **Add to Grid** button.
- **Not Authorized** (Select this radio button when you know GDSP is not going to pay for this service.)
 - Click the **Not Authorized** radio button.
 - Enter the date of service.
 - Click the **Add to Grid** button.

Results Determined From Services

NOTE: If the SLOS testing services status is “Provided” or “Not Authorized,” then SLOS results are required.

- Select one of the options under the Smith-Lemli-Opitz syndrome results drop-down list.
 - Fetus Affected
 - Fetus Not Affected
- Enter 7-DHC ng level.
- Skip field “Can 8-Dehydrocholesterol ng level be detected?”

- Skip field “8-Dehydrocholesterol ng level.”
- Select Kennedy-Krieger Institute.
- Click the **Add to Grid** button.
- Click the **Save** button.
- Click the **Next** button.

4.4.8 Enter PSR – Other/Karyotype Screen

Other Services - 1st Trimester Referral

Other services: Fetal Tissue Karyotype Placental Biopsy PUBS

Date Procedure Provided: 08-06-2008

Provider: Select

Add to Grid Update Cancel

Services

Select	Service	Date	Provider
C	Fetal Tissue Karyotype	08-06-2008	

Edit Delete

Results Determined From Services

Authorized Karyotype or Subsequent Karyotype

Fetus Letter: A

Karyotype diagnosis: Not Performed Abnormal
 Normal Male (Include normal variants) Normal Female (Include normal variants)
 Normal (Gender not revealed) Culture Failed

Name of lab completing study (or lab code number):

Cytogenetic lab specimen number:

If Karyotype abnormal, designate abnormality: Down syndrome Trisomy 18 Other Abnormal Karyotype
 Mosaic

Abnormal Karyotype result/cytogenetic diagnosis (ISCN short form):

Add to Grid Update Cancel

Results

Select	Fetus	Karyotype Diagnosis	Abnormal Karyotype (ISCN)
C	A	Normal Male (Include normal variants)	

Edit Delete

Save Back Next

Figure 4.13: Enter PSR – Other/Karyo (Karyotype) Screen

Other Services – 1st Trimester Referral

This screen is used to enter other types of diagnostic services (Fetal Tissue Karyotype, Placental Biopsy, or PUBS) and karyotype results. You should select only one of the choices below if a Special Authorization was authorized by GDSP prior to the procedure being performed.

- Fetal Tissue Karyotype—Authorized by GDSP via Special Authorization
 - Click the **Fetal Tissue Karyotype** radio button.
 - Enter the date of service.
 - Click the **Add to Grid** button.
- Placental Biopsy or PUBS—Authorized by GDSP via Special Authorization.
 - Click the **Placental Biopsy** or **PUBS** radio button.
 - Enter the date of service.
 - Select the CVS provider from the drop-down list. If the name is not in the list, contact GDSP.
 - Click the **Add to Grid** button.

If amniocentesis was declined, click the **Next** button to move to the Enter PSR - Pregnancy Status screen.

If amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS was performed, use the right scroll bar to navigate to the bottom of the screen to enter the karyotype results.

Authorized Karyotype or Subsequent Karyotype

NOTE: If there is more than one fetus, you must enter karyotype result for each fetus. If the twins are monoamniotic/monochorionic, you only need to enter one karyotype result; therefore, select and enter all results for fetus A.

- **Normal karyotype**
 - Select one of the normal karyotype diagnoses
 - Normal male (include normal variants)
 - Normal female (include normal variants)
 - Normal (gender not revealed)
 - Select the cytogenetic lab that performed the karyotype. If the name lab is not in the drop-down list, contact GDSP.
 - Enter the cytogenetic lab specimen number.
 - Skip “If Karyotype abnormal, designate abnormality.”
 - Skip “Abnormal Karyotype result/cytogenetic diagnosis (ISCN short form)”
 - Click the **Add to Grid** button.
 - Click the **Save** button.
 - Click the **Next** button.

- **Abnormal karyotype**
 - Select **Abnormal** in the karyotype diagnosis field.
 - Select the cytogenetic lab that performed the karyotype. If the name lab is not in the drop-down list, contact GDSP.
 - Enter the cytogenetic lab specimen number.
 - Select one of the options under “If Karyotype abnormal, designate abnormality.”
 - Down syndrome
 - Trisomy 18
 - Other Chromosomal Abnormality
 - Enter the Abnormal Karyotype result/cytogenetic diagnosis (ISCN short form). Use the commas, slashes, minus signs, plus signs, spaces in the appropriate places; double check that you have typed in the correct nomenclature for the karyotype.
 - Click the **Add to Grid** button.
 - Click the **Save** button.
 - Click the **Next** button.
- **No Karyotype result** (Only select if amniocentesis, fetal tissue karyotype, placental biopsy, or PUBS was performed.) Select one of these options from the karyotype diagnosis field”
 - **Not Performed:** If this option is selected, enter an explanation on the next screen (Enter PSR– PregStatus) in the **Comments** box.
 - **Culture Failed:** If this option is selected, enter the following:
 - Enter the cytogenetic lab.
 - Enter an explanation, if applicable, on the next screen (Enter PSR - PregStatus) in the **Comments** box.
 - Click the **Add to Grid** button.
 - Click the **Save** button.
 - Click the **Next** button.

4.4.9 Enter PSR – PregStatus (Pregnancy Status) Screen

Data Intake > Case Summary (PNS) > Enter PSR - PregStatus

Client/Case Search
View Client Profile
Case Summary (PNS)

Appointments Services History Enter PSR GA Calculator

Back Next

Client Name: _____ Date of Birth: 8/27/1956
Accession Number: _____

Pregnancy Status- 1st Trimester Referral

Fetus letter: A
Pregnancy Status:
Update if pregnancy status has changed

Termination date: - -

Subsequent Information Acquired After Initial PDC Visit

Information acquired by:
Specify other source:
List additional diagnoses:

- NO ABNORMALITY
- NO ABNORMALITY ON ULTRASOUND
- FETAL DEMISE < 20 WEEKS
- FETAL DEMISE > OR = 20 WEEKS
- ANENCEPHALY
- SPINA BIFIDA
- ABDOMINAL WALL DEFECT
- OLIGOHYDRAMNIOS
- PLACENTAL ABNORMALITIES
- SPINA BIFIDA WITH HYDROCEPHALY
- ENCEPHALOCELE
- GASTROSCHISIS
- OMPHALOCELE

Other NTD (specify):
Other abnormalities (specify):
Later abnormal Karyotype result/cytogenetic diagnosis:

Add to Grid Update Cancel

Select Fetus	Pregnancy Status	Information Acquired By	Additional Diagnosis

Edit Delete

NOTE: If there were several methods (later ultrasound, later karyotype, pathology/autopsy/visual report, or other method) used to obtain a diagnosis for the fetus after the initial PDC visit, indicate the one method providing the definitive diagnosis. If a later karyotype was the method, please enter the karyotype above.

Comments:

Save Back Next

Figure 4.14: Enter PSR – PregStatus (Pregnancy Status) Screen

Pregnancy Status – 1st Trimester Referral

Use this screen to enter pregnancy status, additional diagnostic information, if applicable, and comments.

- Status of the Pregnancy:
 - Select one of the options; however, if there is a fetal demise > or = 20 weeks or fetal demise <20 weeks, skip the status of the pregnancy
 - Continuing pregnancy
 - Patient intends to have fetal reduction
 - Patient Undecided/Lost to follow-up or unknown
 - Fetal loss after PDC visit (Fetal Demise/SAB/Missed abortion)
 - Patient intends to have an elective termination
- Known elective termination after PDC visit: Enter a Termination date in the next field
- Skip termination date unless pregnancy status is “Known elective termination after PDC visit.”

Subsequent Information Acquired after Initial PDC Visit

- Skip “Information acquired by” unless AF-AFP is over 2.0 MoM and/or AChE is positive, or patient had later ultrasound or later karyotype or fetal autopsy/pathology/visual report or other methodology.
- Skip “Specify other source” unless “other” was selected under the “Information acquired by” field.
- Skip “List additional diagnoses” unless you selected “later ultrasound” or “later karyotype” or “autopsy/pathology/visual report” or “other” under “Information acquired by” field. **NOTE:** Multiple boxes can be checked at one time, depending on the diagnostic findings.

Please note the section on the PSR giving you instructions about completing the PSR if a later karyotype is available. Contact GDSP if you need further clarification on how to enter this information in the Screening Information System (SIS).

- Click the **Add to Grid** button.
- Skip “Comments” unless you have additional information about the patient’s visit, pregnancy, or fetal findings. The “Comments” field has 200 spaces available.
- Click the **Save** button.
- Click the **Next** button, which will take you to the **Enter PSR (Main Screen)**, the first screen shown in this chapter.

4.5. Helpful Hints for PSR Data Entry

If you click the **Add to Grid** button repeatedly, you will receive the following error message: “Duplicate Services entered. Please enter only one service status per PSR” or “Duplicate services for the same fetus letter entered. Please enter only one service status per fetus letter per PSR” or “Duplicate rows for a fetus is not permitted.” To resolve these error messages, you should click either the **Next** or the **Back** button to get rid of the error message. When you navigate back to the screen you navigated from, you will notice that the red text error message is gone and the data you entered is in the grid.

Table 4.2 describes the function of buttons and links on the **Enter PSR (Main Screen)**.

Table 4.2: Functions of Buttons and Links on the Enter PSR (Main Screen)

Button or Link Name	Description of Function	How to Use It
Modify Service button	<p>Allows you to navigate to PSR screens quickly, with the least amount of mouse clicking, and to modify the service status (Provided, Declined, Not Indicated, or Not Authorized), date of service, or provider name.</p> <p>Contact GDSP if you need specific instructions to modify CVS or amniocentesis results.</p>	<ul style="list-style-type: none"> • Click the radio button for the service (genetic counseling, ultrasound, amniocentesis, SLOS, pregnancy status, and list of additional diagnoses) in the Services Authorized gridbar. • Click the Modify Service button. • Change the service status, date of service, or provider name. • Click the Update button. • Click the Save button.
Review Results button	<p>Allows you to navigate to another screen quickly and to modify the results such as Interpretation Factor(s) changes, ultrasound results, AF-AFP/AChE results, and karyotype results.</p>	<ul style="list-style-type: none"> • Click the radio button for the service (genetic counseling, ultrasound, amniocentesis, SLOS, pregnancy status, and list of additional diagnoses). • Click the Review Results button. • Click the radio button for the row on the gridbar you want to modify. • Click the Edit button. • Click the Update button. • Click the Save button.

Button or Link Name	Description of Function	How to Use It
View or Add Other Service link	Allows you to navigate to the Enter PSR-Other/Karyo screen to enter or modify a karyotype, fetal tissue karyotype, placental biopsy, or PUBS service in one click of the mouse.	<ul style="list-style-type: none"> • Click the radio button for the Other service gridbar or the karyotype gridbar. • Click the Edit button. • Change the information. • Click the Update button. • Click the Save button.
View Subsequent Information Acquired and Pregnancy Status link	Allows you to navigate to the Enter PSR-PregStatus screen in one click of the mouse to modify the pregnancy status gridbar or to enter the pregnancy status and/or subsequent information.	<ul style="list-style-type: none"> • Click the radio button for the pregnancy status gridbar. • Click the Edit button. • Change the information for the pregnancy status or the subsequent information/additional diagnoses. • Click the Update button. • Click the Save button.
Edit button	Allows you to change/modify data for a gridbar that was already created.	<ul style="list-style-type: none"> • Click the radio button for the gridbar you want to modify. • Click the Edit button. • Change the data. • Click the Update button. • Click the Save button.
Update button	Allows you to save the data that you modified for a gridbar that was already created.	<ul style="list-style-type: none"> • Click the radio button for the gridbar you want to modify. • Click the Edit button. • Change the data. • Click the Update button. • Click the Save button.

Chapter 5: Invoicing/Billing Instructions

This chapter provides examples of screens in the Screening Information System (SIS) related to invoicing and billing for Prenatal Screening services. Instructions for viewing information on the screens are included with each screen.

5.1. PSR Status Screen

Figure 7.1 displays the **PSR Status** screen, which lists Patient Service Reports (PSRs) that are pending submission.

1. Click the **Follow Up Center** tab.

2. Click the **PSR Status** link.

Clicking the words "Accession Number" will sort the column by ascending or descending accession numbers.

Clicking the radio button and the **Enter PSR** button will take you to the on-line submission of the Patient Service Report (PSR).

Select	Accession Number	Last Name	First Name	Days PSR Pending	Status
<input type="radio"/>	026-32-013/P -2008-11			21	PSR Needed
<input type="radio"/>	032-62-068/P -2008-12			15	PSR Needed
<input type="radio"/>	033-09-011/P -2008-12			13	PSR Needed
<input type="radio"/>	040-43-047/P -2008-11			7	PSR Needed

Figure 5.1: PSR Screen

5.2. Services Rendered Screen

Figure 5.2 provides instructions for generating **Services Rendered** screens (aka Patient List screens) and viewing services rendered based on data entry on PSR, GDSP payment rules, and Special Authorizations.

The screenshot shows the 'Services Rendered' screen in the GDB-SIS system. The interface includes a navigation menu on the left with links for 'Cases Referred', 'PSR Status', 'Services Rendered', 'Invoice Status', 'Quart Rept- Specimens', 'Quart Rept- Patients', and 'View Special Auth'. The 'Services Rendered' link is highlighted. The main content area has a breadcrumb trail 'Follow Up Center >> Services Rendered' and a search form with fields for '*PDC Code:', '*PDC Name:', 'Month:', and 'Invoice Number:'. There are also 'Go' and 'Clear' buttons. A search bar with a 'Search' button is at the top right. Three callout boxes provide instructions: 1. Click the 'Follow Up Center' tab. 2. Click the 'Services Rendered' link. 3. Select a 'Month' from the drop-down menu. A large box contains steps 4 through 8, detailing the selection of time periods and the format for the invoice number.

1. Click the **Follow Up Center** tab.

2. Click the **Services Rendered** link (Known as 2nd level link).

(NOTE: This screen shot was taken after the link was clicked; therefore, in Step 1, you will see the **Cases Referred** screen under the **Follow Up Center** module tab).

3. Select a **Month** from the drop-down menu.

If you know the invoice number, skip to Step 8. Otherwise, continue to Step 4.

4. Select one of two periods of time from **Days** (either 1–15 or 16–31).

5. Enter a **Year** (YYYY).

6. Click the **Go** button on this screen or the **Enter** key on your keyboard.

7. Enter the Invoice Number (2-digit PDC code, followed by a dash, followed by the fiscal year, followed by a dash, followed by the invoice number, xx-yyyy-xx).

8. Click the **Go** button on this screen or the **Enter** key on your keyboard.

Figure 5.2: Services Rendered Screen

5.3. Services Rendered Screen Navigation

The **Services Rendered** screen allows you to view a summary of all services that have been provided by a given PDC and submitted to the Genetic Disease Screening Program for payment during a given time frame. Once a search has been performed, the results are displayed in a grid, by satellite code, as illustrated in Figure 5.3.

Follow-up 087 - View PDC Service Rendered Summary - Microsoft Internet Explorer

GDB-SIS

Data Intake Entity Monitor Follow Up Center CCC Utilities

Follow Up Center » Services Rendered

*PDC Code: Search

*PDC Name:

Month: February Days: 1-15 Year: 2008

Invoice Number:

Go Clear

Satellite Code	Accession Number	Last Name	First Name	Date of Service	GC	US	Amnio	Karyo	AF-AFP	FTK	PUBS	PB/CVS SLOS	2nd Op US	Admin Fee
a	010-23-404/P - 2008-32			01/17/2008	1	1	1	1	1	0	0	0	0	1
a	018-19-147/P - 2008-32			01/25/2008	1	1	1	0	1	0	0	0	1	1
a	019-72-179/P - 2008-31			02/01/2008	1	1	1	1	1	0	0	0	0	1
a	035-64-051/P - 2008-31			02/12/2008	1	1	0	0	0	0	0	0	0	1
a	024-34-192/P - 2008-31			02/01/2008	1	1	0	0	0	0	0	0	0	1
			Total		5	5	3	2	3	0	0	0	1	5

1

Click the **Accession Number** link to view the **Case Summary (PNS)** screen for the selected case. You can click the **Enter PSR** link on the **Case Summary** screen to verify that services were entered on the PSR. You can click the **Service History** link on the **Case Summary** screen to cross-check the invoice number per service provided.

Figure 5.3: Services Rendered Search Results

5.4. Invoice Status

The **Invoice Status** screen displays the payment status for a particular invoice. It also provides the Schedule Number, Schedule Date, Warrant Number, and Warrant Date. See Figure 5.4.

1. Click the **Follow Up Center** tab.

2. Click the **Invoice Status** link.

3. Select **From** and **To** dates to search invoice status payment ranges.

4. Click the **Go** button on this screen or the **Enter** key on your keyboard.

Invoice Number	Schedule Number	Schedule Date	Warrant Number	Warrant Date
01-2006-17	286506	03/30/2007	6693966	05/11/2007
01-2006-16	286477	03/20/2007	6709879	05/17/2007
01-2006-15	286477	03/20/2007	6709879	05/17/2007
01-2006-13	286399	02/20/2007	6493499	03/16/2007
01-2006-Q2	286376	02/07/2007	6428334	02/26/2007
01-2006-12	286361	01/24/2007	6425418	02/23/2007
01-2006-11	286348	01/18/2007	6428343	02/26/2007

Figure 5.4: Invoice Status Screen

5.5. Invoice Payments

Follow the steps below to verify, in SIS, if a check was issued by Sacramento for a particular invoice. These steps are illustrated in Figure 5.5.

1. Click the **Follow-up Center** tab.
2. Click the **Invoice Status** link (in the blue section on the left side of the screen).
3. Verify that your Comprehensive PDC code appears in the PDC code field.
4. Type in the beginning date of the search in the **Date From** field or select the date from the Icon calendar next to the **Date From** field. This beginning date can be the SIS start-up date (July 11, 2005).

5. Type in the end date of the search in the **Date To** field or select the date from the Icon calendar next to the **Date To** field.
6. Click the **Go** button or click the **Enter** key on the keyboard.

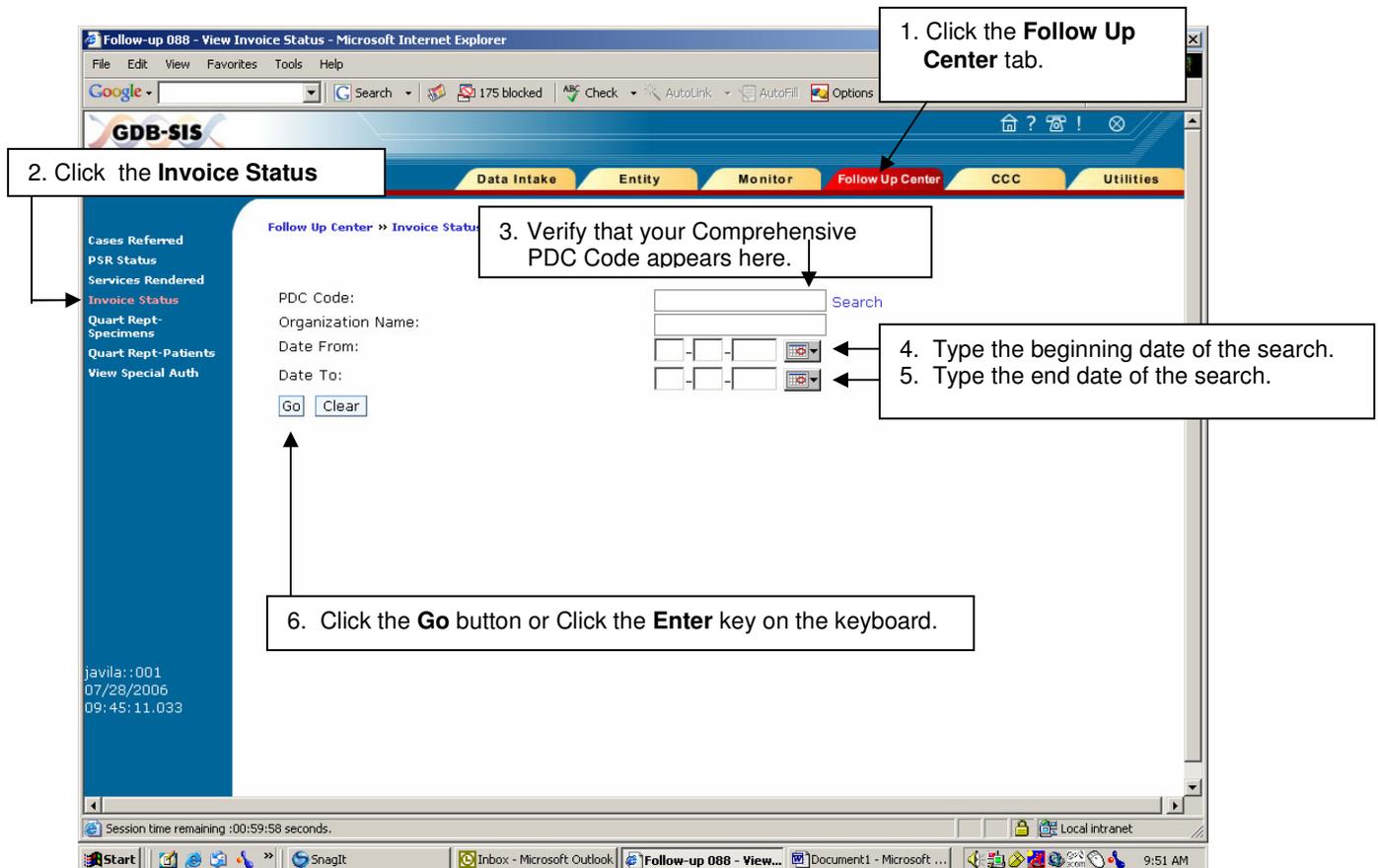


Figure 5.5: Invoice Status Screen

Notice the columns displayed after the “Go” button is clicked.

- Invoice Number—Number linking the user to the **Service Rendered** screen, which lists patients who were provided service(s) and whose PSRs were completed, submitted, and resolved of all inconsistencies during the invoice period. Invoice numbers prior to July 11, 2005, could appear in this column; however, no patients will be listed on the screen and the red text message, “No Records Found,” will appear. Quarterly bonus(es) numbers are also listed here and designated by PDC code, fiscal year, and calendar quarter. No patients are listed for the quarterly bonus.
- Schedule Number—Number for the Sacramento document that includes the invoice.
- Schedule Date—Calendar time the schedule number was sent to Sacramento.
- Warrant Number—Number of the check Sacramento issued to pay the invoice.
- Warrant Date—Calendar time the check was sent by Sacramento to the PDC.

Chapter 6: Chart Documentation and Reporting Requirements

6.1. Updating Appointment Status in SIS

The PDC must update the appointment status in SIS within three (3) business days of any change in the patient's appointment status. An alert is generated through SIS to the PDC Appointment Scheduler if the appointment has not been updated within three (3) business days. Appointment statuses are as follows:

- Scheduled
- GC Appointment Kept-Dx Not Scheduled (This status is to be used for first trimester referrals in which the patient declined a CVS and is either awaiting a second trimester diagnostic appointment or a second trimester revised risk assessment)
- Kept (This status can only be used in the first trimester if the patient had a CVS; otherwise, this status is used for first trimester referrals seen in the second trimester or for second trimester referrals)
- Error-Appointment Not Kept
- Cancelled
- No Show

6.2. PDC/CCC Communication

If the patient cannot be seen within seven (7) days of the referral and chooses to be seen at another PDC, the PDC must notify the CCC to cancel the referral. (See Appendix A for a current list of CCCs by region.) If the patient wishes to switch PDC referral assignments within the same PDC, a Special Authorization is required in advance from GDSP. (See Appendix A for a current list of GDSP staff to contact.)

For patients who are seen for a *First Trimester Combined Screen Positive* result, the PDC must communicate to the CCC the patient's decision regarding services within one (1) business day of the patient's appointment. This communication may be via telephone or fax. (See PDC Fax to CCC re: 1st T Positive in Appendix A.) The communication must disclose if the patient

- Has had a diagnostic procedure (CVS) in the first trimester;
- Has opted to have additional services in the second trimester;
- Has opted to have a second trimester blood specimen drawn for Full Integrated Screening;
- Is undecided; or
- Has decided to decline any further services.

Any change to an Interpretation Factor identified during genetic counseling or ultrasound must be called into the CCC assigned to the case. The Interpretation Factor changed must be saved to the case, and a new interpretation must be communicated to the patient before she leaves the

PDC or before proceeding with further authorized services. The new interpretation can be obtained through the **Case Summary** screen in SIS and printed for the patient’s chart, or it can be obtained via fax from the CCC. The new interpretation should include the initials of the Genetic Counselor who discussed the modified results with the patient.

6.3. Alerts Generated Through SIS

If the prenatal care provider calls the CCC and asks for a reinterpretation based on new information, and the interpretation or risk assessment changes, SIS notifies the PDC scheduler via an alert. This alert states, “Please note that one or more Interpretation Factors have changed for this case since first referred. Please notify the Genetic Counselor immediately.”

If the patient is no longer eligible for follow-up services, an alert will be generated that says, “This patient is no longer authorized for follow-up services and the referral has been cancelled. Please notify the Genetic Counselor.” The case will move to the Cancelled Referral grid on the **Cases Referred** screen. Please see Table 6.1 and Table 6.2 for other PDC Alerts on the **Monitor Module-View Alerts** screen:

Table 6.1 Appointment Scheduler Alerts

Alert Code	Alert Statement
1	We have taken this case off your inventory. Please notify the Genetic Counselor.
7	Appointment has not been scheduled. Please follow-up with coordinator. (Note: This alert is sent every 5 days until the patient’s appointment is scheduled in SIS.)
10	Please note that one or more Interpretation Factors have changed for this case since first referred. Please notify the Genetic Counselor.
37	Appointment date and time has passed. Please update appointment status.
39	Site visit is scheduled in 30 days at a PDC. Please notify the PDC Director.
79	Case approaching 24 weeks gestational age.
81	This patient has just been referred to your PDC.
84	A site visit has been scheduled. You may review the list of cases that will be examined for this audit. Please give the list to the PDC Director.
130	Please note that the first trimester result is now superseded. Inform the Genetic Counselor.

Table 6.2 PSR Contact Alerts

Alert Code	Alert Statement
2	Service authorized. Please notify the Genetic Counselor.
3	Service denied. Please notify the Genetic Counselor.

NOTE : Individuals with multiple roles in SIS (e.g., PDC appointment scheduler and PSR contact) will see alert codes for all their respective roles.

6.3.1 How to Filter Alerts Based on an Alert Code

- Log into the SIS application at <http://sis.dhs.ca.gov/>
- Navigate to the Monitor Module tab and the View Alerts screen will appear.
- Select **Alert Code** from the “Filter by” dropdown list, and a textbox will appear.
- Enter an alert code such as “81” if you are a PDC appointment scheduler.
- Click the **Search** button or press the **Enter** key on your keyboard.
- Notice only alerts with that alert code will appear in the grid on the **View Alerts** screen.
- Click once on the “Date” column header to sort alerts by the oldest date first.
- Click a second time on the “Date,” and the sort will have the most recent alerts first.
- “Group” Cancellation of Alerts.

Follow the directions above for filtering alerts based on alert code.

- Click the **Check All** button to place a check in the box under the “Cancel Alert” column header in the grid for all the alerts.
- Remove the check in the box for the alerts you do not want to cancel under the “Cancel Alert” column header in the grid.
- Click the **Save** button and a message “Are you sure you want to cancel the checked alerts?” will appear.
- Click “Yes” to this message to reconfirm a “group” cancellation should occur. If there are no alerts remaining after the group cancellation, the message “No records found” will appear.
- Click “No” to this message to stop the “group” cancellation. No changes will occur.

6.4. Protocol for Prenatal Genetic Screening Questionnaire/Pedigree

GDSP’s Continuous Quality Improvement (CQI) Committee recommends (but does not require) that PDCs/satellites use both a questionnaire and a pedigree as tools for obtaining a patient’s pregnancy and family history. A pedigree is required if the patient, partner, or fetus is at increased risk for a genetic disorder due to a significant family history indicated on the

questionnaire or by referral. See Appendix A for examples of prenatal questionnaire/pedigree tools.

NOTE: Use of the GDSP Prenatal Genetic Screening Questionnaire is optional. However, if this form is not utilized at the PDC/satellite, one of the following must occur:

- The PDC/satellite may use its own prenatal questionnaire form, which must contain all the required elements listed on the GDSP questionnaire. If specific elements are not contained on the questionnaire, they must be addressed and documented elsewhere in the patient's chart.
- The PDC/satellite may use the GDSP Prenatal Genetic Screening Pedigree.
- The PDC/satellite may use its own pedigree form. The patient's chart must contain documentation that all of the required elements listed on the GDSP form have been addressed and documented elsewhere in the patient's chart.

NOTE: The GDSP Prenatal Genetic Screening Questionnaire or one of the above listed alternative forms must be completed for all patients referred to the PDC/satellite

- Who are referred for follow-up through the Prenatal Screening Program.
- When the indication for referral is one of those listed on the PDC Quarterly Report Form, regardless of gestational age.

The questionnaire can be completed in one of two ways:

- By the patient (or another person on behalf of the patient, e.g., translator) in advance of talking with the Genetic Counselor. The patient should sign and date the completed form.
- During the genetic counseling session, with the Genetic Counselor asking the questions and documenting the patient's responses on the form.

The Genetic Counselor must review the questionnaire with the patient. If the patient did not provide a response to a particular question, the Genetic Counselor should clarify the question and solicit a response. The counselor should inquire if any questions were not understood or if the patient gave any responses she was not sure of. The Genetic Counselor must sign and date the completed form.

6.5. Clinical Geneticist Chart Review

In accordance with the PDC Standards, a Clinical Geneticist must supervise all professional services and be responsible for the evaluation of work performance by reviewing and signing off on all patient charts. The Clinical Geneticist must sign the chart within 30 days of the patient's final date of service. The signature indicates a review of the family history, ultrasound findings, amniocentesis, or CVS consent form or documentation of decline to ensure that appropriate services meeting State Standards have been provided. The chart must also be reviewed for the amniocentesis or CVS results and documentation that the results and all significant clinical information have been sent to the referring physician. If the amniocentesis or CVS results are normal, this last review may be performed by a Board Certified Genetic Counselor. If the results

are abnormal, this review must be performed by a Clinical Geneticist, and the chart must also document that appropriate consultation was offered to the patient. A Clinical Geneticist may be responsible for supervision of services to no more than 200 prenatal patients per week.

All copies of reports and correspondence in the chart should be **signed and dated**. All patients seen for Prenatal Screening follow-up must have charts that include the following (if applicable):

- Prenatal Screening Interpretation Factors with documentation of confirmation with the patient.
- A genetic risk assessment via a questionnaire and/or pedigree. (See Appendix A.)
- Genetic counseling summary with risks and information reviewed with patient.
- The ultrasound report should document the BPD on all Prenatal Screening referrals and should include the signature of the Consultative Sonologist performing the ultrasound exam.
- Amniocentesis or CVS consent/refusal forms signed by the patient. (See Appendix A.)
- Cytogenetic and AFAFP/AChE results.
- Copies of correspondence to referring clinicians.
- Telephone consultations, including communication of genetic screening or diagnostic test results.
- Consent for release of Prenatal Screening results, if necessary.
- Documentation of offer of Clinical Geneticist involvement in abnormal cases.
- Documentation of explanation for delay in follow-up services if Prenatal Screening patient is not seen within seven days of the initial contact.
- Written documentation of communication with the CCC (for any reinterpretation of Prenatal Screening results).
- Signed and dated Clinical Geneticist review of the chart. (A Chart Review Form documenting all required elements to be reviewed in the patient's chart can be found in Appendix A.)

6.6. Electronic Medical Records (EMR)

EMR can be a paperless system, a “scanned document system,” or a combination. From GDSP’s perspective, information from the genetic counseling appointment must be accessible for GDSP monitoring. A document titled Electronic Medical Record in Appendix A reviews the data fields/information GDSP will need to access for review on a site visit.

The type of system the PDC can use will vary. The system needs to be HIPAA compliant and should be developed keeping EPHI (Electronic Patient Health Information) secure and confidential.

As the system is developed, the following issues should be addressed:

- Access to the EMR.
- Transmission of patient information via e-mail.
- Storage of patient information.
- Discard of patient information.

- Discard of computers, laptops, etc. containing patient information.

The following websites on HIPAA may be useful as PDCs develop their EMR system:

<http://www.cms.hhs.gov/SecurityStandard/>

<http://www.cms.hhs.gov/SecurityStandard/Downloads/SecurityGuidanceforRemoteUseFinal122806.pdf>

<http://www.cchit.org/files/CCHITPhysiciansGuide2007.pdf> (physicians' guide to certification of ambulatory medical records)

<http://www.cchit.org/choose/ambulatory/2007/index.asp> (products certified by certification commission for Healthcare Information Technology)

<http://documents.iss.net/marketsolutions/RegulatoryComplianceDataSheet.pdf>

6.7. Informed Consent for Services Not Covered by the Prenatal Screening Program

The California Expanded AFP Screening Program is a voluntary program involving a blood test and, depending upon the results, follow-up diagnostic services. Some of these services are authorized by the Prenatal Screening Program and involve no additional costs to the patient. These no-cost services include the following :

- Genetic risk assessment by a state-approved Genetic Counselor.
- Ultrasound (if indicated).
- Amniocentesis (if indicated) and testing of the amniotic fluid for:
 - Chromosome problems like Down syndrome and Trisomy 18
 - Neural tube defects like spina bifida (open spine)
 - Smith-Lemli-Opitz syndrome, a rare birth defect causing mental retardation (only if indicated).

Although other testing may be indicated based upon the family and medical history or ultrasound study, these additional tests are not covered by the Program. These other services can be billed to the patient's health insurance or to the patient directly. The patient should check her policy to determine coverage of any additional services.

Examples of services NOT covered by the Prenatal Screening Program include the following:

- Rhogam injection following amniocentesis, if the blood type is Rh negative.
- Follow-up ultrasound to monitor the growth of the fetus.
- Special ultrasound of the baby's heart (fetal echocardiography).
- Genetic blood tests on the patient or her partner.
- Consultation with other specialists.
- Other testing of the amniotic fluid, such as FISH (a rapid chromosome test) or DNA analysis for viral exposures or genetic conditions.

An Informed Consent for Services Not Covered Form to give to patients is located in Appendix A. The form indicates that the patient understands what follow-up services are covered by the Prenatal Screening Program and that they will be financially responsible for any additional services.

6.7.1 Internal CQI Program

In January 2001, the Genetic Disease Branch (now the Genetic Disease Screening Program) required that all State-approved Prenatal Diagnosis Centers implement an internal Continuous Quality Improvement (CQI) program. The purpose of this program is twofold: First, to have each PDC establish its own evaluation of the patient care their center is providing. This evaluation is to be accomplished by an internal comprehensive review of the charts and of the center's overall performance from initial patient contact to the final provision of services. Secondly, if any deficiencies are noted in the documentation below a 95% threshold, the PDC should develop a method of corrective action to ensure that the documentation is included in the chart in a timely manner. GDSP will not be collecting any of the information generated by these internal CQI programs. The programs will be reviewed during PDC site visits to ensure that the internal CQI program is in process. Therefore, it is important for each PDC to be responsible for its own follow-up on any issues that may be found through its internal CQI program.

To assist in this process, GDSP and the CQI Committee have developed tools that can be adapted or modified as needed. These tools are available in Appendix A and include

- A sample PDC Monitors Checklist (to be used with each chart).
- A sample Chart Monitoring Tally Sheet.
- A sample Corrective Action Report.

6.8. Non-AFP Quarterly Reports

All state-approved Prenatal Diagnosis Centers (PDC) are required to submit summaries of their non-AFP PDC services and report the abnormal chromosomes and neural tube defects detected in their patients. This information is entered in the Screening Information System (SIS) by the quarterly report contact. A copy of the Confidential Case Report of a Birth Defect (CCR) to report the abnormal chromosomes and neural tube defects can be found in Appendix A.

A complete Non-AFP PDC Quarterly Report consists of the "Summary of Prenatal Diagnostic Services" submitted on-line in SIS and the case reports (CCRs), with a specimen-sampling date within the specific reporting period.

The Non-AFP PDC reports are due at the end of each quarter. Please note the following reporting schedule:

**Chapter 6:
Chart Documentation and Reporting Requirements**

Non-AFP PDC Quarterly Reporting Schedules	Reporting Period	Due Date
1 st	July–September	November 15 th
2 nd	October–December	February 15th
3 rd	January–March	May 15th
4 th	April–June	August 15 th

If you have any questions or comments, please contact Corinna Tempelis by telephone at (510) 412-1528 or e-mail Corinna.Tempelis@cdph.ca.gov

Appendix A: References

Reference	Chapter
1. PDC Guide to GDSP Personnel	1 & 6
2. List of Case Coordinators	1 & 6
3. Flowchart	1
4. State Approved Prenatal Diagnostic Centers	1
5. PDC Standards 2009	3
6. Entity Protocol Table	3
7. Prenatal Genetic Screening Questionnaire/Pedigree (English and Spanish)	3 & 6
8. Consent Forms (Amniocentesis and CVS in English, Spanish, Chinese, Vietnamese)	3
9. Ultrasound Abnormality PSR	4
10. PDC Fax to CCC re: 1 st T Positive	6
11. Chart Review Form	6
12. Electronic Medical Record	6
13. Informed Consent for Services Not Covered Form	6
14. PDC Monitors Checklist	6
15. Chart Monitoring Tally Sheet	6
16. Corrective Action Report	6
17. Confidential Case Report of a Birth Defect	6
18. Adverse Outcome Study	6

California Department of Public Health
Genetic Disease Screening Program
Expanded AFP Screening Program

850 Marina Bay Parkway, Richmond, CA 94804 • Phone 510/412-1502 • Fax 510/412-1547

PDC GUIDE TO GDSP PERSONNEL
Communication with Prenatal Screening Program

Area of Concern

PDC Policy Issues
PDC applications, personnel changes
Special authorizations
PSRs
PDC Invoicing and reimbursement
Prenatal Screening Program Policy Issues
Coordinator issues
Program Statistics
Prenatal Screening Educational Materials
PDC quarterly reports
NTD reporting system
Cytogenetic reporting system
Adverse Outcome Study

Primary Staff Person & Backup

Sara
Sara, Cheryl, Jackie, Steve
Sara, Cheryl, Jackie
Sara, Cheryl, Jackie
Lisa, Gerry
Monica
Eileen
Bob
Jeanne
Corinna, Marie
Marie
Linda M., Marie
Steve

GDSP STAFF PERSON/TELEPHONE NUMBER: (Area Code 510 and prefix 412)

Jackie Avila	6205
Jeanne Boxley	6214
Gerry Cayanan	231-7466
Bob Currier	1443
Monica Flessel	1456
Sara Goldman	1463
Lisa Holcomb	1479
Cheryl Ikeda	1475
Linda Malm	1495
Steve Purser	1491
Marie Roberson	1522
Eileen Stoner	1527
Corinna Tempelis	1528

Mailing address for all GDSP Staff:
Genetic Disease Screening Program
850 Marina Bay Parkway, Room F175
Richmond, CA 94804

Email Address: firstname.lastname
@cdph.ca.gov

GDB general line: (510) 412-1502

Prenatal Screening Program supplies:
phone – (510) 412-1441
fax – (510) 412-1553
Toll free number (866) 718-7915

SIS Help Desk (510) 307-8928

FAX for PDC-related issues: (510) 412-1551

**CALIFORNIA PRENATAL SCREENING PROGRAM
COORDINATOR REGIONAL ASSIGNMENTS
As of January 2009**

CCC #40

Safie Yaghoubi 510-412-1518 Erica Gordon 510-231-7464
Eileen Stoner 510-412-1527 Monica Flessel 510-412-1456

All cases that cannot be assigned to Coordinator sites 41–54 (due to lack of a ZIP code) are assigned to CCC 40. They are then re-assigned to the appropriate Coordinator site by CCC 40 staff.

CCC #41

LAURA LORBEER
KATHERINE ENSIGN

Backups: Laura Eppelmann
 Siri Stokesberry

Assistant: Michael Strabola
 Heidi Maas

U.C. DAVIS
(800) 559-5616 toll free
(916) 734-6551
FAX # (916) 734-0637

Assigned Counties:

Del Norte	Napa
Humboldt	San Francisco
Lake	Solano
Marin	Sonoma
Mendocino	

San Mateo ZIP Codes:

94005	94018	94066
94010	94019	94080
94011	94030	94401
94013	94037	94402
94014	94038	94403
94015	94044	94404

CCC #42

PRATIMA SWARUP
EMILY MALOUF
ANNA SUTTON

Backup: Laura Eppelmann
 Siri Stokesberry

Assistant: Michael Strabola
 Heidi Maas

U.C. DAVIS
(800) 391-8669 toll free
(916) 734-6575
FAX # (916) 734-0625

Assigned Counties:

Alpine	Sacramento
Amador	Shasta
Butte	Sierra
Calaveras	Siskiyou
Colusa	Stanislaus
El Dorado	Sutter
Glenn	Tehama
Lassen	Trinity
Modoc	Tuolumne
Nevada	Yolo
Placer	Yuba
Plumas	

ALL ZIP CODES FOR THE STATE OF NEVADA

CCC #43

TERI THOMPSON
BRET HUTCHINSON

Backups: Laura Eppelmann
 Siri Stokesberry

Assistant: Michael Strabola
 Heidi Maas

U.C. DAVIS
(800) 428-4279 toll free
(916) 734-6228
FAX # (916) 734-0637

Assigned Counties:

Alameda
Contra Costa
San Joaquin

**CALIFORNIA PRENATAL SCREENING PROGRAM
COORDINATOR REGIONAL ASSIGNMENTS
As of January 2009**

CCC #44	CCC #45	
BEATKA SHEETZ PAT MILLER	KAREN ELDREDGE JAMY HATHCOAT KIM PASQUINI MELANIE SEVERSON JESSICA GATES	Laura Eppelmann (916) 734-6553
Backups: Laura Eppelmann Siri Stokesberry	Assistant: Debbie Jacobs	Linda Healy (323) 625-3720
Assistant: Michael Strabala Heidi Maas		
U.C. DAVIS (877) 871-6467 toll free (916) 734-6078 FAX # (916) 734-0625	CHILDREN'S HOSPITAL CENTRAL CALIFORNIA (800) 237-7466 toll free (559) 353-6645 FAX # (559) 353-7215	
<u>Assigned Counties:</u> Monterey San Benito San Luis Obispo Santa Cruz Santa Clara	<u>Assigned Counties:</u> Fresno Kern Kings Madera Merced Mariposa Tulare	
San Mateo ZIP Codes: 94002 94028 94065 94020 94060 94070 94021 94061 94074 94025 94062 94303 94026 94063 94027 94064		

**CALIFORNIA PRENATAL SCREENING PROGRAM
COORDINATOR REGIONAL ASSIGNMENTS
As of January 2009**

CCC #47

TAMARA PURNELL
CYNTHIA KENNEDY
SHARIE CHENG

Backup: Linda Healy

Assistant: Raven Kelly

CEDARS-SINAI MEDICAL CENTER
(877) 568-9237 toll free
(323) 866-6790
FAX # (323) 866-6791

Assigned Counties:

Santa Barbara
Ventura

Los Angeles ZIP Codes:

90024	90401	91320	91360	93510
90034	90402	91321	91361	93532
90035	90403	91322	91362	93534
90043	90404	91324	91364	93535
90045	90405	91325	91367	93536
90049	90406	91326	91384	93537
90066		91328	91387	93538
90073	91040	91331	91390	93539
90077	91042	91332		93543
90095		91335	91401	93544
	91102	91340	91402	93550
90213	91126	91341	91403	93551
90214		91342	91404	93552
90215	91301	91343	91405	93553
90245	91302	91344	91406	93563
90265	91303	91345	91411	93591
90272	91304	91350	91412	
90290	91305	91351	91423	
90291	91306	91354	91436	
90292	91307	91355		
90293	91310	91356		
	91311	91358		
90301	91316			
90302				
90303				
90304				
90305				
90306				
90308				

CCC #48

DEBORAH GLASSER
GISELLE PEREZ DE LA GARZA
CHAI SIBOLIBAN

Backup: Linda Healy

Assistant: Yolanda Hayes

CEDARS-SINAI MEDICAL CENTER
(888) 330-9237 toll free
(323) 866-6750
FAX # (323) 866-6755

Assigned County:

Los Angeles ZIP Codes:

90004	90057	91101	91501
90005	90065	91103	91502
90006	90068	91104	91503
90010	90069	91105	91504
90012	90071	91106	91505
90014	90072	91107	91506
90015	90089	91108	
90016		91109	91601
90017	90210	91123	91602
90018	90211	91125	91604
90019	90212	91201	91605
90020		91202	91606
90026	91001	91203	91607
90027	91006	91204	91608
90028	91007	91205	91609
90029	91010	91206	
90030	91011	91207	91706
90031	91016	91208	91747
90036	91020	91209	91769
90038	91024	91214	91775
90039	91030	91225	91776
90041	91046	91226	91778
90042			91780
90046		91352	91793
90048			

**CALIFORNIA PRENATAL SCREENING PROGRAM
COORDINATOR REGIONAL ASSIGNMENTS
As of January 2009**

CCC #49

TERESA ARELLANO
KELLY CHARBONEAU

Backup: Linda Healy

Assistant: Marlene Sermenio

CEDARS-SINAI MEDICAL CENTER
(888) 844-9237 toll free
(323) 866-6788
FAX (323) 866-6789

Assigned County:

Los Angeles ZIP Codes:

90001	90230	91750
90002	90232	91754
90003	90234	91755
90007	90255	91765
90008	90270	91766
90011		91767
90013	90640	91768
90021		91770
90022	91702	91773
90023	91711	91789
90025	91722	91790
90032	91723	91791
90033	91724	91792
90037	91731	
90040	91732	91801
90044	91733	91803
90047	91734	
90056	91740	
90058	91741	
90062	91744	
90063	91745	
90064	91746	
90067	91748	

CCC #50

MARGY HOWELL
LUISA CORDOVA
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Assistants: Alberta Winslow
Charlette Johnson (float)

CEDARS-SINAI MEDICAL CENTER
(877) 567-9237 toll free
(323) 866-6795
FAX (323) 866-6796

Assigned County:

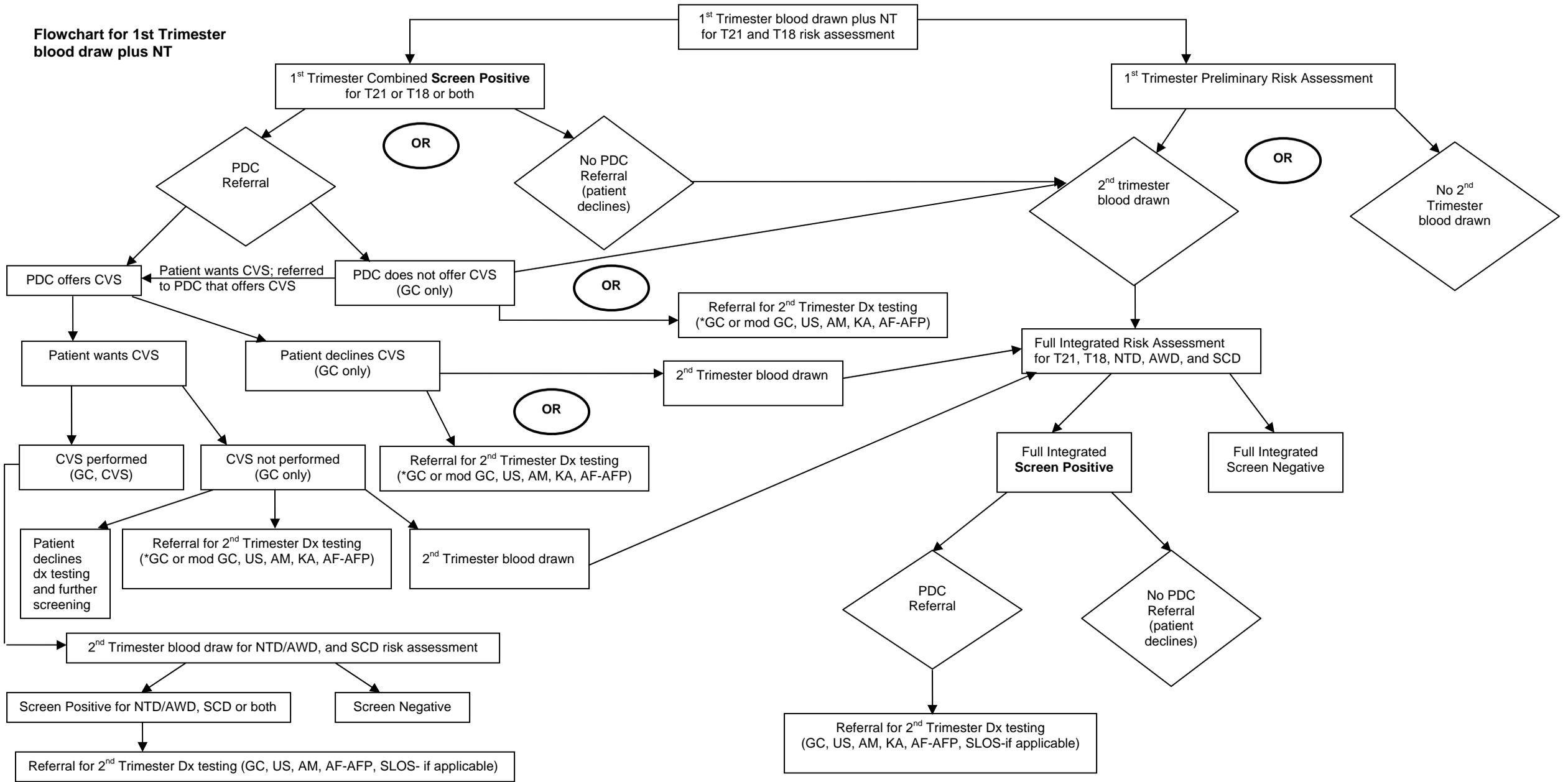
Los Angeles ZIP Codes:

90009	90260	90601	90701	90801
90051	90261	90602	90703	90802
90054	90262	90603	90704	90803
90059	90266	90604	90706	90804
90061	90267	90605	90710	90805
	90274	90606	90711	90806
90201	90275	90613	90712	90807
90206	90277	90637	90713	90808
90220	90278	90638	90714	90809
90221	90280	90639	90715	90810
90222	90294	90650	90716	90813
90223	90295	90651	90717	90814
90224	90296	90660	90723	90815
90231		90670	90731	90822
90240	90501		90732	90840
90241	90502		90733	
90242	90503		90744	
90247	90504		90745	
90248	90505		90746	
90249	90506		90747	
90250	90507		90749	
90254	90508			
90256	90509			
	90510			

**CALIFORNIA PRENATAL SCREENING PROGRAM
COORDINATOR REGIONAL ASSIGNMENTS
As of January 2009**

<p>CCC #51</p> <p>PAULETTE VODENICHAR THEA WILLS-OLSEN ARLYN IGLESIAS SUNNYE SALWAY JENNY GILLEY</p> <p>Assistants: Susan Koskela Judy Castillon</p> <p>U.C. IRVINE (877) 224-4373 toll free (714) 456-5994 FAX: (877) 757-5437</p> <p><u>Assigned Counties:</u> Inyo Mono Orange San Bernardino</p>	<p>CCC #52</p> <p>JENNIFER WRIGHT LISA MANAHAN LINDA TALLEY KRISTYN CAETANO (vacancy)</p> <p>Assistant: Jackie Stauch</p> <p>U.C. SAN DIEGO (866) 366-4408 toll free (858) 822-1280 FAX (858) 822-1284</p> <p><u>Assigned Counties:</u> Imperial Riverside San Diego</p>
<p>CCC #53</p> <p>PEGGY FEHLEN-QUIZON ROBYN KRIEGER MONICA WOHLFERD ELISE OBOLENSKY CAROL NOREM</p> <p>Assistant: Rachel Lee Ana Flores</p> <p>KAISER – NORTHERN CALIFORNIA</p> <p>(510) 752-6190 FAX: (510) 752-6800</p> <p>Serves clinicians and patients within the Kaiser Permanente health care system in Northern California.</p>	<p>CCC #54</p> <p>CALEEN DANON AKI LOGG AMY LIOU</p> <p>Assistants: John Criscione Rachiel Reyes</p> <p>KAISER - SOUTHERN CALIFORNIA</p> <p>(626) 564-3322 FAX: (626) 564-3311</p> <p>Serves clinicians and patients within the Kaiser Permanente health care system in Southern California.</p>

Flowchart for 1st Trimester blood draw plus NT



NOTE: If a patient has had a 1st Trimester Screen Positive result and once she has had a valid 2nd Trimester risk assessment, no further follow-up is authorized under the Prenatal Screening Program. In this case, the patient has only had genetic counseling and has not had a CVS, or US, and/or AM and the 2nd Trimester risk assessment is screen negative, then US and/or AM are not authorized. The 1st Trimester screen positive risk assessment has been superceded by the 2nd Trimester risk assessment. If the 2nd Trimester risk assessment is screen positive, then follow-up is authorized. However, the 2nd trimester screen positive risk assessment is considered a new referral and requires the CCC to re-assign the patient to the same PDC or assign the patient to a different PDC.

NOTE: * Genetic Counseling versus Modified Genetic Counseling depends on if the referral is at the same PDC.

STATE APPROVED PRENATAL DIAGNOSIS CENTERS

CVS	SITE#	SITE NAME	CVS	SITE#	SITE NAME
*	03a	Prenatal Diagnosis Northern Calif.	*	36a	Genzyme Pasadena
*	03b	Stockton	*	36h	U.S.C. - California
*	03f	Perinatal Associates of Sacramento		36j	Tarzana
*	04a	Cedars-Sinai Medical Center		36l	Ventura
*	05a	Prenatal Diagnostic & Perinatal Ctr		36p	Whittier
*	05b	Apple Valley	*	36r	LA-Grand Avenue
*	05c	Indio		36t	White Memorial
*	05d	Riverside	*	36ab	Walnut Creek
*	05e	Corona	*	36ai	Obstetrix Med Grp of Central Coast-SLO
*	05f	Wildomar	*	36aj	Santa Maria Palisades
	05g	West Covina	*	36bc	Burbank
	05h	Pasadena	*	36be	Hoag Memorial Hospital
*	05i	Montclair		36bi	Desert Hospital
	05o	Moreno Valley	*	36bm	Hollywood Presbyterian
	05p	Arrowhead	*	36bn	Providence Holy Cross
*	05q	San Gabriel Valley PMG Pomona		36bo	Devore-Lancaster
*	05r	San Gabriel Valley PMG West Covina		36bp	Garden Grove Hospital Perinatal Center
*	05s	San Gabriel Valley PMG Garfield	*	38a	U.C. Irvine
	05t	San Gabriel Valley PMG Arcadia		38c	St. Jude Medical Center
	05u	San Gabriel Valley PMG Montebello	*	39ae	Genzyme Genetics - Platt and Assoc
*	06a	U.C. San Diego	*	39b	Saddleback Memorial Med Ctr
	06c	El Centro		39g	Fountain Valley
*	08a	Sacramento Maternal Fetal Medicine		39i	Harbor Center For Genetics, Torrance
*	08b	Chico	*	39s	Modesto
	09a	Peninsula Prenatal Diagnostics		39z	Valley Presbyterian/Van Nuys
	09b	Palo Alto		39aa	San Diego Pregnancy Specialists
	09c	San Jose		39ab	Ogundipe-Lynwood
	09e	Fremont		39ac	Ogundipe-Inglewood
	09f	San Mateo		39ad	Samadi-Lancaster
*	14a	Genetics Center	*	39af	San Dimas Medical Group
	14g	Kern Prenatal Diagnostic Center		39ag	Valencia Perinatal Services
	14i	Perinatal Dx Ctr - Ventura		39aj	Children's Hospital of Central California
	14j	Perinatal Dx Ctr - Thousand Oaks		39ak	Perinatal Associates of Central California
	14k	Perinatal Dx Ctr - Santa Monica	*	39al	Obstetrix-San Jose
	15a	U.C. Davis	*	39am	Magella-Long Beach
	15b	Mercy-Redding	*	39an	Magella-Torrance
*	16a	U.C. San Francisco	*	39ar	Encinitas Perinatal Center
*	16f	Santa Rosa Clinic		39as	C. Scott Naylor, M.D.
*	16h	Monterey	*	39au	San Diego Perinatal Center
*	16j	Pleasanton	*	39av	Temecula Valley Perinatal Center
*	17a	East Bay Perinatal		39aw	Tarzana, Dr. Doany
*	17f	San Ramon		39ax	Ctr Maternal Fetal Medicine Santa Monica
*	17g	Fremont		39ay	Mission Hospital Regional Medical Ctr
*	18a	Stanford University	*	56a	San Gabriel Valley PDC
*	18b	Stanford at El Camino	*	56c	Fetal Diagnostic Center of Southern Calif.
*	18d	Santa Cruz		60a	Linda S. Cowan, M.D.
*	18e	Fremont		67a	Santa Clara Valley Medical Center
*	18f	Salinas		67b	O'Connor Hospital
*	24a	California Pacific Medical Center		70a	Central California Prenatal Diagnostics
*	24c	Marin	*	75c	Kaiser - North/Oakland
*	24e	Santa Rosa	*	75a	San Jose
*	24g	Mills Peninsula-San Mateo	*	75b	Hayward
*	28a	U.C.L.A. Medical Center		75d	Sacramento
*	28b	Northridge Hospital	*	75e	San Francisco
	28c	Olive View Medical Center	*	75f	San Rafael
*	28d	Santa Monica Hospital	*	75h	Santa Clara - Homestead
*	28k	Tabsh-UCLA		76a	Kaiser - South/Panorama City
*	29a	LAC+USC PDC	*	76b	Bellflower
	30g	Genesis Labs/Loma Linda		76c	Fontana
	30e	Hemet Valley Med Ctr		76d	South Bay
			*	76e	Los Angeles
				76f	Orange County
			*	76g	Riverside
				76h	West Los Angeles
				76i	Woodland Hills
				76j	Baldwin Park
			*	76z	San Diego
			*	80a	San Francisco Perinatal Associates
				92a	Clinical Genetic Ctr-La Mirada
				92b	Clinical Genetic Ctr-Fountain Valley

26 PDCs and 104 satellites
pdc&sat2CVS:03/30/2009

California Department of Public Health Genetic Disease Screening Program

850 Marina Bay Parkway, F175, Richmond, CA 94804 • Phone 510/412-1502 • Fax 510/412-1551

PRENATAL DIAGNOSIS CENTER STANDARDS AND DEFINITIONS

2009

I. Comprehensive Prenatal Diagnosis Center

A Comprehensive Prenatal Diagnosis Center is an established center with a complete range of prenatal diagnosis services and full-time multidisciplinary medical staff as follows:

- A. Has a Director who is certified in Clinical Genetics by the American Board of Medical Genetics. The Director is responsible for such administrative duties as:
 - 1. The supervision and the quality of testing, counseling, and medical care provided by all clinical members of the Prenatal Diagnosis Center staff, including Satellites;
 - 2. Assurance that participation in prenatal diagnosis procedures by any pregnant woman is voluntary;
 - 3. Notifying the GDSP within ten working days of:
 - a. Any changes in locations where services are provided or in staffing of locations, including a plan to meet the Standards on an interim and permanent basis; and
 - b. Any case of maternal mortality that could possibly be related to or associated with prenatal diagnosis;
 - 4. The acceptance for prenatal diagnosis of all pregnant women referred from state funded or administered programs; and
 - 5. The timely submission of required reports including reports on each patient with abnormal results, quarterly prenatal diagnosis center reports, and all practitioners' Adverse Outcome Studies.
 - 6. The acceptance of site visits by state staff or designated state agents and to make available confidential patient reporting systems, billing and medical records needed to monitor compliance with the PDC Standards and California Prenatal Screening Program Guidelines.

- B. The Director or other Clinical Geneticist(s) certified in Clinical Genetics by the American Board of Medical Genetics must:
1. Be available to provide consultation in person within three working days to all families with abnormal or questionable results. The consultation offer must be documented in the patient's chart; and
 2. Supervise all professional services and be responsible for the evaluation of work performance by reviewing and signing off on all patient charts. The Clinical Geneticist must sign the chart within 30 days of the final date of service. The signature indicates a review of the family history, ultrasound findings, amniocentesis or CVS consent form or documentation of decline to ensure appropriate services meeting State Standards have been provided. The chart must also be reviewed for the amniocentesis or CVS results and documentation that the results and all significant clinical information have been sent to the referring physician. If the amniocentesis or CVS results are normal, this last review may be performed by a Board certified Genetic Counselor. If the results are abnormal, this review must be performed by a Clinical Geneticist and there must also be documentation of appropriate consultation offered to the patient. A Clinical Geneticist may be responsible for supervision of services to no more than 200 prenatal patients per week.

In Sections C, K, and L, the number of procedures excludes repeat insertions.

- C. Has ultrasound-guided amniocentesis available at greater than/equal to 15 weeks gestation (standard amniocentesis). This is performed by physicians who:
1. Have previously completed 100 second trimester procedures under the supervision of a physician experienced in ultrasound guided amniocentesis. Each of the physicians performing or supervising the procedures must have specific knowledge and experience with:
 - a. The collection by second trimester amniocentesis of 50 amniotic fluid samples per year for cell culture and analysis. Once the practitioner has successfully completed two Adverse Outcome Studies for amniocentesis, the practitioner must collect at least 25 amniotic fluid samples annually;
 - b. Obstetrical ultrasonography; and
 - c. Basic genetic information and appropriate counseling procedures for chromosomal, biochemical and neural tube defects.

2. Provisional approval to perform amniocentesis may be given to Fellows in Training. Each Fellow in Training must:
 - a. Currently be in a Maternal-Fetal Medicine Training Program; and
 - b. Have completed the first 50 second trimester amniocentesis procedures performed under the direct supervision of an approved amniocentesis practitioner with the supervising practitioner in the room; and
 - c. Have the second 50 second trimester procedures performed with the supervising practitioner onsite, within immediate access to the patient; and
 - d. Submit a log of the second 50 supervised procedures to the Genetic Disease Screening Program for review. The practitioner will be granted full approval and then they must begin collecting Adverse Outcome data on consecutive procedures.
3. Provisional approval to perform amniocentesis may be given to Interim Approval Practitioners. Each Interim Approval Practitioner must:
 - a. Have completed a Maternal-Fetal Medicine Training Program; and
 - b. Have completed the first 50 second trimester amniocentesis procedures during their Fellowship training, performed under the direct supervision of an approved amniocentesis practitioner with the supervising practitioner in the room; and
 - c. Complete the deficient number of second trimester procedures under supervision within a defined time period; with the supervising practitioner onsite, within immediate access to the patient; and
 - d. Submit a log of the second 50 supervised procedures to the Genetic Disease Screening Program for review. The practitioner will be granted full approval and then they must begin collecting Adverse Outcome data on consecutive procedures.
4. If amniocentesis is offered between 13 and 15 weeks gestation (early amniocentesis), the practitioner must have been approved to perform standard amniocentesis and must have performed 100 supervised standard amniocentesis procedures including or in addition to 10 supervised early amniocentesis procedures.
5. If the amniocentesis is performed by a physician other than an Obstetrician/Gynecologist (OB/GYN), an OB/GYN with American

Board of Obstetrics and Gynecology certification/Active Candidate status or equivalent must be available on an emergency on-call basis in case of amniocentesis complications. The Genetic Disease Screening Program is to be notified whenever such an OB/GYN specialist is called on an emergency basis.

- D. Has consultative ultrasonography available which is performed by a physician on site who:
1. Is Board certified in Radiology or OB/GYN or the equivalent. (If they are not Board certified, they must be active status candidates for the next Board examination.); and
 2. Has completed a fellowship and had supplemental subspecialty training in:
 - a. maternal/fetal medicine or clinical genetics; or
 - b. diagnostic radiology, body imaging or the equivalent with an emphasis upon fetal medicine.

The supplemental training must be at a facility that performs at least 2000 second trimester fetal ultrasound exams a year and that meet the anatomical guidelines of the AIUM/ACR for complete fetal examinations. The supplemental training must include at least three months of targeted fetal ultrasound examinations that involve high-risk obstetric imaging and must include basic physics, techniques, performance, and interpretation followed by three months of proctoring, i.e., co-reading, by a qualified consultative sonologist; and

3. Has previously performed 500 detailed second trimester ultrasound exams on patients referred specifically for the detection of fetal abnormalities. Indications would include:

twins, early growth delay, oligohydramnios, polyhydramnios, abnormality observed at another facility, history of genetically transmitted disease, insulin dependent diabetes, family history of malformation and advanced maternal age.

The emphasis of these examinations is a detailed and targeted survey of fetal anatomy for malformations and must include:

fetal number, fetal presentation, documentation of fetal life, placental localization, amniotic fluid volume, gestational dating, detection and evaluation of maternal pelvic mass, and a survey of fetal anatomy for malformations; and

4. The solo practice consultative sonologist must perform a minimum of 200 detailed second trimester prenatal ultrasound exams annually on pregnancies at risk for fetal abnormalities. Each group practice consultative sonologist shall perform at least 150 detailed

second trimester prenatal ultrasound exams annually on pregnancies at risk for fetal abnormalities. A group practice is defined as a situation where all consultative sonologists practice at the same location.

5. All ultrasound practices at State-approved Prenatal Diagnosis Centers must be accredited by the American College of Radiology (ACR) or the American Institute of Ultrasound in Medicine (AIUM).

Any exceptions to the above criteria may be presented to an ultrasonographers' review panel for their recommendations regarding equivalent background and experience. Final decisions are made by the Genetic Disease Screening Program.

- E. Provides all clients choosing to terminate the pregnancy referral to a facility with assured access to second trimester abortions by a method which usually allows confirmation of diagnosis unless medically contraindicated.
- F. Has genetic counseling services which are performed by a qualified Genetic Counselor, Clinical Geneticist, or Ph.D. Medical Geneticist. At least one Genetic Counselor at each approved center must be Board certified or have Active Candidate status and pass the next Board examination.

1. A qualified Genetic Counselor is defined as:

- a. A person who has been certified by the American Board of Medical Genetics (ABMG) or American Board of Genetic Counseling (ABGC); or
- b. A recent graduate from a Master's or Ph.D. program in genetic counseling from an ABMG/ABGC or equivalent accredited training program who as soon as possible after graduation has applied and been accepted to take the next scheduled ABMG/ABGC examination; or
- c. An individual who fails the ABMG/ABGC certification exam for the first time but is still entitled to sit for re-examination in the next cycle. However, a Board certified Genetic Counselor, Ph.D. Medical Geneticist or Clinical Geneticist at the center must review and sign off on all cases on a weekly basis until Board certification is achieved in the next cycle.

An individual who fails the ABMG/ABGC certification exam for the second time will be designated as a Genetic Assistant until declared an Active Candidate by the ABMG/ABGC. A Board certified Genetic Counselor, Ph.D. Medical Geneticist or Clinical Geneticist at the center must review and sign off on all cases on a weekly basis until Board certification is achieved.

An individual who fails the ABMG/ABGC certification exam for a third time or more will be designated as a Genetic Assistant until Board certification is achieved.

2. Genetic Assistant is defined as an R.N. licensed by the Board of Registered Nursing with a minimum of a bachelor's degree in nursing and/or a person who has an earned bachelors, masters, or doctoral degree in nursing, biological sciences or other appropriate field. Genetic Assistants may only counsel patients who do not have a high risk of unusual or other potentially complex genetic abnormalities that are suspected as the result of previous family history, clinical investigation or laboratory tests. All Genetic Assistants must be directly supervised by a Board certified Genetic Counselor, Clinical Geneticist, or Ph.D. Medical Geneticist. Direct supervision is defined as the review of all cases on a weekly basis and documentation of such by signature.
 3. Prior to CVS and/or amniocentesis procedures, each woman must be offered genetic counseling under the supervision of a Clinical Geneticist and provide informed consent.
 4. Genetic Counselors, Ph.D. Medical Geneticists, and Genetic Assistants must attend continuing education courses, conferences, and/or grand rounds on genetically related topics for 30 hours within a two year period.
 5. All patients seen for genetic counseling must have a genetic risk assessment that includes the minimal elements contained in the Prenatal Genetic Screening Questionnaire and/or Pedigree. A pedigree is required if the patient, partner or fetus is at increased risk for a genetic disorder due to a significant family history indicated on the questionnaire.
- G. Utilizes a Genetic Disease Screening Program approved cytogenetics laboratory as evidenced by:
1. Direction by a cytogeneticist (M.D. and/or Ph.D.) certified as such by the American Board of Medical Genetics, and licensed by the State of California to direct a clinical cytogenetics laboratory.
 2. Testing performed by boarded or certified testing personnel, licensed by the State of California to perform clinical cytogenetic testing.
 3. Compliance with the current Pacific Southwest Regional Genetics Network (PSRGN) cytogenetic testing guidelines.
 4. Prior to independent prenatal cell culture and analysis, a new applicant center must meet criteria 1-3 above and provide evidence of quality assurance and quality control policies for the

implementation of prenatal testing. This must include establishing a consultative affiliation with an approved laboratory which is financially independent of the applicant agency for the first 25 samples of any new prenatal test, a review of the first 25 results of any test, and ongoing quality assurance/quality control indicators once the test is validated. Review of the first 25 samples by both applicant and consultative affiliate laboratories must be documented in a letter itemizing the cases and include a general overview of the quality assurance/quality control policies enacted for the ongoing monitoring of prenatal testing. A record of this review must be kept for ten years in an easily retrievable form.

5. Ability to perform or arrange for dysmorphism evaluation and/or pathologic examination of abortuses as well as cytogenetic and biochemical procedures.
 6. Continuing analyses of not less than 200 prenatal cell cultures per year with final results to meet turnaround times as outlined in the approved cytogenetic testing guidelines.
 7. Participation in and successful completion of any State of California provided or approved laboratory inspection, proficiency testing and/or quality control program including submission of appropriate documentation of participation and results.
 8. Submission of a written list identifying other recognized laboratories which have been used for the performance of specialized investigations for inherited diseases including biochemical and DNA studies.
 9. Assured access to resources for the determination of alpha fetoprotein and acetylcholinesterase concentrations in amniotic fluid for diagnosis of neural tube defects.
- H. Any trainee performing genetic services in a Prenatal Diagnosis Center must be under the direct, constant and on-site supervision of an appropriate specialist on the staff of an approved Comprehensive Prenatal Diagnosis Center.
- I. An interdisciplinary meeting of the Comprehensive Prenatal Diagnosis Center staff including ultrasonography, amniocentesis practitioner, genetic counseling, and medical genetics staff must be held at least once every three months.
- J. If a staff change(s) occurs such that an approved center no longer has the services of a Clinical Geneticist, a Medical Geneticist, Cytogeneticist, and/or Genetic Counselor certified by their respective Boards, whenever certification is required by these Standards, the personnel hired must be certified or have Active Candidate status and pass the next sitting of their Board certification exams. Until the time of Board certification, the center Director will arrange for consultation and supervision of the appropriate

areas by outside personnel who are Board certified and notify the Genetic Disease Screening Program of the consultation arrangement. All other changes in Prenatal Diagnosis Center staff referred to in these Standards must meet the criteria as described in Sections C, D, K, and L (if applicable).

- K. Where all transcervical Chorionic Villus Sampling (TC CVS) procedures are performed by:
1. Physicians who are American Board of Obstetrics and Gynecology certified/Active Candidates or equivalent, and have had specific training and special expertise in prenatal diagnosis. This training must include detailed obstetrical ultrasonography, as well as basic genetic information and appropriate counseling procedures for chromosomal, biochemical, and neural tube defects; and
 2. Physicians who have performed a total of at least 25 TC CVS procedures. These may be performed on women who are not planning to continue their pregnancies or on women referred for prenatal genetic indications and planning to continue their pregnancies. However, a minimum of 5 TC CVS procedures must be performed on women referred for prenatal genetic indications and planning to continue their pregnancies. All procedures performed on continuing pregnancies must have on-site supervision in the procedure room by an OB/GYN who is experienced in TC CVS. (Experienced is defined as having performed at least 25 TC CVS procedures on women continuing their pregnancies.); and
 3. Physicians who are approved as TA CVS practitioners or meet the Standards for approval as a TA CVS practitioner; and
 4. Each of the physicians performing or supervising TC CVS must perform at the rate of at least 25 CVS procedures, with at least 5 being TC CVS procedures, annually on women planning to continue their pregnancies.
- L. Where all transabdominal Chorionic Villus Sampling (TA CVS) procedures are performed by:
1. Physicians who are American Board of Obstetrics and Gynecology certified/Active Candidates or equivalent; and have had specific training and special expertise in prenatal diagnosis. This training must include detailed obstetrical ultrasonography, as well as basic genetic information and appropriate counseling procedures for chromosomal, biochemical, and neural tube defects.
 2. Such physicians shall have:
 - a. Been approved for and experienced in the performance of amniocentesis with ultrasound guidance, and have

performed at least 25 TA CVS procedures. These may be performed on women who are not planning to continue their pregnancies or on women referred for prenatal genetic indications and planning to continue their pregnancies. However, a minimum of 5 TA CVS procedures must be performed on women referred for prenatal genetic indications and planning to continue their pregnancies. All procedures performed on continuing pregnancies must have on-site supervision in the procedure room by an OB/GYN who is experienced in TA CVS. (Experienced is defined as having performed a minimum of 25 TA CVS procedures on women planning to continue their pregnancies); or

- b. Performed a minimum of 10 Percutaneous Umbilical Blood Sampling (PUBS) procedures or fetal intravenous transfusions on women planning to continue their pregnancies followed by a minimum of 15 TA CVS procedures with on-site supervision in the procedure room of a physician experienced in TA CVS; or
 - c. Been approved as a TC CVS practitioner and an amniocentesis practitioner; or
 - d. Met the Standards for approval as a TC CVS practitioner and amniocentesis practitioner; and
3. Each of the physicians performing or supervising TA CVS must perform at the rate of at least 25 CVS procedures (TC CVS, TA CVS or combination of both) annually on women planning to continue their pregnancies.
- M. In Centers offering CVS, if either TA or TC CVS is clinically contraindicated or unsuccessful, an appropriate alternative prenatal diagnosis procedure must be available either at that Prenatal Diagnosis Center or a referral must be made to another State-approved Prenatal Diagnosis Center.
- N. All amniocentesis and CVS practitioners must accumulate pregnancy outcome data as indicated on the adverse outcome form. Practitioners performing second trimester amniocentesis procedures must report on a statistically significant number of women who have had prenatal diagnostic amniocentesis or CVS procedures and who are planning to continue their pregnancies. Early amniocentesis practitioners are required to report the outcome of each procedure performed between 13 and 15 weeks gestation. Centers must submit each prenatal diagnostic practitioner's individual adverse outcome rate. (The adverse outcome rate is defined as the fetal loss rate minus the anomaly loss rate. The fetal loss rate is defined as the number of fetuses expiring at any time after the prenatal diagnostic procedure up to/ equal to 28 weeks gestation divided by the total number of viable fetuses at the time of the prenatal diagnosis

procedures. The anomaly loss rate is defined as the number of fetuses with congenital anomalies, genetic disorders, or chromosome abnormalities expiring at any time after the prenatal diagnosis procedure up to/equal to 28 weeks gestation divided by the total number of viable fetuses at the time of the prenatal diagnosis procedure. Studies with a start date of January 1, 2007, will have the fetal loss rate defined as the number of fetuses expiring at any time after the prenatal diagnostic procedure up to/ equal to 24 weeks gestation divided by the total number of viable fetuses at the time of the prenatal diagnosis procedures. Studies with a start date of January 1, 2007, will have the anomaly loss rate defined as the number of fetuses with congenital anomalies, genetic disorders, or chromosome abnormalities expiring at any time after the prenatal diagnosis procedure up to/equal to 24 weeks gestation divided by the total number of viable fetuses at the time of the prenatal diagnosis procedure. The adverse outcome rate is statistically adjusted to account for patients who are lost to follow-up). If the adjusted adverse outcome rate of an amniocentesis practitioner is greater than 3 percent, an early amniocentesis practitioner is greater than 5 percent, or CVS practitioner is greater than 6 percent, the Genetic Disease Screening Program with the assistance and advice of an appointed committee of Prenatal Diagnosis Center experts will evaluate and make a determination regarding the appropriateness of that physician to continue as an approved prenatal diagnosis practitioner. The Genetic Disease Screening Program will review all data received and will ask some practitioners who perform fewer than 50 amniocenteses per year or fewer than 25 CVS procedures per year to submit a progress report every six months until the study is complete.

- O. Each Prenatal Diagnosis Center must maintain a minimum annual volume of 100 women seen for prenatal genetic services. Prenatal genetic services are defined as those genetic services relating to the outcome of pregnancies.
- P. Each Prenatal Diagnosis Center must have an Internal Continuous Quality Improvement Program. The PDC Director must provide oversight to the program and work with PDC staff to achieve improvement goals.
- Q. Whenever a Satellite Prenatal Diagnosis Center decides to switch affiliation to another Comprehensive Prenatal Diagnosis Center, the Satellite Center must submit a letter to the original Prenatal Diagnosis Center Director and to the Genetic Disease Screening Program informing them of their intent to switch affiliations. The original Prenatal Diagnosis Center Director must then submit a letter to the Genetic Disease Screening Program acknowledging the intent of the Satellite Center switch. The letter must include whether or not there are any outstanding issues with the site regarding compliance to the PDC Standards and California Prenatal Screening Program Guidelines. The letter must be sent to the Genetic Disease Screening Program within thirty days from the original request of the Satellite Center requesting the switch.

If there are outstanding data that are required to be submitted (e.g. quarterly Prenatal Diagnosis Center reports, Adverse Outcome Studies), the Genetic Disease Screening Program will not approve the switch until the data have been successfully completed and submitted to the Screening Program. It is the responsibility of the Satellite center requesting the switch to ensure that the data are complete before requesting the switch in affiliation. If the Comprehensive Prenatal Diagnosis Center that is taking on the new site(s) wants to assume the responsibility for completing the data, they must submit assurance in writing to the Genetic Disease Screening Program that the data will be submitted within a specified time period. The approval will be time limited until the data are submitted.

- R. Abnormal or ambiguous results of amniocentesis or CVS procedures must be verbally communicated to referring physicians and/or patients by clinical genetics staff such as M.D. Clinical Geneticists, Ph.D. Medical Geneticists, Clinical Cytogeneticists, or Genetic Counselors. The center must have a written protocol in place and must take responsibility for reporting normal results of amniocentesis or CVS procedures.
- S. Providers will be given provisional approval as TA CVS practitioners, TC CVS practitioners, or amniocentesis providers under the following terms:

- 1. Delinquent reporter

Approved amniocentesis and CVS practitioners who fail to provide adverse outcome data for more than two years from the study start date or submission of a progress report will be given provisional approval as a Delinquent Reporter. These Delinquent Reporters must report the outcomes of pregnancy to 28 weeks on at least 90% of procedures via a progress report or log every six months until their studies are completed. Delinquent Reporters that are in studies with a start date of January 1, 2007, must report the outcomes of pregnancy to 24 weeks on at least 90% of procedures via a progress report or log every six months until their studies are completed. If there is no submission of data or documentation of attempts to submit the data within six months of being given provisional status, practitioner approval will be withdrawn.

- 2. Category A Provisional Practitioner

Approved amniocentesis and CVS practitioners that have two consecutive years of low volume, defined as less than 50 second trimester amniocentesis procedures per year or less than 25 CVS procedures per year; and:

- a. Have not completed any adverse outcome studies; or
- b. Have completed one adverse outcome study; or
- c. Are Delinquent Reporters

will be given provisional approval as a Category A Provisional Practitioner.

These Category A Provisional Practitioners must complete a new adverse outcome study by reporting the outcomes of pregnancy to 28 weeks on at least 90% of procedures via a progress report or log every six months until their studies are completed. Category A Provisional Practitioners that are in studies with a start date of January 1, 2007, or later must report the outcomes of pregnancy to 24 weeks on at least 90% of procedures via a progress report or log every six months until their studies are completed. If there is no submission of data or documentation of attempts to submit the data within six months of being given provisional status, practitioner approval will be withdrawn.

3. Category B Provisional Practitioner

Approved amniocentesis and CVS practitioners that have successfully completed two adverse outcome studies, and then have two consecutive years of low volume, defined as less than 25 CVS or at least 25 second trimester amniocentesis procedures per year, will be given provisional approval as a Category B Provisional Practitioner.

These Category B Provisional Practitioners must:

- a. Complete a new adverse outcome study by reporting the outcomes of pregnancy to 24 weeks on at least 90% of procedures via a progress report or log every six months until their studies are completed; and
- b. Be re-reviewed for approval status by the CQI Committee within one year of the Category B Provisional Practitioner status being awarded.

If there is no submission of data or documentation of attempts to submit the data within six months of being given provisional status, practitioner approval will be withdrawn.

4. A practitioner requesting a leave of absence for personal reasons or sabbatical leave should submit a letter to the Genetic Disease Screening Program (GDSP) outlining specific reasons for the request and listing any outstanding approval conditions or reporting compliance issues. Leave of absence requests will be reviewed by the Perinatal Subcommittee with final approval by GDSP.
5. In order to re-approve a TC CVS practitioner after one year of having practitioner approval status withdrawn, the practitioner must perform 5 TC CVS procedures with on-site supervision in the procedure room by an OB/GYN who is experienced in performing TC CVS. (Experienced is defined as having performed a minimum

of 25 TC CVS procedures on women planning to continue their pregnancies)

6. In order to re-approve a TC CVS practitioner after two years of having practitioner approval status withdrawn, the practitioner must perform 10 TC CVS procedures with on-site supervision in the procedure room by an OB/GYN who is experienced in performing TC CVS. (Experienced is defined as having performed a minimum of 25 TC CVS procedures on women planning to continue their pregnancies)
7. In order to reapprove a TA CVS practitioner after having practitioner approval status withdrawn, the practitioner must be an approved amniocentesis practitioner and must perform 5 TA CVS procedures with on-site supervision in the procedure room by an OB/GYN who is experienced in performing TA CVS. (Experienced is defined as having performed a minimum of 25 TA CVS procedures on women planning to continue their pregnancies)

II. SATELLITE PRENATAL DIAGNOSIS CENTER

A Satellite Prenatal Diagnosis Center is a center with the following prenatal diagnosis services:

- A. Provides on site genetic counseling, ultrasonography, and the collection of amniotic fluid/CVS specimens at a site which is not in the same suite as an existing prenatal diagnosis center/satellite.
- B. Provides on-site counseling by a Board certified/Active Candidate Clinical Geneticist, Ph.D. Medical Geneticist, or Genetic Counselor prior to CVS and/or amniocentesis. The Clinical Geneticist must be available whenever possible at the satellite site within three working days to provide consultation in person to all families with abnormal or questionable results. The Clinical Geneticist must be available within reasonable travel time (less than eight hours) to all assigned sites of service.
- C. The Clinical Geneticist will conduct monthly meetings with clinical staff who are assigned to the site to include, but not be limited to, case review.
- D. Has a written agreement with a State-approved Comprehensive Prenatal Diagnosis Center and an approved cytogenetic laboratory which also performs necessary laboratory studies. The Clinical Geneticist of the Comprehensive Prenatal Diagnosis Center will assume complete responsibility for the accuracy of genetic counseling. The Director of the Comprehensive Prenatal Diagnosis Center is responsible for the adequacy of the amniotic fluid/CVS samples and follow-up services.

- E. Has an established mechanism for safely and rapidly delivering satisfactory amniotic fluid/CVS samples to the affiliated Comprehensive Prenatal Diagnosis Center or to the approved cytogenetics laboratory.
- F. Complies with criteria B, C, D, E, F, G, H, I, J, K, L, M, N, O, P, Q, R, and S as described for a Comprehensive Prenatal Diagnosis Center.

Any requests for exceptions to the Standards must be documented in a letter to the Genetic Disease Screening Program requesting a waiver and outlining temporary coverage as well as future plans to comply with the Standards. Waivers will be considered only in extreme circumstances and must be justified as necessary to provide access to services in underserved areas. Final decisions are made by the Genetic Disease Screening Program.

California Department of Public Health
 Genetic Disease Screening Program
 850 Marina Bay Parkway, F175
 Richmond, CA 94804

Required Documentation for New Entity Requests

**Entity	GDSP attachment (completed, signed, and dated)	Documentation of Professional Status and/or Credentialing	Screening Information System (SIS)		
			SIS Oath of Confidentiality	SIS PDC e-learning assessment	E-mail address
Genetic Counselor	C-6	√	√	√	√
Ph.D. Medical Geneticist	C-7	√	√	√	√
Amniocentesis Practitioner	C-1 (2-sided form)				
Transabdominal (TA) CVS practitioner	C-4 (2-sided form)				
Transcervical (TC) CVS practitioner	C-3 (2-sided form)				
Consultative Sonologist	C-5 (2-sided form)	Curriculum Vitae (CV)			
Clinical Geneticist	B	√	√		* If applicable
PDC Director	A	√	√		* If applicable
Cytogenetic Laboratory	D				
PDC Scheduling Contact			√	√	√
PSR Contact			√	√	√
PDC Invoice Liaison			√		√

* If applicable- means if PDC Director and/or Clinical Geneticist will need access to SIS, then an e-mail address is required.

** If an entity already exists with the Comprehensive PDC and the request is to add the entity to PDC satellites already affiliated with the Comprehensive PDC, then the appropriate and completed GDSP attachment can be faxed by the PDC Director or PDC Contact for the Comprehensive PDC. The request must be received within 10 business days of any changes in locations or staffing of locations. Originals should be mailed to the PSQA Branch.

PRENATAL GENETIC SCREENING QUESTIONNAIRE

Name _____ Date of Birth _____ LMP _____

How old will **you** be when the baby is born? _____ Your occupation? _____

How old will the **baby's father** be when the baby is born? _____ His occupation? _____

Answer each question below and provide an explanation when applicable.

(Please mark all that apply because some genetic disorders are more common in certain ethnic groups.)

1. Is your family or your baby's father's family...
 - a. Southeast Asian, Taiwanese, Chinese, or Filipino? No Yes
 - b. Italian, Greek, Middle Eastern, East Indian or Pakistani? No Yes
 - c. African or African-American (Black)? No Yes
 - d. Jewish? No Yes
 - e. Cajun or French Canadian? No Yes
 - f. Caucasian? No Yes

2. Are you and the baby's father related by blood (such as cousins)? No Yes

3. Have you, the baby's father, or anyone in either of your families ever had any of the following disorders or pregnancy histories?
 - a. Any chromosomal abnormalities (such as Down syndrome) No Yes
 - b. Any neural tube defect (spina bifida, anencephaly) No Yes
 - c. Any blood disorder (such as hemophilia, sickle cell, thalassemia, clotting disorder) No Yes
 - d. Any nerve or muscle disorder (neurofibromatosis, muscular dystrophy) No Yes
 - e. Any bone or skeletal disorder (such as dwarfism) No Yes
 - f. Cystic fibrosis (a lung disease) No Yes
 - g. Polycystic kidney disease No Yes
 - h. Heart defect (at birth) No Yes
 - i. Cleft lip/palate No Yes
 - j. Mental retardation No Yes
 - k. Any genetic condition not listed above No Yes
 - l. Any birth defect not listed above No Yes
 - m. A serious medical problem that you are concerned about (such as diabetes) No Yes
 - n. Had a baby who died shortly after birth or in childhood? No Yes
 - o. Had a stillbirth or two or more pregnancy losses? No Yes
 - p. Needed surgery before one year of age? No Yes
 - q. Any childhood cancer or early malignancy? No Yes

4. Have you or the baby's father had any genetic tests (such as cystic fibrosis, Tay-Sachs, Canavan or sickle cell screening)? Other: _____ No Yes
(specify)

5. Was this pregnancy achieved through in-vitro fertilization (IVF) or other assisted reproductive methods?..... No Yes
 If yes, was there: Sperm donor Egg donor ICSI Other: _____

6. Have you used tobacco, alcohol or street drugs since being pregnant or since your last menstrual period?..... No Yes

7. Excluding vitamins and iron, have you taken any medications since being pregnant or since your last menstrual period?..... No Yes

8. Have you had the Expanded AFP Screening blood test?..... No Yes
 If yes, when?

9. If yes to any question above, explain:

Completed by _____

Date _____

Reviewed by _____

Date _____

CUESTIONARIO DE TAMIZ GENÉTICO PRENATAL

Nombre _____ Fecha de Nacimiento _____ Última menstruación _____

¿Qué edad tendrá **usted** cuando nazca su bebé? _____ ¿Cuál es su ocupación? _____

¿Qué edad tendrá **el padre** cuando nazca el bebé? _____ ¿Cuál es la ocupación del padre? _____

Conteste las siguientes preguntas y explique si es necesario.

(Marque todo lo que le aplique ya que algunas enfermedades genéticas son más comunes en ciertos grupos étnicos)

1. ¿Es su familia o la familia del padre del bebé...
 - a. Del Sureste Asiático, Taiwanes, Chino o Filipino? No Sí
 - b. Italiano, Griego, del Medio Oriente, de la India Oriental o Pakistán? No Sí
 - c. Africano o Africano-Americano (Negro)? No Sí
 - d. Judío? No Sí
 - e. Cajún o Canadiense Francés? No Sí
 - f. Caucásico/Anglosajón/Angloamericano? No Sí
2. ¿Está Ud. emparentada por sangre con el padre del bebé (por ejemplo, primos)? No Sí
3. ¿Usted, o el padre del bebé o algún familiar de ustedes ha tenido alguna de las siguientes enfermedades o antecedentes de embarazos?
 - a. Anormalidad cromosómica (como el síndrome de Down) No Sí
 - b. Defecto del tubo neural (espina bífida, anencefalia) No Sí
 - c. Enfermedad de la sangre (hemofilia, anemia falciforme, talasemia, enfermedad de coagulación) No Sí
 - d. Enfermedad de los nervios o los músculos (neurofibromatosis, distrofia muscular) No Sí
 - e. Enfermedad de los huesos o del sistema esquelético (por ejemplo, enanismo) No Sí
 - f. Fibrosis quística (una enfermedad de los pulmones) No Sí
 - g. Enfermedad de riñones poliquísticos No Sí
 - h. Defecto del corazón (de nacimiento) No Sí
 - i. Labio / paladar hendido (leporino) No Sí
 - j. Atraso mental No Sí
 - k. Alguna condición genética no mencionada arriba No Sí
 - l. Algún defecto de nacimiento no mencionado arriba No Sí
 - m. Un problema médico serio que le preocupe a Ud. (tal como diabetes) No Sí
 - n. ¿Ha perdido algún bebé poco tiempo después de nacido o en la infancia? No Sí
 - o. ¿Le ha nacido muerto un bebé o ha perdido 2 o más embarazos? No Sí
 - p. ¿Un bebé que haya requerido operación antes del primer año de edad? No Sí
 - q. ¿Algún cáncer infantil o tumor maligno temprano? No Sí
4. ¿Le han hecho alguna prueba genética a Ud. o al padre del bebé (por ejemplo, para fibrosis quística, Tay-Sachs, Canavan o células falciformes)? Otras: _____ No Sí
(especifique)
5. ¿Este embarazo se logró mediante fertilización in vitro (IVF) o por otro método de asistencia reproductiva?..... No Sí
De ser así, hubo un donante de espermatozoides donante de óvulo ICSI Otro: _____
6. ¿Ha consumido tabaco, alcohol o drogas de calle desde que empezó su embarazo o desde su última menstruación?..... No Sí
7. Excluyendo vitaminas y hierro, ¿ha tomado algún medicamento desde que empezó su embarazo o desde su última menstruación?..... No Sí
8. ¿Le han hecho la prueba de sangre llamada Extendida de AFP (alpha-fetoproteína)?.... No Sí
De ser así, ¿cuándo? _____
9. Si contestó "sí" a alguna pregunta, por favor explique: _____

Completado por: _____

Fecha _____

Reviewed by _____

Date _____

PRENATAL GENETIC SCREENING PEDIGREE

Patient Name _____ Patient MR# _____ LMP _____

Patient's Age _____ Patient's Occupation _____ FOB's Age _____ FOB's Occupation _____

Family and Patient History				KEY		
	No	Yes		No	Yes	
Consanguinity	<input type="checkbox"/>	<input type="checkbox"/>	Other genetic condition or birth defect	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Down syndrome	<input type="checkbox"/>	<input type="checkbox"/>	Any serious medical problem (diabetes, Lupus)	<input type="checkbox"/>	<input type="checkbox"/>	
Other chromosomal abnormalities	<input type="checkbox"/>	<input type="checkbox"/>	Any baby who died shortly after birth orin childhood	<input type="checkbox"/>	<input type="checkbox"/>	
Neural tube defect	<input type="checkbox"/>	<input type="checkbox"/>	Stillborn or 2 or more first trimester spontaneouspregnancy losses	<input type="checkbox"/>	<input type="checkbox"/>	
Blood disorder	<input type="checkbox"/>	<input type="checkbox"/>	Needed surgery before 1 year of age	<input type="checkbox"/>	<input type="checkbox"/>	
Nerve or Muscle disorder	<input type="checkbox"/>	<input type="checkbox"/>	Childhood cancer or early malignancy	<input type="checkbox"/>	<input type="checkbox"/>	
Bone or skeletal disorder	<input type="checkbox"/>	<input type="checkbox"/>	Any genetic tests or screenings	<input type="checkbox"/>	<input type="checkbox"/>	
Cystic fibrosis	<input type="checkbox"/>	<input type="checkbox"/>	IVF, ICSI, or other assisted reproductive methods	<input type="checkbox"/>	<input type="checkbox"/>	
Tay-Sachs/Canavan	<input type="checkbox"/>	<input type="checkbox"/>	Tobacco/Alcohol/Drug exposure(s)	<input type="checkbox"/>	<input type="checkbox"/>	
Polycystic kidney disease	<input type="checkbox"/>	<input type="checkbox"/>	Medication exposure(s)		<input type="checkbox"/>	
Heart defect (at birth)	<input type="checkbox"/>	<input type="checkbox"/>				
Cleft lip/palate	<input type="checkbox"/>	<input type="checkbox"/>				

Check all that apply for the patient or baby's father's ancestry:

- | | Patient | FOB | | Patient | FOB |
|---|--------------------------|--------------------------|------------------------------|--------------------------|--------------------------|
| a. Southeast Asian, Taiwanese, Chinese or Filipino? | <input type="checkbox"/> | <input type="checkbox"/> | e. Cajun or French Canadian? | <input type="checkbox"/> | <input type="checkbox"/> |
| b. Italian, Greek, Middle Eastern, East Indian, or Pakistani? | <input type="checkbox"/> | <input type="checkbox"/> | f. Caucasian? | <input type="checkbox"/> | <input type="checkbox"/> |
| c. African or African-American (Black)? | <input type="checkbox"/> | <input type="checkbox"/> | g. Other? Patient: _____ | | |
| d. Jewish? | <input type="checkbox"/> | <input type="checkbox"/> | FOB: _____ | | |

Comments: _____

Completed by _____ Date _____

Reviewed by _____ Date _____

STATEMENT OF INFORMED CONSENT/REFUSAL FOR AMNIOCENTESIS

1. I have been informed that the purpose of amniocentesis is to detect fetal chromosomal disorders, neural tube defects, and other specific disorders of the fetus.
2. I have been informed that before the amniocentesis is performed I will have an ultrasound examination to help locate the placenta and fetus. Ultrasound may also detect twins, incorrect dating of the pregnancy, and some other conditions.
3. I have been informed that amniocentesis involves inserting a needle through the woman's abdomen into the fluid in her uterus which surrounds the fetus. A small amount of fluid (less than one ounce) is taken out and tested. There may be some discomfort when the needle is inserted.
4. I have been informed that there are serious complications in less than 1% of amniocenteses performed, based on currently available information. These include miscarriage, hemorrhage, infection, premature rupture of the membranes, or injury to the fetus or fetal death. Minor complications include cramping, vaginal spotting or slight leakage of amniotic fluid, and soreness where the needle was inserted.

_____ I have requested early amniocentesis (13 weeks 0 days to 14 weeks 6 days gestation). I have been informed that early amniocentesis may be associated with a higher risk than standard amniocentesis (at or after 15 weeks gestation) for pregnancy loss, amniotic fluid leakage, and/or club foot deformity.

5. I have been informed that fewer than 1 in 100 amniocenteses need to be repeated because not enough fluid is obtained the first time. Occasionally, even though fluid is obtained, a diagnosis cannot be made.
6. I have been informed that amniocentesis can identify over 99 percent of all chromosomal disorders and over 90 percent of all open neural tube defects. However, a complete and correct diagnosis of the condition of the fetus cannot be guaranteed.
7. I have been informed that not all birth defects can be detected by amniocentesis or ultrasonography.
8. I have been informed that in the case of twins or triplets, the results may pertain to only one of the fetuses.
9. I have been informed that all abnormal findings will be explained to me. Treatment alternatives will be discussed. The decision to continue or to have the pregnancy terminated is entirely mine.
10. I have been informed that my participation in this procedure is entirely voluntary. Refusing this procedure will not make me ineligible for any services supported by State funding.
11. My signature below indicates that:

I have read, or had read to me, the above information and I understand it. I have had an opportunity to discuss it, including the purpose and possible risks of amniocentesis, with my doctor or the doctor performing the procedure. I have received all of the information I want. My questions all have been answered.

Yes	<p>I REQUEST that Dr. _____ and/or associates perform amniocentesis. I understand and accept the consequences of this decision.</p> <p>Signed _____ Date _____</p> <p>Witnessed by _____ Date _____</p>
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No	<p>I DECLINE to have amniocentesis. I understand and accept the consequences of this decision.</p> <p>Signed _____ Date _____</p> <p>Witnessed by _____ Date _____</p>
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The California Information Practices Act of 1977 (Civil Code 1798 et seq.) requires that the following information be provided. The California State Department of Health Services, Genetic Disease Branch, will receive a confidential report of all abnormal tests performed by state-approved prenatal diagnosis centers. This information is collected under the authority of California Code of Regulations Sections 6531 and 6532. This information will be used to ensure that all approved prenatal diagnostic centers meet state standards for services and to improve the detection, prevention, and treatment of birth defects.

The Department may contact you to collect additional information in connection with special studies of birth defects. Your participation in these studies is voluntary and all information collected by the Department is strictly confidential and personal information will not be released to any other person or agency without your written approval. If you have any questions, requests, or complaints about the use of your personal health information or desire a copy of the Notice of Information and Privacy Practices as required by the Health Insurance Portability and Accountability Act of 1996 (HIPAA), please contact the Chief of the Genetic Disease Branch, 850 Marina Bay Parkway; Richmond, CA 94804, (510) 412-1502.

**STATEMENT OF INFORMED CONSENT/REFUSAL FOR
TRANSCERVICAL OR TRANSABDOMINAL CHORIONIC VILLUS SAMPLING
(TC CVS OR TA CVS) ≥ 10 weeks post L.M.P. or Equivalent**

1. I have been informed that the purpose of Chorionic Villus Sampling is to detect fetal chromosomal disorders and other specific disorders of the fetus.
2. I have been informed that there is another method of obtaining diagnostic information about the chromosomal or biochemical status of the fetus. This procedure, called amniocentesis, is usually done in the second trimester at 15 to 18 weeks from my last menstrual period or equivalent. It involves placing a needle through the abdominal wall into the fluid which surrounds the fetus in the uterus and removing a small amount of fluid. The risks and benefits of amniocentesis versus CVS have been explained to me in detail.
3. I have been informed that before the CVS is performed, I will have an ultrasound examination to help locate the placenta and fetus. Ultrasound may also detect twins, incorrect dating of the pregnancy, and some other conditions.
4. I have been informed that the TC CVS procedure consists of inserting a catheter (sterile plastic tube) under ultrasound guidance through the woman's cervix to obtain placental tissue. I have been informed that the TA CVS procedure consists of inserting a needle under ultrasound guidance through the woman's abdomen to the placenta to obtain placental tissue.
5. I have been informed that the exact risk of serious complications is unknown but has been reported to be between 1 in 200 and 6 in 200 above the background risk. The most common serious complication is miscarriage. Other complications are bleeding, rupture of the membranes, uterine perforation, or infections. Minor complications include cramping, vaginal spotting, or slight leakage of amniotic fluid for up to 2 weeks.
6. Some studies have suggested that CVS may be associated with a risk for fetal limb abnormalities. Most studies, however, seem to indicate that procedures performed at 10 weeks of pregnancy or greater pose little if any increased risk.
7. I have been informed that occasionally CVS procedures need to be repeated because not enough tissue is obtained the first time. It is possible that, even though tissue is obtained, a diagnosis cannot be made. However, in over 95% of cases, sufficient tissue is obtained and a diagnostic evaluation can be performed.
8. I have been informed that if multiple conceptions (twins, triplets, etc.) are present, attempts to obtain chorionic villi surrounding the developing fetus may not be successful. Amniocentesis may be required during the second trimester of pregnancy.
9. I have been informed that not all birth defects can be detected by CVS and the accompanying ultrasonography, but CVS can identify over 99% of all chromosomal disorders. However, a complete and correct diagnosis of the fetus cannot be guaranteed.
10. I have been informed that an amniocentesis or fetal blood sampling may be recommended to clarify the results of CVS.
11. I have been informed that all abnormal findings will be explained to me. Treatment alternatives will be discussed. The decision to continue or to have the pregnancy terminated is entirely mine.
12. I have been informed that there is a small chance that the chorionic villi obtained do not represent the genetic make-up of the fetus, either due to chromosomal mosaicism (2 or more cell types) or maternal cell contamination.
13. I have been informed that my participation in this procedure is entirely voluntary. I can refuse this procedure and still receive medical care.
14. I have been informed that CVS procedures do not detect most neural tube defects. Therefore, a blood test which can screen for neural tube defects (MS AFP test) will be offered to me at 16-18 weeks from my last menstrual period or equivalent.

Yes	<p>I REQUEST that Dr. _____ and/or associates perform CVS. I understand and accept the consequences of this decision.</p> <p>Signed _____ Date _____</p> <p>Witnessed by _____ Date _____</p>
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No	<p>I DECLINE to have CVS. I understand and accept the consequences of this decision.</p> <p>Signed _____ Date _____</p> <p>Witnessed by _____ Date _____</p>
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The California Information Practices Act of 1977 (Civil Code 1798 et seq.) requires that the following information be provided. The California State Department of Health Services, Genetic Disease Branch, will receive a confidential report of all abnormal tests performed by state-approved prenatal diagnosis centers. This information is collected under the authority of California Code of Regulations Sections 6531 and 6532. This information will be used to ensure that all approved prenatal diagnostic centers meet state standards for services and to improve the detection, prevention, and treatment of birth defects.

The Department may contact you to collect additional information in connection with special studies of birth defects. Your participation in these studies is voluntary and all information collected by the Department is strictly confidential and personal information will not be released to any other person or agency without your written approval. If you have any questions, requests, or complaints about the use of your personal health information or desire a copy of the Notice of Information and Privacy Practices as required by the Health Insurance Portability and Accountability Act of 1996 (HIPAA), please contact the Chief of the Genetic Disease Branch, 850 Marina Bay Parkway; Richmond, CA 94804, (510) 412-1502.
DHS 4457 (05-12)

Declaración de consentimiento o rechazo fundamentado de amniocentesis

1. Se me ha informado que el propósito de la amniocentesis es detectar trastornos cromosómicos del feto, defectos del tubo neural, y otros trastornos específicos del feto.
2. Se me ha informado que antes de hacer la amniocentesis, se me hará un examen de ultrasonido para ayudar a localizar la placenta y el feto. Además, es posible que el ultrasonido identifique mellizos (o gemelos), fecha incorrecta del embarazo, y algunas otras condiciones.
3. Se me ha informado que la amniocentesis consiste en la introducción de una aguja a través del abdomen de la mujer, hasta el líquido que rodea al feto en el útero. Se extrae una pequeña cantidad de líquido (menos de una onza) para ser examinada. Es posible que sienta cierta molestia cuando se introduzca la aguja.
4. Se me ha informado que ocurren complicaciones graves en menos del 1% de las amniocentesis que se llevan a cabo, de acuerdo a los datos disponibles en la actualidad. Las complicaciones incluyen el aborto espontáneo, hemorragias, infección, rotura prematura de las membranas, lesión al feto y la muerte del feto. Las complicaciones menores incluyen cólicos abdominales, sangrado vaginal leve o goteo leve de fluido amniótico, y dolor en el área en que se introdujo la aguja.

_____ He solicitado una amniocentesis temprana (13 semanas y 0 días a 14 semanas y 6 días de gestación). Se me ha informado que la amniocentesis temprana puede presentar un riesgo más elevado de pérdida del embarazo, pérdidas de líquido amniótico y de deformidades de pie zambo, que la amniocentesis que se realiza normalmente (después de las 15 semanas de gestación).

5. Se me ha informado que menos de una de cada 100 amniocentesis necesitan repetirse debido a que no se obtiene el líquido suficiente la primera vez. Ocasionalmente, aun cuando se obtiene el líquido, no se puede dar un diagnóstico.
6. Se me ha informado que la amniocentesis puede identificar más del 99 por ciento de todos los trastornos cromosómicos, y más del 90 por ciento de todos los defectos de tubo neural abierto. Sin embargo, no se puede garantizar un diagnóstico completo y correcto de la condición del feto.
7. Se me ha informado que no todos los defectos de nacimiento se pueden detectar por amniocentesis o ultrasonografía.
8. Se me ha informado que en caso de mellizos o trillizos, es posible que los resultados sólo sean pertinentes a uno de los fetos.
9. Se me ha informado que me explicarán cualquier resultado anormal. Me hablarán sobre las alternativas de tratamiento. La decisión de continuar o terminar el embarazo será completamente mía.
10. Se me ha informado que mi participación en este procedimiento es completamente voluntaria. Negarme a participar en este procedimiento no afectará mi elegibilidad para recibir servicios financiados con fondos estatales.
11. Mi firma a continuación indica que:

He leído, o me han leído la información anterior, y la entiendo. He tenido la oportunidad de hablar con mi doctor o el doctor que hará el procedimiento, sobre el propósito y los posibles riesgos de la amniocentesis. He recibido toda la información que deseo. Todas mis preguntas han sido contestadas.

SI	<p>SOLICITO que el Dr. / la Dra. _____ y/o sus asociados me hagan una amniocentesis. Entiendo y acepto las consecuencias de esta decisión.</p> <p>Firma _____ Fecha _____</p> <p>Firma del testigo _____ Fecha _____</p>
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NO	<p>RECHAZO que me hagan la amniocentesis. Entiendo y acepto las consecuencias de esta decisión.</p> <p>Firma _____ Fecha _____</p> <p>Firma del testigo _____ Fecha _____</p>
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La Ley de California de 1977 de Prácticas de Información (Código Civil § 1798 y siguientes) requiere que se proporcione la siguiente información. La División de Enfermedades Genéticas del Departamento de Servicios de Salud de California recibirá un informe confidencial de todas las pruebas anormales realizadas por centros de diagnóstico prenatal aprobados por el estado. Esta información se obtiene bajo la autoridad de las secciones 6531 y 6532 del Código de Reglamentaciones de California. Esta información se utilizará para asegurar que todos los centros de diagnóstico prenatal aprobados cumplan las normas estatales de servicios y para mejorar la detección, prevención y tratamiento de defectos de nacimiento.

El Departamento se podrá poner en contacto con usted para obtener más información relativa a estudios especiales de defectos de nacimiento. Su participación en esos estudios es voluntaria y toda la información obtenida por el Departamento es estrictamente confidencial y no se entregará información personal a ninguna otra persona o entidad sin su aprobación por escrito. Si tiene alguna pregunta, pedido o queja sobre el uso de información personal sobre su salud o desea obtener una copia del Aviso de prácticas de información y privacidad, como lo requiere la Ley de 1996 de Transferibilidad y Responsabilidad del Seguro de Salud (HIPAA), póngase en contacto con el jefe de la división de enfermedades genéticas: Chief of the Genetic Disease Branch, Dr. George C. Cunningham, M.P.H.; 850 Marina Bay Parkway; Richmond, CA 94804, (510) 412-1502.

**Declaración de consentimiento o rechazo fundamentado de toma transcervical
o transabdominal de muestras de vellosidades coriónicas
(TC CVS O TA CVS) > 10 semanas después del último período menstrual o equivalente**

1. Se me ha informado que el propósito de la toma de muestras de vellosidades coriónicas es detectar trastornos cromosómicos y otros trastornos específicos del feto.
2. Se me ha informado que hay otro método para obtener información diagnóstica sobre el estado cromosómico o el estado bioquímico del feto. Este examen diagnóstico, llamado amniocentesis, por lo general se hace en el segundo trimestre, a las 15 a 18 semanas desde mi último período menstrual o equivalente. Requiere la inserción de una aguja a través de la pared abdominal hasta el líquido que rodea al feto en el útero y extraer una pequeña cantidad de líquido. Me han explicado en detalle los riesgos y los beneficios de la amniocentesis comparados con los de la CVS.
3. Se me ha informado que antes de hacerme la CVS me harán un examen de ultrasonido para ayudar a ubicar la placenta y el feto. El ultrasonido también puede detectar mellizos, fechas incorrectas del embarazo y algunos otros trastornos.
4. Se me ha informado que el examen CVS transcervical consiste en insertar un catéter (tubo de plástico estéril) con guía de ultrasonido por el cuello del útero de la mujer para obtener tejido de la placenta. Se me ha informado que el examen CVS transabdominal consiste en insertar una aguja con guía de ultrasonido a través del abdomen de la mujer hasta la placenta para obtener tejido de la placenta.
5. Se me ha informado que el riesgo exacto de que ocurran complicaciones serias se desconoce, pero se ha informado que es de entre 1 en 200 y de 6 en 200 por encima del riesgo de fondo. La complicación más seria es el aborto espontáneo. Otras complicaciones son hemorragia, ruptura de las membranas, perforación del útero o infecciones. Las complicaciones menores incluyen dolores, manchas de sangre vaginal o goteo leve de líquido amniótico por hasta 2 semanas.
6. Algunos estudios han sugerido que la CVS puede estar vinculada a riesgo de anomalías en los miembros del feto. Sin embargo, la mayoría de los estudios parecen indicar que los exámenes realizados a las 10 semanas de embarazo o más presentan poco, o ningún incremento del riesgo.
7. Se me ha informado que ocasionalmente es necesario repetir los exámenes CVS porque no se obtuvo suficiente tejido la primera vez. Es posible que, incluso si se obtiene tejido, no se pueda hacer un diagnóstico. Sin embargo, en más del 95% de los casos se obtiene suficiente tejido para hacer un diagnóstico y la evaluación se puede realizar.
8. Se me ha informado que en casos de concepciones múltiples (mellizos, trillizos, etc.), los intentos de obtener vellosidades coriónicas que rodean al feto en desarrollo pueden no ser exitosos. Puede ser necesario hacer una amniocentesis en el segundo trimestre del embarazo.
9. Se me ha informado que no todos los defectos de nacimiento se pueden detectar por CVS y la ultrasonografía que la acompaña, pero la CVS puede identificar más del 99% de los trastornos cromosómicos. Sin embargo, no se puede garantizar un diagnóstico completo y correcto del feto.
10. Se me ha informado que la amniocentesis o la obtención de una muestra de sangre del feto se puede recomendar para aclarar los resultados de la CVS.
11. Se me ha informado que me explicarán todos los resultados anormales y me hablarán sobre las alternativas de tratamiento. La decisión de continuar el embarazo o de darle fin es enteramente mía.
12. Se me ha informado que hay una pequeña posibilidad de que las vellosidades coriónicas obtenidas no representen la configuración genética del feto, ya sea por mosaiquismo cromosómico (2 ó más tipos de células) o por contaminación de células maternas.
13. Se me ha informado que mi participación en este examen es completamente voluntaria. Puedo rechazar este examen e igual recibir atención médica.
14. Se me ha informado que los exámenes CVS no detectan la mayoría de los defectos del tubo neural. Por lo tanto, se me ofrecerá un análisis de sangre a las 16 a 18 semanas de mi último período menstrual o equivalente que puede detectar defectos del tubo neural (prueba MS AFP).

Sí	<p>SOLICITO que el Dr. / la Dra. _____ y/o sus asociados me hagan una CVS. Entiendo y acepto las consecuencias de esta decisión.</p> <p>Firmado _____ Fecha _____</p> <p>Firma del testigo _____ Fecha _____</p>
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No	<p>RECHAZO que me hagan la prueba CVS. Entiendo y acepto las consecuencias de esta decisión.</p> <p>Firmado _____ Fecha _____</p> <p>Firma del testigo _____ Fecha _____</p>
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El Departamento se podrá poner en contacto con usted para obtener más información relativa a estudios especiales de defectos de nacimiento. Su participación en esos estudios es voluntaria y toda la información obtenida por el Departamento es estrictamente confidencial y no se entregará información personal a ninguna otra persona o entidad sin su aprobación por escrito. Si tiene alguna pregunta, pedido o queja sobre el uso de información personal sobre su salud o desea obtener una copia del Aviso de prácticas de información y privacidad, como lo requiere la Ley de 1996 de Transferibilidad y Responsabilidad del Seguro de Salud (HIPAA), póngase en contacto con el jefe de la división de enfermedades genéticas: Chief of the Genetic Disease Branch, Dr. George C. Cunningham, M.P.H.; 850 Marina Bay Parkway; Richmond, CA 94804, (510) 412-1502.

羊膜穿刺術知情同意書／拒絕書

1. 我已得知羊膜穿刺術的目的是偵測胎兒的染色體異常、神經管缺陷和其他具體異常。
2. 我已得知在進行羊膜穿刺術之前要做一次超音波檢查，以確定胎盤和胎兒的位置。超音波也可以偵測雙胞胎、懷孕日期的錯誤推斷，以及其他某些狀況。
3. 我已得知羊膜穿刺術是將一根針從女性的腹壁穿入子宮中環繞胎兒的液體，抽出少量液體（少於一盎司）進行檢測。針頭刺入時可能會感到一些不適。
4. 我已經得知，根據目前已知的資料，羊膜穿刺術產生嚴重併發症的機率不到1%。這些併發症包括流產、出血、感染、胎膜過早破裂、或傷及胎兒或胎兒死亡。輕微併發症包括痙攣、陰道小量出血、或輕微羊水滲漏，以及穿刺部位感到疼痛。

_____ 我已經要求接受早期羊膜穿刺術（懷孕13週0天到14週6天）。我已經得知早期羊膜穿刺術可能比標準羊膜穿刺術（懷孕15週或以上）更容易導致妊娠失敗、羊水滲漏及（或）畸形足。

5. 我已經得知因為第一次採集的羊水不夠而需要重做羊膜穿刺術的比例不到百分之一。有時候即使採集了足夠的羊水，也無法做出診斷。
6. 我已經得知羊膜穿刺術可以辨識99%以上的染色體異常以及90%以上的開放性神經管缺陷。然而，並無法保證能對胎兒的狀況做出完整而確實的診斷。
7. 我已經得知羊膜穿刺術或超音波無法偵測所有的先天性缺陷。
8. 我已經得知在雙胞胎或三胞胎的情況下，也許只能獲得其中一個胎兒的檢測結果。
9. 我已經得知我會獲得所有異常結果的解釋，也會討論各種可能的治療方案。有關繼續或中斷妊娠的決定完全操之在我。
10. 我已經得知我參與這項程序純屬自願。拒絕這項程序不會使我無法接受由州政府資助的任何服務。
11. 我在下方簽名即表示：

我已經閱讀或由別人讀給我聽上述的資訊並了解其內容。我有機會和我的醫生或進行該程序的醫生做討論，包括羊膜穿刺術的目的和可能的風險。我已經收到所需的一切資料，而且我的問題都已獲得解答。

是	我要求 _____ 醫生及（或）同事進行羊膜穿刺術。 我了解並接受這項決定的後果。
	簽名 _____ 日期 _____
	見證人 _____ 日期 _____

否	我拒絕接受羊膜穿刺術。 我了解並接受這項決定的後果。
	簽名 _____ 日期 _____
	見證人 _____ 日期 _____

1977年「加州資訊執行政案」(民法 1798 et seq.) 規定我們要提供以下資料。加州衛生服務部遺傳疾病局 (California State Department of Health Services, Genetic Disease Branch) 會收到州政府核准的產前診斷中心關於所有異常測試結果的機密報告。這份資訊的收集係根據「加州法規」第 6531 和 6532 節的授權，目的是確保所有核准的產前診斷中心都符合州政府的服務標準，並改善先天性缺陷的偵測、預防及治療。

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經子宮頸或經腹部絨毛膜取樣

知情同意書／拒絕書

(TC CVS 或 TA CVS) > 上一次經期 (L.M.P.) 後 10 週或同等時間

1. 我已經得知絨毛膜取樣 (CVS) 的目的是偵測胎兒的染色體異數和其他具體異數。
2. 我已經得知可從另一種方法取得有關胎兒的染色體或生化狀態的診斷資訊。這項程序稱為羊膜穿刺術，通常在妊娠第二期進行，亦即上一次月經之後的15至18週或同等時間。方法是將一根針從腹壁穿入子宮中環繞胎兒的液體，抽出少量液體。我已獲得有關羊膜穿刺術與CVS之風險和好處的詳細解釋。
3. 我已經得知在進行CVS之前要做一次超音波檢查，以確定胎盤和胎兒的位置。超音波也可以偵測雙胞胎、懷孕日期的錯誤推斷，以及其他某些狀況。
4. 我已經得知經子宮頸絨毛膜取樣 (TC CVS) 的程序是將一根導管 (消毒的塑膠管) 藉由超音波導引插入女性的子宮頸，以採集胎盤組織。我已經得知經腹部絨毛膜取樣 (TA CVS) 的程序是將一根導管藉由超音波導引插入女性的腹部，以採集胎盤組織。
5. 我已經得知嚴重併發症的確切風險尚屬未知，但是據報告約為背景風險的200分之1到200分之6之間。最常見的嚴重併發症是流產。其他併發症包括出血、胎膜破裂、子宮穿孔，或感染。輕微併發症包括痙攣、陰道少量出血、或羊水輕微滲漏達兩週之久。
6. 某些研究顯示CVS可能涉及胎兒四肢異數的風險。然而，大部分的研究顯示在妊娠第10週或以上所做的程序極少增加風險。
7. 我已經得知CVS程序有時會因為第一次採集的組織不夠而需要重做。即使採集了足夠的組織，也可能無法做出診斷。然而，95%以上的案例都能取得足量的組織，並做出診斷評估。
8. 我已經得知在多重受孕 (雙胞胎、三胞胎等) 的情形下，可能無法順利從發展的胎兒週圍取得絨毛膜。在妊娠第二期可能需要做羊膜穿刺術。
9. 我已經得知CVS以及伴隨的超音波無法偵測所有的先天性缺陷，但是CVS可以識別99%以上的染色體異常。然而，並無法保證能對胎兒做出完整而確實的診斷。
10. 我已經得知為了澄清CVS的檢驗結果，可能要做羊膜穿刺術或胎兒採血。
11. 我得知我會獲得所有異常結果的解釋，也會討論各種可能的治療方案。有關繼續或中斷妊娠的決定完全操之在我。
12. 我已經得知在極少的機率下所取得的絨毛膜無法代表胎兒的遺傳基因，原因是染色體混嵌 (兩種或以上的細胞種類) 或母體細胞污染。
13. 我已經得知我參與這項程序純屬自願。我可以拒絕這項程序，但仍然獲得醫療照顧。
14. 我已經得知CVS程序無法偵測大部分的神經管缺陷。因此，我可以在上一次月經之後16-18週或同等時間內接受驗血，以篩檢神經管缺陷 (MS AFP測試)。

是	我要求 _____ 醫生及 (或) 同事進行CVS。 我了解並接受這項決定的後果。
	簽名 _____ 日期 _____
	見證人 _____ 日期 _____

否	我拒絕接受CVS。我了解並接受這項決定的後果。
	簽名 _____ 日期 _____
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1977年「加州資訊執行政案」(民法 1798 et seq.) 規定我們要提供以下資料。加州衛生服務部遺傳疾病局 (California State Department of Health Services, Genetic Disease Branch) 會收到州政府核准的產前診斷中心關於所有異常測試結果的機密報告。這份資訊的收集係根據「加州法規」第 6531 和 6532 節的授權，目的是確保所有核准的產前診斷中心都符合州政府的服務標準，並改善先天性缺陷的偵測、預防及治療。

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BẢN ỨNG THUẬN/TỪ CHỐI PHƯƠNG THỨC CHỌC RÚT NƯỚC ỒI SAU KHI HIỂU RÕ VẤN ĐỀ

- Tôi đã được cho biết mục đích của phương thức chọc rút nước ối là để thử nghiệm các chứng rối loạn nhiễm sắc thể thai nhi, các khuyết tật ống thần kinh, và các chứng rối loạn nhất định khác của thai nhi.
- Tôi đã được cho biết là trước khi chọc rút nước ối, tôi sẽ được rọi siêu âm để giúp biết được chỗ nhau thai và thai nhi. Siêu âm cũng có thể tìm ra song thai, tính ngày có thai sai, và một số tình trạng khác.
- Tôi đã được cho biết chọc rút nước ối là dùng kim chích vào bụng dưới của người phụ nữ vào chỗ nước ối bọc quanh thai nhi. Một ít nước ối (dưới một ounce) sẽ được rút ra để thử nghiệm. Khi chích kim vào bụng có thể gây cảm giác hơi khó chịu.
- Tôi đã được cho biết là dưới 1% những trường hợp chọc rút nước ối gây biến chứng nghiêm trọng, dựa theo dữ kiện hiện có. Những biến chứng này gồm xảy thai, xuất huyết, nhiễm trùng, vỡ bọc nước sớm, hoặc gây thương tích cho thai nhi hoặc làm thai nhi thiệt mạng. Những trường hợp biến chứng nhẹ gồm đau bụng, âm đạo ra máu lấm thẫm hoặc rỉ nước ối, và chỗ chích bị đau.

Tôi đã yêu cầu chọc rút nước ối sớm (13 tuần 0 ngày đến 14 tuần 6 ngày của thai kỳ). Tôi đã được cho biết là chọc rút nước ối sớm có thể gây rủi ro nhiều hơn trường hợp chọc rút nước ối tiêu chuẩn (vào hoặc sau 15 tuần của thai kỳ) làm xảy thai, rỉ nước ối, và/hoặc gây dị hình vẹo chân.

- Tôi đã được cho biết là chưa đến 1 trong 100 vụ chọc rút nước ối cần phải được thực hiện lại vì không rút đủ lượng nước ối lần đầu. Thành thạo, tuy đã rút đủ lượng nước ối nhưng vẫn không chẩn đoán được.
- Tôi đã được cho biết là chọc rút nước ối có thể tìm biết được hơn 99 phần trăm tất cả những chứng rối loạn nhiễm sắc thể và hơn 90 phần trăm tất cả những khuyết tật hở ống thần kinh. Tuy nhiên, không thể bảo đảm chẩn đoán được toàn bộ và đúng về tình trạng thai nhi.
- Tôi đã được cho biết là không thể phát hiện được tất cả các khuyết tật bẩm sinh bằng phương thức chọc rút nước ối hoặc siêu âm.
- Tôi đã được cho biết là trong trường hợp song hoặc tam thai, kết quả có thể là chỉ về một trong các thai nhi này.
- Tôi đã được cho biết là tôi sẽ được giải thích về tất cả các kết quả bất bình thường. Tôi sẽ được cho biết về những cách điều trị khác. Quyết định có tiếp tục hoặc chấm dứt thai là hoàn toàn do tôi quyết định.
- Tôi đã được cho biết là việc tôi tham gia vào phương thức này là hoàn toàn tự nguyện. Nếu tôi từ chối phương thức này thì cũng không phải vì thế mà khiến tôi không hội đủ điều kiện hưởng bất cứ dịch vụ nào được Tiểu Bang tài trợ.
- Chữ ký của tôi dưới đây chứng nhận rằng:

Tôi đã đọc, hoặc có người đọc cho tôi nghe, chi tiết trên đây và tôi hiểu rõ các chi tiết đó. Tôi đã có cơ hội thảo luận, gồm cả mục đích và các rủi ro có thể xảy ra của phương thức chọc rút nước ối, với bác sĩ của tôi và bác sĩ thực hiện phương thức này. Tôi đã nhận được tất cả các chi tiết tôi muốn có. Tôi đã được giải đáp tất cả mọi thắc mắc.

Có	Tôi YÊU CẦU Dr. _____ và/hoặc các đồng sự thực hiện phương thức chọc rút nước ối. Tôi hiểu và chấp nhận hậu quả của quyết định này. Ký tên _____ Ngày _____ Nhân chứng _____ Ngày _____
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Không	Tôi TỪ CHỐI phương thức chọc rút nước ối. Tôi hiểu và chấp nhận hậu quả của quyết định này. Ký tên _____ Ngày _____ Nhân chứng _____ Ngày _____
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Đạo Luật Thông Tin California Năm 1977 (Bộ Luật Dân Sự 1798 và kế tiếp.) đòi hỏi phải cung cấp chi tiết sau đây. California State Department of Health Services, Genetic Disease Branch (Bộ Y Tế California, Ngành Bệnh Di Truyền), sẽ nhận một phúc trình giữ kín về tất cả những kết quả thử nghiệm bất bình thường của các trung tâm chẩn đoán tiền sản được tiểu bang phê chuẩn. Chi tiết này được thu thập theo thẩm quyền của Bộ Luật Điều Lệ California Các Đoạn 6531 và 6532. Chi tiết này sẽ được xử dụng để bảo đảm là tất cả các trung tâm chẩn đoán tiền sản được phê chuẩn đáp ứng các tiêu chuẩn về dịch vụ và để cải tiến phát hiện, phòng ngừa, và điều trị các chứng khuyết tật bẩm sinh.

Bộ có thể liên lạc với quý vị để thu thập thêm chi tiết liên quan đến những cuộc nghiên cứu đặc biệt về khuyết tật bẩm sinh. Việc quý vị tham gia các cuộc nghiên cứu này là do tự nguyện và tất cả chi tiết do Bộ thu thập đều được tuyệt đối giữ kín và chi tiết cá nhân sẽ không được tiết lộ cho bất cứ người hoặc cơ quan nào nếu không có giấy cho phép của quý vị. Nếu quý vị có bất cứ thắc mắc, yêu cầu, hoặc khiếu nại nào về việc xử dụng chi tiết sức khỏe cá nhân của mình hoặc muốn có một bản Thông Báo về Cách Thông Tin và Tôn Trọng Quyền Riêng Tư theo đòi hỏi của Đạo Luật Năm 1996 về Tính Cách Lưu Động và Trách Nhiệm Bảo Hiểm Sức Khỏe (HIPAA), xin liên lạc với Chief of the Genetic Disease Branch (Trưởng Ngành Bệnh Di Truyền), George C. Cunningham, M.D., M.P.H.; 850 Marina Bay Parkway; Richmond, CA 94804, (510) 412-1502.

BẢN ỨNG THUẬN/TỪ CHỐI
PHƯƠNG THỨC CHORIONIC VILLUS SAMPLING QUA CỔ TỬ CUNG HOẶC QUA THÀNH BỤNG
(TC CVS HOẶC TA CVS) ≥ 10 tuần sau L.M.P. hoặc Tương Đương

1. Tôi đã được cho biết mục đích của phương thức Chorionic Villus Sampling là để phát hiện các chứng rối loạn nhiễm sắc thể thai nhi và các chứng rối loạn nhất định khác của thai nhi.
2. Tôi đã được cho biết có một phương pháp khác để thu thập dữ kiện chẩn đoán về tình trạng nhiễm sắc thể hoặc sinh hóa của thai nhi. Phương thức này, được gọi là chọc rút nước ối, thường được thực hiện trong tam cá nguyệt thứ nhì từ 15 đến 18 tuần kể từ kinh kỳ sau cùng của tôi hay tương đương. Phương thức này là dùng kim chích qua thành bụng vào nước ối bọc quanh thai nhi trong tử cung và rút ra một ít nước ối. Tôi đã được giải thích chi tiết về các rủi ro và lợi ích của phương thức chọc rút nước ối so với CVS.
3. Tôi đã được cho biết trước khi thực hiện CVS, tôi sẽ được rọi siêu âm để giúp tìm chỗ nhau thai và thai nhi. Siêu âm cũng có thể tìm ra song thai, tính ngày có thai sai, và một số tình trạng khác.
4. Tôi đã được cho biết TC CVS là dùng một ống luồn (ống plastic khử trùng) theo hình rọi siêu âm qua cổ tử cung của người phụ nữ để lấy tế bào nhau thai. Tôi đã được cho biết là TA CVS là dùng kim chích theo hình rọi siêu âm qua qua bụng dưới của người phụ nữ để lấy tế bào nhau thai.
5. Tôi đã được cho biết là không biết chính xác về rủi ro đưa đến biến chứng nghiêm trọng nhưng được biết là trong 200 vụ thì có từ 1 đến 6 bị rủi ro hơn mức cơ bản. Biến chứng nghiêm trọng nhất là xảy thai. Các biến chứng khác là xuất huyết, vỡ bọc nước, lủng tử cung, hoặc nhiễm trùng. Các biến chứng nhẹ gồm đau bụng, âm đạo ra máu lấm thấm, hoặc rỉ chút ít nước ối đến tối đa là 2 tuần.
6. Một số cuộc nghiên cứu cho thấy là CVS có thể gây ra rủi ro thai nhi bị chân tay dị hình. Tuy nhiên, đa số các cuộc nghiên cứu xem ra đều cho thấy là nếu thực hiện phương thức này sau khi có thai được 10 tuần thì hầu như không tăng thêm rủi ro nào.
7. Tôi đã được cho biết thỉnh thoảng có thể cần phải thực hiện lại CVS vì không lấy đủ tế bào trong lần đầu. Cũng có thể không chẩn đoán được dù đã lấy đủ tế bào. Tuy nhiên, trong hơn 95% mọi trường hợp, có thể lấy đủ tế bào và đánh giá chẩn đoán.
8. Tôi đã được cho biết là nếu mang đa thai (song thai, tam thai), các nỗ lực lấy chorionic villi xung quanh thai nhi đang phát triển có thể không thành công. Có thể cần phải chọc rút nước ối trong tam cá nguyệt thứ nhì của thai kỳ.
9. Tôi đã được cho biết là CVS và rọi siêu âm kèm theo không thể phát hiện được những trường hợp khuyết tật bẩm sinh, nhưng CVS có thể tìm ra hơn 99% tất cả những chứng rối loạn nhiễm sắc thể. Tuy nhiên, không thể bảo đảm chẩn đoán được toàn bộ và đúng về tình trạng thai nhi.
10. Tôi đã được cho biết là tôi có thể được đề nghị chọc rút nước ối hoặc lấy mẫu máu thai nhi để hiểu rõ hơn về kết quả CVS.
11. Tôi đã được cho biết là tôi sẽ được giải thích về tất cả các kết quả bất bình thường. Tôi sẽ được cho biết về những cách điều trị khác. Quyết định có tiếp tục hoặc chấm dứt thai là hoàn toàn do tôi quyết định.
12. Tôi đã được cho biết là mẫu chorionic villi thu thập có thể không tiêu biểu được thành phần di truyền của thai nhi dù rủi ro này rất thấp, vì lẫn nhiều nhiễm sắc thể (2 loại tế bào trở lên) hoặc bị nhiễm tế bào người mẹ.
13. Tôi đã được cho biết là việc tôi tham gia vào phương thức này là hoàn toàn tự nguyện. Tôi có thể từ chối phương thức này và vẫn được chăm sóc y tế.
14. Tôi đã được cho biết là phương thức CVS không phát hiện được đa số các khiếm khuyết ống thần kinh. Do đó tôi sẽ được đề nghị thử máu để dò tìm khiếm khuyết ống thần kinh (thử nghiệm MS AFP) vào 16-18 tuần kể từ kinh kỳ sau cùng của tôi hoặc tương đương.

Có	<p>Tôi YÊU CẦU Dr. _____ và/hoặc các đồng sự thực hiện phương thức CVS. Tôi hiểu và chấp nhận hậu quả của quyết định này.</p> <p>Ký tên _____ Ngày _____</p> <p>Nhân chứng _____ Ngày _____</p>
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Không	<p>Tôi TỪ CHỐI phương thức CVS. Tôi hiểu và chấp nhận hậu quả của quyết định này.</p> <p>Ký tên _____ Ngày _____</p> <p>Nhân chứng _____ Ngày _____</p>
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Đạo Luật Thông Tin California Năm 1977 (Bộ Luật Dân Sự 1798 và kế tiếp.) đòi hỏi phải cung cấp chi tiết sau đây. California State Department of Health Services, Genetic Disease Branch (Bộ Y Tế California, Ngành Bệnh Di Truyền), sẽ nhận một phúc trình giữ kín về tất cả những kết quả thử nghiệm bất bình thường của các trung tâm chẩn đoán tiền sản được tiểu bang phê chuẩn. Chi tiết này được thu thập theo thẩm quyền của Bộ Luật Điều Lệ California Các Đoan 6531 và 6532. Chi tiết này sẽ được sử dụng để bảo đảm là tất cả các trung tâm chẩn đoán tiền sản được phê chuẩn đáp ứng các tiêu chuẩn về dịch vụ và để cải tiến phát hiện, phòng ngừa, và điều trị các chứng khuyết tật bẩm sinh.

Bộ có thể liên lạc với quý vị để thu thập thêm chi tiết liên quan đến những cuộc nghiên cứu đặc biệt về khuyết tật bẩm sinh. Việc quý vị tham gia các cuộc nghiên cứu này là do tự nguyện và tất cả chi tiết do Bộ thu thập đều được tuyệt đối giữ kín và chi tiết cá nhân sẽ không được tiết lộ cho bất cứ người hoặc cơ quan nào nếu không có giấy cho phép của quý vị. Nếu quý vị có bất cứ thắc mắc, yêu cầu, hoặc khiếu nại nào về việc sử dụng chi tiết sức khỏe cá nhân của mình hoặc muốn có một bản Thông Báo về Cách Thông Tin và Tôn Trọng Quyền Riêng Tư theo đòi hỏi của Đạo Luật Năm 1996 về Tính Cách Lưu Động và Trách Nhiệm Bảo Hiểm Sức Khỏe (HIPAA), xin liên lạc với Chief of the Genetic Disease Branch (Trưởng Ngành Bệnh Di Truyền), George C. Cunningham, M.D., M.P.H.; 850 Marina Bay Parkway; Richmond, CA 94804, (510) 412-1502.

ULTRASOUND FINDINGS

<input type="checkbox"/> No Abnormality on Ultrasound for the fetus/Pregnancy	<input type="checkbox"/> Conjoined Twins
<input type="checkbox"/> Molar Pregnancy	<input type="checkbox"/> Twin-Twin Transfusion
<input type="checkbox"/> Pregnancy Not Detected	<input type="checkbox"/> Acardiac Twin
<input type="checkbox"/> Fetal Demise < 20 weeks	<input type="checkbox"/> Monoamniotic/Monochorionic Twins
<input type="checkbox"/> Fetal Demise > or = 20 weeks	<input type="checkbox"/> Other Ultrasound Abnormality(ies) check below
CENTRAL NERVOUS SYSTEM	HEART/LUNG
<input type="checkbox"/> Acrania	<input type="checkbox"/> Pleural Effusion
<input type="checkbox"/> Agenesis of Corpus Callosum (Absent Calvarium)	<input type="checkbox"/> Small Chest
<input type="checkbox"/> Anencephaly	<input type="checkbox"/> Cystic Adenomatous Malformation of the Lung (CAML)
<input type="checkbox"/> Arnold Chiari Malformation	<input type="checkbox"/> Chest Mass (CCAM, Sequestration)
<input type="checkbox"/> Banana sign	<input type="checkbox"/> Cardiac Defect- Abnormal Outflow Tracts
<input type="checkbox"/> Cerebellar Hypoplasia	<input type="checkbox"/> Cardiac Defect- Atrial Septal Defect (ASD)
<input type="checkbox"/> Choroid Plexus Cyst(s)	<input type="checkbox"/> Cardiac Defect- Cardiomegaly
<input type="checkbox"/> Craniorachischisis	<input type="checkbox"/> Cardiac Defect- Coarctation of Aorta
<input type="checkbox"/> Dandy Walker	<input type="checkbox"/> Cardiac Defect- Dextrocardia/Heart on Right Side
<input type="checkbox"/> Dolichocephaly	<input type="checkbox"/> Cardiac Defect- Echogenic Intracardiac Foci (EIF)
<input type="checkbox"/> Encephalocele	<input type="checkbox"/> Cardiac Defect- Endocardial Cushion Defect/A-V canal
<input type="checkbox"/> Enlarged Atrium	<input type="checkbox"/> Cardiac Defect- Enlarged Atrium
<input type="checkbox"/> Exencephaly	<input type="checkbox"/> Cardiac Defect- Ebstein Anomaly
<input type="checkbox"/> Holoprosencephaly	<input type="checkbox"/> Cardiac Defect- Hypoplastic Left Heart
<input type="checkbox"/> Hydrocephalus NOT with spina bifida (unknown etiology)	<input type="checkbox"/> Cardiac Defect- Pericardial Effusion
<input type="checkbox"/> Iniencephaly	<input type="checkbox"/> Cardiac Defect- Tetralogy of Fallot
<input type="checkbox"/> Lemon sign	<input type="checkbox"/> Cardiac Defect- Transposition of the Great Vessels
<input type="checkbox"/> Meckel Gruber	<input type="checkbox"/> Cardiac Defect- Tricuspid Regurgitation
<input type="checkbox"/> Mega Cisterna Magna	<input type="checkbox"/> Cardiac Defect- Ventricular Septal Defect (VSD)
<input type="checkbox"/> Myelomeningocele	<input type="checkbox"/> Other Heart Defect (specify in OTHER section)
<input type="checkbox"/> Pentalogy of Cantrell	
<input type="checkbox"/> Porencephaly	ABDOMEN
<input type="checkbox"/> Rachischisis	<input type="checkbox"/> Amniotic Band syndrome
<input type="checkbox"/> Spina Bifida	<input type="checkbox"/> Ascites/Anasarca/Edema
<input type="checkbox"/> Spina Bifida with hydrocephalus	<input type="checkbox"/> Diaphragmatic Hernia
<input type="checkbox"/> Ventriculomegaly	<input type="checkbox"/> Double Bubble
<input type="checkbox"/> Neural Tube Defect- unspecified	<input type="checkbox"/> Duodenal Obstruction
<input type="checkbox"/> Other Brain Anomaly (specify in OTHER section)	<input type="checkbox"/> Echogenic Bowel
	<input type="checkbox"/> Gastroschisis
FACE	<input type="checkbox"/> Limb Body Wall Defect
<input type="checkbox"/> Cleft Lip	<input type="checkbox"/> Omphalocele
<input type="checkbox"/> Cleft Palate	<input type="checkbox"/> Unspecified Abdominal Wall Defect
<input type="checkbox"/> Cleft Lip & Palate	<input type="checkbox"/> Peritoneal Calcifications
<input type="checkbox"/> Flat Nasal Bridge	<input type="checkbox"/> Absent Stomach
<input type="checkbox"/> Micrognathia	<input type="checkbox"/> Small Stomach
	<input type="checkbox"/> Dilated Stomach
	<input type="checkbox"/> Other Abdominal Problem (specify in OTHER section)
NECK	
<input type="checkbox"/> Cystic Hygroma	SKELETAL SYSTEM
<input type="checkbox"/> Nuchal Fold (> 5 - < 6 mm)	<input type="checkbox"/> Caudal Regression <input type="checkbox"/> Severe Scoliosis
<input type="checkbox"/> Nuchal Fold (> or = 6 - < 7 mm)	<input type="checkbox"/> Dwarf
<input type="checkbox"/> Nuchal Fold (> or = 7 - < 8 mm)	<input type="checkbox"/> Other Skeletal System Problem (specify in OTHER section)
<input type="checkbox"/> Nuchal Fold (> or = 8 - < 9 mm)	<input type="checkbox"/> Upper Extremity- Abnormal position or number of bones
<input type="checkbox"/> Nuchal Fold (> or = 9 - < 10 mm)	<input type="checkbox"/> Upper Extremity- Absent bones
<input type="checkbox"/> Nuchal Fold (> 10 mm)	<input type="checkbox"/> Upper Extremity- Arthrogryposis Multiplex Congenita
SIZE/GROWTH/OVERALL APPEARANCE	<input type="checkbox"/> Upper Extremity- Clenched hands
<input type="checkbox"/> Hydrops fetalis	<input type="checkbox"/> Upper Extremity-Clinodactyly
<input type="checkbox"/> Intrauterine Growth Retardation (IUGR)	<input type="checkbox"/> Upper Extremity- Polydactyly
<input type="checkbox"/> Generalized Edema	<input type="checkbox"/> Lower Extremity- Abnormal position or number of bones
<input type="checkbox"/> Macrosomia	<input type="checkbox"/> Lower Extremity- Unilateral Clubfoot
<input type="checkbox"/> Multiple Congenital Anomalies (MCA)	<input type="checkbox"/> Lower Extremity- Bilateral Clubfeet
	<input type="checkbox"/> Lower Extremity- Short Femurs
AMNIOTIC FLUID VOLUME	
<input type="checkbox"/> Mild/Moderate Oligohydramnios	KIDNEY/URINARY BLADDER/PELVIS
<input type="checkbox"/> Severe Oligohydramnios	<input type="checkbox"/> Absent Kidney- Unilateral
<input type="checkbox"/> Mild/Moderate Polyhydramnios	<input type="checkbox"/> Absent Kidney- Bilateral
<input type="checkbox"/> Severe Polyhydramnios	<input type="checkbox"/> Potter's syndrome
	<input type="checkbox"/> Echogenic Kidneys
UMBILICAL CORD	<input type="checkbox"/> Enlarged Kidneys
<input type="checkbox"/> 2 Vessel Cord/Single Umbilical Artery	<input type="checkbox"/> Hydronephrosis
<input type="checkbox"/> Velamentous Insertion	<input type="checkbox"/> Multicystic Dysplastic Kidneys
<input type="checkbox"/> Very Curled	<input type="checkbox"/> Renal Cysts
	<input type="checkbox"/> Absent Bladder
PLACENTA	<input type="checkbox"/> Empty Urinary Bladder
<input type="checkbox"/> Chorioangioma <input type="checkbox"/> Placental Abruption	<input type="checkbox"/> Dilated Urinary Bladder
<input type="checkbox"/> Chorion Separation <input type="checkbox"/> Placental Lakes	<input type="checkbox"/> Pelvic Cysts
<input type="checkbox"/> Fibroids in Placenta <input type="checkbox"/> Placenta Previa	<input type="checkbox"/> Pyelectasis
<input type="checkbox"/> Hematoma in Placenta <input type="checkbox"/> Thickened Placenta	<input type="checkbox"/> Other kidney/bladder problem (specify in OTHER section)
<input type="checkbox"/> Other Placental problem (specify in OTHER section)	

California Department of Public Health
Genetic Disease Screening Program
California Prenatal Screening Program



850 Marina Bay Parkway, Richmond, CA 94804 • Phone (510) 412-1502 • Fax (510) 412-1547

TO: Prenatal Screening Coordinator at CCC _____

Fax# _____

FROM: PDC# _____

Fax# _____ Phone# _____

Genetic Counselor: _____

RE: Patient: _____

Accn# _____

This patient was seen on ____/____/20____ for genetic counseling only. Her result was First Trimester Combined Screen Positive.

She has elected:

CVS Provided/scheduled on _____
 To be scheduled at PDC _____ (name) _____ (PDC#)

Amniocentesis Scheduled on _____

To proceed with Second Trimester Blood Draw for Full Integrated Screening

Undecided

Declined all further services

Notes:

CLINICAL GENETICIST CHART REVIEW

(Within 30 days of the final date of service)

Patient's Name _____

Patient MR# _____

Not Applicable Reviewed

- Family History Assessment (Pedigree and/or Questionnaire) – Document reviewed and dated by a genetic counselor and appropriate follow-up of a positive family history
- Expanded AFP Interpretation factors – Confirmed with patient
- Invasive prenatal diagnostic procedure consent/decline form – Consent signed by the patient or documentation of decline on the form or in the chart
- Preliminary ultrasound findings

Signature of Clinical Geneticist

Date

Not Applicable Reviewed

- Complete ultrasound report – Signed by consultative sonologist
- All laboratory results – e.g.: karyotype, AFAFP, DNA
- Documentation of all significant clinical information and results to the referring physician

Signature of Clinical Geneticist
or genetic counselor (if all results are normal)

Date

Reviewed

- Document appropriate follow-up regarding abnormal results

Signature of Clinical Geneticist

Date

ELEMENTS NEEDED IN ELECTRONIC CHART-DRAFT 021408

TYPE OF INFORMATION	CURRENT PAPER CHART	ELECTRONIC VERSION (EMR)	FUTURE TREND
	Information required in the paper chart (currently)-for GDSP monitoring	Required Information - letters/reports scanned into EMR or required elements present in paperless system	
Demographic	Patient Name	Patient Name	
	DOB (Date of Birth)	DOB (Date of Birth)	
	Patient Contact Information (address, telephone # (s))	Patient Contact Information (address, telephone # (s))	
	Date when appt. made	Date of contact (date when appt. made)	
	Patient identifier from the referral source (optional)	Patient identifier from the referral source (optional)	
Referring Physician (OB)	Name	Name	
	Contact Information (address, telephone # (s))	Contact Information (address, telephone # (s))	
Billing/Insurance Info	Although not required for GDSP monitoring, it is required for billing; should include patient Prenatal Screening referral, third party billing etc.	Although not required for GDSP monitoring, it is required for billing; should include patient Prenatal Screening referral, third party billing etc.	
Information from Date of Service	Date of Service	Date of Service	
	Site of Service	Site of Service	
	Gestational Age	Gestational Age (also available from scanned utz report)	
	Number of fetuses	Number of fetuses (also available from scanned utz report)	
	Referral Indication	Referral Indication	
	Ethnicity Information for biological parents	Ethnicity Information for biological parents (also available from scanned pedigree)	
	Questionnaire completed by patient / GC	Questionnaire completed by patient / GC	
	Pedigree completed by GC GC Letter (copy of the letter generated and mailed to physician)	Pedigree completed by GC GC Letter	
Prenatal Screening Pts	AFP Printout; Recal Printout if indicated; Indication that interpretation factors were verified by GC (could be an initial of the GC or notification in the chart)	Prenatal Screening Printout; Recalc Printout if indicated; Indication that interpretation factors were verified by GC	(GDSP should be able to send AFP results/authorization or printout electronically to the various EMR via interfaces; the printouts should be concise and user friendly so that information can be viewed on one screen rather than scrolling down 4-5 pages. In addition, Recal results/authorization or printouts should specify RECAL and authorization status and indicate the reason for RECAL)
Procedure Information	Ultrasound Report	Ultrasound Info: Date of ultrasound, Gestational age information, number of fetuses, referral indication, Anatomy Information Normal Vs Abnormal; if abnormal need to know the details. Procedure done CVS / Amnio or declined or rescheduled. Report will need some form of signature, e.g. scanature, electronically signed etc.	Interfaces between GC system and the ultrasound reporting system, with transfer of ultrasound report data directly into GC system (rather than having to scan the document)
	Procedure consent, if procedure performed	Procedure consent, if procedure done	Consent could be available on the paperless system and patient could directly sign the PDA form
Lab / Medical Record Information	Results of CVS / Amnio, if indicated	Results of CVS / Amnio, if indicated	Interfaces between GC system and the lab system with transfer of results directly into GC system
	Other lab results, if indicated-e.g. Blood type, other medical records	Other lab results, if indicated-e.g. Blood type, other medical records	
	Follow-up for abnormal results, if indicated	Follow-up of abnormal results, if indicated	
Result Reporting	Documentation of result reporting to patient and/or physician	Documentation of result reporting to patient and/or physician	
	Documentation re. follow-up for abnormal prenatal result, if indicated	Documentation re. follow-up for abnormal prenatal result, if indicated	
Clinical Geneticist Chart Review	Documentation of GC chart reviewed by clinical geneticist	Documentation of GC chart reviewed by clinical geneticist	

Services Covered by the Prenatal Screening Program

The California Prenatal Screening Program is a voluntary program involving a blood test and, depending upon the results, follow-up diagnostic services. Some of these services are covered by the Prenatal Screening Program and involve no additional cost to you.

These include:

1. Genetic Risk Assessment by a state-approved Genetic Counselor
2. Ultrasound* (if indicated)
3. Amniocentesis* (if indicated) and testing of the amniotic fluid for:
 - chromosome problems like Down syndrome and trisomy 18
 - neural tube defects like spina bifida (open spine)
 - Smith-Lemli-Opitz syndrome, a rare birth defect causing mental retardation (only if indicated).

Although other testing may be indicated based upon your family and medical history or your ultrasound study, these additional tests are not covered by the Program. These services can be billed to your health insurance or to you directly. Your financial responsibility is determined by your insurance company and you should check your policy to determine coverage of any additional services.

Examples of services NOT covered by the Prenatal Screening Program include:

1. Rhogam injection following amniocentesis, if your blood type is Rh negative
2. Follow-up ultrasound to monitor the growth of the fetus
3. Special ultrasound of the baby's heart (fetal echocardiography)
4. Genetic blood tests on you or your partner
5. Consultation with other specialists
6. Other testing of the amniotic fluid, such as FISH (a rapid chromosome test) or DNA analysis for viral exposures or genetic conditions.

I have read the above information about follow-up services that are covered by the Prenatal Screening Program and understand that I will be financially responsible for any additional services.

I understand that my participation in the Program is voluntary and that I can decline any services.

Patient's signature: _____ Date: _____

Patient's Name: _____

* In order for the ultrasound and amniocentesis to be covered by the Prenatal Screening Program, you must meet with the genetic counselor.

CQI Chart Monitoring Tally Sheet

Date of Review: _____ Reviewer: _____

PDC/Satellite Site: _____

Total Number of Charts Reviewed: _____

Chart Monitor	Is the documentation in the chart?		
	threshold goal	number	percent
For all PDC Patients:			
1. Documentation of genetic counseling and clinical/history risk assessment (questionnaire and/or pedigree)	95%		
2. Documentation of a signed consent/decline for an invasive prenatal diagnostic procedure	95%		
3. Documentation of a signed ultrasound report	95%		
4. Documentation of a signed copy of correspondence with the referring physician/clinic	95%		
5. Documentation of karyotype and AFAFP/AChE results	95%		
6. Documentation of communication of genetic screening or diagnostic test results to the patient and/or referring physician/clinic	95%		
7. Documentation of the signed clinical geneticist chart review within 30 days of the last day of service	95%		
8. Documentation of the offer of clinical geneticist involvement in abnormal cases	95%		
For Prenatal Screening patients only:			
1. The time from initial contact to the Prenatal screening appointment is within 7 days or a documented explanation for the reason for the delay	95%		
2. Documentation of confirming the Prenatal Screening interpretation factors with the patient	95%		
3. Written documentation of communication with the Case Coordinator	95%		

Comments: _____

PDC Director: _____ Date of Review _____

**CQI Chart Monitoring
Corrective Action Report**

Date: _____ Reviewer: _____

PDC Staff Being Reviewed: _____

Monitor(s) Reviewed: _____

Threshold Value: 95 % Measured Value: _____

Corrective Action: _____

Follow-up of Corrective Action: _____

Date of Follow-up: _____

Signatures:

Follow-up Reviewer: _____	Date: _____
PDC Staff Being Reviewed: _____	Date: _____
PDC Director: _____	Date: _____

Mother's or Infant's Name (in case of page separation):

HOSPITAL INFORMATION

37. NAME AND ADDRESS OF BIRTH HOSPITAL:
38. MOTHER'S MEDICAL RECORD AT BIRTH HOSPITAL:
39. INFANT'S MEDICAL RECORD AT BIRTH HOSPITAL:
40. NAME AND ADDRESS OF TRANSFER HOSPITAL:
41. INFANT'S MEDICAL RECORD AT TRANSFER HOSPITAL:
42. DATE OF TRANSFER TO TRANSFER HOSPITAL:

PHYSICIAN INFORMATION

43. NAME AND ADDRESS OF MOTHER'S PHYSICIAN:
44. TELEPHONE NUMBER OF MOTHER'S PHYSICIAN:
45. NAME AND ADDRESS OF INFANT'S PHYSICIAN:
46. TELEPHONE NUMBER OF INFANT'S PHYSICIAN:

BIRTH DEFECT DIAGNOSIS

A. CHROMOSOMAL ABNORMALITIES

Do not report: a) Heterochromatin Variants; b) Satellite/Stalk Variants of Chromosomes 13,14,15,21,22; c) Inv(2) (p11;q13); d) Inv (9) (p11;q12 or q13); e) Familial Y Variants; or d) Pseudomosaics.

47. CYTOGENETIC DIAGNOSIS (ISCN Short Form)

48. INHERITANCE OF STRUCTURAL REARRANGEMENTS: DE NOVO, PATERNAL, MATERNAL, UNKNOWN
49. CYTOGENETIC LAB SPECIMEN NUMBER:
50. LAB NAME:
51. SPECIMEN TYPE: STILLBORN TISSUE/BLOOD, AMNIOTIC FLUID, LIVERBORN BLOOD, CHORIONIC VILLUS (CVS), LIVERBORN TISSUE, PERCUTANEOUS UMBILICAL BLOO, LIVERBORN CORD BLOOD, ABORTUS SPECIMEN, LIVERBORN BONE MARROW, OTHER Specify:

52. SAMPLING DATE Check if final result date used
53. GESTATIONAL AGE AT TIME OF SAMPLING: (Prenatal: Abortus, Fetal Demise or Stillborn only)
54. METHOD USED TO DETERMINE GESTATIONAL AGE AT TIME OF SAMPLING: LMP, ULTRASOUND, EXAM

55. REASON FOR SAMPLE: DYSMORPHIC FEATURES, CONGENITAL ABNORMALITIES, CONFIRMATION OF PRENATAL DIAGNOSIS, OTHER

B. NEURAL TUBE DEFECTS

56. NEURAL TUBE DEFECT DIAGNOSIS: (ICD 9 Codes 740.0-742.0) ANENCEPHALY, ACRANIA, SPINA BIFIDA, RACHISCHISIS, EXENCEPHALY, SPINA BIFIDA, CRANIORACHISCHISIS, MECKEL GRUBER, with HYDROCEPHALUS, INIENCEPHALY, UNSPECIFIED, Spina Bifida includes Meningocele, Myelomeningocele, Myelomeningocele, ENCEPHALOCELE, OTHER Specify below: and Lipmenigocele, MYELOMENINGOCELE
57. SPINA BIFIDA TYPE: OPEN, CLOSED, UNSPECIFIED
58. IS HYDROCEPHALY PRESENT? YES, NO, UNKNOWN
59. NTD PART OF A SYNDROME?
60. IF YES, SPECIFY SYNDROME:
61. ARE OTHER ABNORMALITIES PRESENT? Specify:

PRENATAL GENETIC PROCEDURES PERFORMED TO DETECT NEURAL TUBE DEFECT

62. ULTRASOUND: PRENATAL PROCEDURE, DATE PROCEDURE PERFORMED (MM/DD/YYYY), NAME & ADDRESS OF FACILITY or PDC CODE WHERE PROCEDURE WAS PERFORMED, GESTATIONAL AGE OF FETUS IN WEEKS/DAYS, DID PROCEDURE DETECT NTD? YES, NO
63. AMNIOCENTESIS: PRENATAL PROCEDURE, DATE PROCEDURE PERFORMED (MM/DD/YYYY), NAME & ADDRESS OF FACILITY or PDC CODE WHERE PROCEDURE WAS PERFORMED, GESTATIONAL AGE OF FETUS IN WEEKS/DAYS, DID PROCEDURE DETECT NTD? YES, NO
64. IF AMNIO PERFORMED, PROVIDE THE AF-AFP LEVEL (in M.o.M.)
65. AF-AChE RESULT: POSITIVE, NEGATIVE, NOT PERFORMED
66. IF NTD WAS DIAGNOSED POSTNATALLY, WHEN DIAGNOSED? AT THE TIME OF LIVE BIRTH, AT PHYSICAL EXAMINATIO, AT TIME OF STILLBIRTH, OTHER Specify Other:
67. DATE OF POSTNATAL DIAGNOSIS:

68. WAS THE FETAL ABNORMALITY CONFIRMED? YES, NO, PENDING, NOT POSSIBLE, UNKNOWN
69. METHOD USED FOR CONFIRMATION OF FETAL ABNORMALITY: AUTOPSY / PATHOLOGY, VISUAL EXAM, OTHER Specify: ULTRASOUND ONLY, ULTRASOUND & AMNIO
70. WHAT WAS THE SOURCE OF SOURCES OF YOUR CONFIRMATION? ULTRASOUND RPT, AMNIO REPORT, CLINICIAN NOTES, UNKNOWN, AUTOPSY / PATH REPORT, DELIVERY ROOM REPORT, OUTCOME OF PREGNANCY, OTHER Specify:

PRIVACY STATEMENT The Information Practices Act of 1977 (Civil Code 1798 et. seq.) requires that the following details be provided when a form is used to obtain information from individuals. The data requested in this form are required by the Genetic Disease Screening Program (GDSP) of the California Department of Public Health and are mandated by C.C.R.17 Section 6532. These data are used to provide information to subjects on the prevention of birth defects, to determine prevalence of chromosomal and neural tube defects and to monitor trends of occurrence. They will also be used to determine the effectiveness of the California Expanded Alpha Fetoprotein Screening Program. It is mandatory that health professionals completing this form provide complete and accurate information. The records maintained by the GDSP are confidential as defined in Civil Code 1798.34 and are exempt from access by any individual except licensed medical personnel designated by the subject. The information may also be used in special studies as defined in Health and Safety Code 100330. The furnishing of such information to the Department or its authorized representative, or any other cooperating individual, agency or organization in any such special study shall not subject any person, hospital or other organization furnishing such information to any actions or damages.

OUTCOME OF INVASIVE PRENATAL DIAGNOSTIC PROCEDURES

INSTRUCTIONS: PLEASE COMPLETE THIS FORM AND RETURN IT TO THE CALIFORNIA DEPARTMENT OF PUBLIC HEALTH, GENETIC DISEASE SCREENING PROGRAM, 850 MARINA BAY PARKWAY, F-175, MAIL STOP 8200, RICHMOND, CA 94804 OR FAX TO: 510/412-1551. CALL STEPHEN PURSER AT 510/412-1491 IF YOU HAVE ANY QUESTIONS OR CONCERNS.

STUDY DESIGN This form is designed to collect the outcome of consecutive prenatal diagnostic procedures. Include all procedures performed in consecutive order (excluding elective terminations). You may include Expanded AFP patients. Data collection must be continuous until the appropriate level of procedures and responses is obtained. Provisional practitioners must achieve at least a 90% or greater follow-up rate. All other practitioners must achieve at least an 80% or greater follow-up rate. The greater the response rate you obtain, the smaller the sample size you need to complete study (see table below). Practitioners who work at multiple centers must notify the Genetic Disease Screening Program in advance of the location(s) of their study and must include all procedures at all sites.

REPORTING REQUIREMENTS For practitioners who perform less than 100 amniocenteses per year or less than fifty (50) CVS procedures per year, we will ask for a progress report every six months until the study is complete. The start date for the study for new practitioners will be the first day of the first completed study, the next data collection period will be scheduled to begin five years after the start date of the previous study.

TO ACHIEVE A FOLLOW-UP RATE OF:	# OF STANDARD AMNIOCENTESES PROCEDURES REQUIRED	NUMBER OF KNOWN OUTCOME	# OF CHORIONIC VILLUS SAMPLE PROCEDURES REQUIRED	NUMBER OF KNOWN OUTCOME	# OF EARLY AMNIOCENTESES PROCEDURES REQUIRED	NUMBER OF KNOWN OUTCOME
100%	100	100	50	50	60	60
99%	105	104	54	53	64	63
98%	111	108	58	57	68	67
97%	116	113	62	61	73	71
96%	123	118	67	65	78	75
95%	129	123	73	69	84	80
94%	136	128	78	74	90	84
93%	144	134	85	79	96	90
92%	152	140	92	84	103	95
91%	160	146	99	90	111	101
90%	169	152	108	97	119	107
89%	179	159	117	104	128	114
88%	189	167	127	112	138	122
87%	200	174	138	120	149	129
86%	212	183	150	129	160	138
85%	225	191	163	139	173	147
84%	239	201	178	150	187	157
83%	254	210	194	161	202	168
82%	269	221	212	174	219	179
81%	286	232	232	188	237	192
80%	305	244	254	203	257	206

PLEASE REPORT OUTCOMES ON REVERSE OF THIS FORM.

OUTCOME OF INVASIVE PRENATAL DIAGNOSTIC PROCEDURES

INSTRUCTIONS: PLEASE COMPLETE THIS FORM AND RETURN IT TO THE CALIFORNIA DEPARTMENT OF PUBLIC HEALTH, GENETIC DISEASE SCREENING PROGRAM, 850 MARINA BAY PARKWAY, F-175, MAIL STOP 8200, RICHMOND, CA 94804 OR FAX TO: 510/412-1551. CALL STEPHEN PURSER AT 510/412-1491 IF YOU HAVE ANY QUESTIONS OR CONCERNS.

STUDY DESIGN This form is designed to collect the outcome of consecutive prenatal diagnostic procedures. Include all procedures performed in consecutive order (excluding elective terminations). You may include Expanded AFP patients. Data collection must be continuous until the appropriate level of procedures and responses is obtained. Provisional practitioners must achieve at least a 90% or greater follow-up rate. All other practitioners must achieve at least an 80% or greater follow-up rate. The greater the response rate you obtain, the smaller the sample size you need to complete study (see table below). Practitioners who work at multiple centers must notify the Genetic Disease Screening Program in advance of the location(s) of their study and must include all procedures at all sites.

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TO ACHIEVE A FOLLOW-UP RATE OF:	# OF STANDARD AMNIOCENTESES PROCEDURES REQUIRED	NUMBER OF KNOWN OUTCOME	# OF CHORIONIC VILLUS SAMPLE PROCEDURES REQUIRED	NUMBER OF KNOWN OUTCOME	# OF EARLY AMNIOCENTESES PROCEDURES REQUIRED	NUMBER OF KNOWN OUTCOME
100%	100	100	50	50	60	60
99%	105	104	54	53	64	63
98%	111	108	58	57	68	67
97%	116	113	62	61	73	71
96%	123	118	67	65	78	75
95%	129	123	73	69	84	80
94%	136	128	78	74	90	84
93%	144	134	85	79	96	90
92%	152	140	92	84	103	95
91%	160	146	99	90	111	101
90%	169	152	108	97	119	107
89%	179	159	117	104	128	114
88%	189	167	127	112	138	122
87%	200	174	138	120	149	129
86%	212	183	150	129	160	138
85%	225	191	163	139	173	147
84%	239	201	178	150	187	157
83%	254	210	194	161	202	168
82%	269	221	212	174	219	179
81%	286	232	232	188	237	192
80%	305	244	254	203	257	206

PLEASE REPORT OUTCOMES ON REVERSE OF THIS FORM.

Appendix B: Program Data

Reference

1. Program Expansion Summary Grid
2. Down Syndrome Detection rates and *Screen Positive* rates
3. Trisomy 18 Detection rates and *Screen Positive* rates
4. Mid-trimester risk for chromosome abnormalities
5. Communicating risks
6. General release (2007)
7. PDC Flow (2007)
8. Quarterly Report statistics (2007)
9. Information available on GDSP website
10. PNS supplies

PNS Program Expansion Summary

www.cdph.ca.gov/PROGRAMS/pns

This summary is for GDSP and it's contractors/vendors. Do not distribute to general public.

Legend:

To use CRL for dating: Exam=7w1d - 14w2d (10 - 84 mm)
 To use NT for Interp: NT exam=11w2d - 14w2d (CRL 45 – 84mm)
 1st T blood draw: 10w0d - 13w6d
 2nd T blood draw: 15w0d - 20w0d

First T analytes: PAPP-A, hCG
 Second T analytes: AFP, hCG, uE3, Inhibin
 Valid specimen = a specimen with a risk assessment
 Associated specimen = a specimen with NO risk assesment

INTERP/Results	Definition	Description	F.U.	Mailers (include NT reminder)	Notes
NT Only:	No blood specimens from either trimester	No accession # No result; No program involvement			
1st T Serum only:	One valid 1st T specimen (No valid NT)	No numerical risk assessment yet	To get a risk assessment - need complete NT or 2nd T valid specimen or do both for Full Integrated results	Acknowledgment mailer to clinician w/reminder at 17w3d if no 2nd T specimen in SIS. Acknowledgment letter to patient.	Case stays closed with no coordinator involvement T.S.: awaiting further data
1st T Combined: Preliminary Risk Assessment (PRA)	Complete NT and 1st T valid blood specimen	Risk assessment for T21 and T18 is Screen Negative	Draw 2nd T blood specimen for Full Integrated screening	Result mailer w/ reminder at at 17w 3d if no 2nd T specimen. Patient Letter	PRA not a headline case T.S.: Awaiting Refined Risk
1st T Combined: Screen Positive	Complete NT and 1st T valid blood specimen	Risk Assessment for T21 and for T18 is Screen Positive	Either: 2nd T blood specimen for Full Integrated OR: referral to PDC	Mailer describes both options. Patient letter also. Reminder at 18 weeks, if 2 nd T specimen chosen	If pt has 2nd trimester specimen: there is a 50% chance Integrated interpretation will be Screen Positive also
Serum Integrated:	1st T valid specimen and 2nd T valid specimen (No valid NT)	Risk assessments Either: Scr Neg OR: Scr Pos -----	Referral to PDC	Screen Negative result mailer OR Screen Positive result mailer	1st specimen NOT used if exam dating only
Full Integrated:	1st T valid blood specimen and 2nd T valid blood specimen and complete NT	Risk Assessments (with highest T21/18 detection rates) Either Scr Neg OR: Scr Pos -----	Referral to PDC	Screen Negative result mailer OR Screen Positive result mailer	

PNS Program Expansion Summary Grid

INTERP/Results	Definition	Description	F.U.	Mailers (include NT reminder)	Notes
Quad Marker	2nd T valid blood specimen; (No valid NT)	Risk Assessments Either: Scr Neg OR: Scr Pos -----	Referral to PDC	Screen Negative result mailer OR Screen Positive result mailer	
Quad + NT:	2nd T valid blood specimen and complete NT (No 1st T valid specimen)	Risk assessments using NT for T21/T18 risks Either: Scr Neg OR: Scr Pos -----	Referral to PDC	Screen Negative result mailer OR Screen Positive result mailer	

CUT-OFFS	1st T combined	Serum Integrated	Full Integrated	Quad Marker	Quad + NT
T21	1 in 100	1 in 200	1 in 200	1 in 150	1 in 200
T18	1 in 50	1 in 100	1 in 100	1 in 100	1 in 100
SCD (SLOS)	--	1 in 250	1 in 250	1 in 250	1 in 250
NTD	--	≥ 2.5 or ≥ 4.5	≥ 2.5 or ≥ 4.5	≥ 2.5 or ≥ 4.5	≥ 2.5 or 4.5

EXPECTED POS RATES	1st T combined	Serum Integrated	Full Integrated	Quad Marker	Quad + NT
T21	2.50%	4.50%	4.50%	4.50%	3.40%
T18	0.20%	0.30%	0.30%	0.30%	0.20%
SLOS	—	0.20%	0.20%	0.20%	0.20%
NTD		1.00%	1.00%	1.00%	1.00%

EXPECTED DETECTION RATES**	Trisomy 21	Trisomy 18	Anencephaly	Open Spina Bifida	AWD	SLOS
Quad Marker Screening:	80%	67%	97%	80%	85%	60%
Quad + NT:	89%	72%	97%	80%	85%	60%
Serum Integrated Screening:	85%	79%	97%	80%	85%	60%
Full Integrated Screening: TOTAL from 1st + 2nd Screen Positive tests	90%	81%	97%	80%	85%	60%
First Trimester serum + NT	75%	69%	N/A	N/A	N/A	N/A
Additional detection after 2nd Trimester Specimen	15%	12%	97%	80%	85%	60%

** Assumes all women with Screen Positive results have Prenatal Diagnosis

Prenatal Screening for Down Syndrome
Estimated Positive Rates and Detection Rates (by maternal age at term)

Age	Quad		Serum Integrated		(Full) Integrated			
	Positive Rate	Detection Rate	Positive Rate	Detection Rate	First Trimester		Total after Second Trimester(*)	
					Positive Rate	Detection Rate	Positive Rate	Detection Rate
18	2%	61%	2%	71%	1%	55%	2%	81%
19	2%	61%	2%	72%	1%	55%	2%	81%
20	2%	61%	2%	73%	1%	55%	2%	81%
21	2%	61%	2%	72%	1%	55%	2%	81%
22	2%	62%	2%	72%	1%	55%	2%	81%
23	2%	62%	2%	72%	1%	55%	2%	81%
24	2%	62%	2%	73%	1%	56%	2%	81%
25	2%	63%	2%	73%	1%	57%	2%	82%
26	2%	63%	2%	73%	1%	58%	2%	83%
27	2%	64%	2%	74%	1%	58%	2%	83%
28	2%	65%	3%	74%	1%	59%	3%	83%
29	3%	67%	3%	76%	1%	61%	3%	84%
30	3%	68%	3%	77%	1%	61%	3%	84%
31	3%	69%	4%	78%	2%	64%	4%	86%
32	4%	72%	4%	80%	2%	66%	4%	87%
33	5%	74%	5%	82%	3%	69%	5%	88%
34	7%	78%	6%	84%	3%	72%	6%	89%
35	8%	81%	7%	86%	5%	75%	8%	91%
36	10%	84%	10%	88%	6%	79%	9%	92%
37	12%	86%	12%	90%	8%	81%	12%	93%
38	16%	89%	15%	92%	10%	84%	14%	94%
39	19%	91%	18%	94%	13%	86%	18%	94%
40	23%	93%	21%	94%	16%	89%	21%	96%
41	26%	94%	25%	95%	20%	90%	25%	96%
42	29%	95%	28%	96%	22%	92%	27%	97%
43	32%	95%	31%	96%	26%	93%	31%	97%
44	34%	96%	33%	97%	28%	94%	33%	97%
45	37%	96%	35%	97%	30%	94%	36%	97%
46	38%	97%	37%	97%	32%	94%	38%	98%
47	40%	97%	39%	98%	35%	95%	39%	98%
48	41%	97%	40%	98%	35%	95%	40%	98%
49	42%	97%	40%	98%	36%	95%	41%	98%
50	43%	97%	41%	98%	37%	95%	42%	98%
< 35	3%	68%	3%	77%	1%	62%	3%	85%
>=35	15%	90%	14%	93%	10%	86%	14%	94%
All Ages	4.5%	80%	4.5%	85%	2.5%	75%	4.5%	90%

(*) Second Trimester rates assume patients with positive results in first trimester accept referral and all patients with preliminary (negative) risk assessments return for second trimester screening

Prenatal Screening for Trisomy 18
Estimated Positive Rates and Detection Rates (by maternal age at term)

Age	Quad		Serum Integrated		(Full) Integrated			
					First Trimester		Total after Second Trimester(*)	
	Positive Rate	Detection Rate	Positive Rate	Detection Rate	Positive Rate	Detection Rate	Positive Rate	Detection Rate
18	0.08%	50%	0.06%	74%	<0.1%	28%	0.08%	68%
19	0.12%	50%	0.07%	74%	<0.1%	29%	0.05%	68%
20	0.07%	51%	0.06%	74%	<0.1%	29%	0.10%	68%
21	0.08%	51%	0.08%	74%	<0.1%	29%	0.07%	68%
22	0.05%	51%	0.07%	74%	<0.1%	29%	0.08%	69%
23	0.10%	51%	0.08%	75%	<0.1%	29%	0.06%	69%
24	0.10%	51%	0.10%	75%	<0.1%	30%	0.10%	69%
25	0.10%	52%	0.10%	75%	<0.1%	30%	0.09%	70%
26	0.11%	52%	0.08%	75%	<0.1%	30%	0.07%	71%
27	0.06%	53%	0.09%	76%	<0.1%	30%	0.09%	71%
28	0.10%	53%	0.09%	75%	0.05%	32%	0.10%	71%
29	0.12%	55%	0.09%	77%	0.10%	33%	0.07%	73%
30	0.13%	55%	0.10%	78%	0.07%	36%	0.19%	74%
31	0.16%	56%	0.12%	78%	0.08%	38%	0.21%	76%
32	0.20%	58%	0.14%	80%	0.11%	40%	0.22%	77%
33	0.30%	62%	0.18%	81%	0.12%	43%	0.25%	79%
34	0.37%	63%	0.31%	83%	0.16%	48%	0.27%	82%
35	0.36%	67%	0.35%	85%	0.23%	57%	0.51%	84%
36	0.78%	69%	0.37%	86%	0.39%	61%	0.81%	86%
37	0.97%	73%	0.65%	88%	0.45%	66%	0.95%	89%
38	1.39%	75%	0.84%	90%	0.72%	70%	1.27%	91%
39	1.70%	78%	1.12%	91%	1.13%	74%	1.92%	92%
40	2.44%	79%	1.27%	92%	1.40%	77%	2.29%	93%
41	2.90%	81%	1.62%	93%	1.65%	79%	2.78%	94%
42	3.94%	83%	1.80%	94%	2.77%	81%	4.12%	95%
43	4.45%	83%	2.17%	95%	2.84%	83%	4.17%	95%
44	5.12%	85%	2.33%	95%	3.78%	84%	5.15%	96%
45	5.89%	85%	2.49%	95%	4.15%	85%	5.54%	96%
46	5.43%	86%	3.07%	95%	4.50%	86%	6.07%	96%
47	6.07%	86%	3.12%	95%	4.75%	86%	6.50%	97%
48	7.24%	87%	3.05%	96%	4.77%	87%	6.62%	97%
49	7.24%	86%	2.95%	96%	5.03%	87%	6.82%	97%
50	7.24%	87%	3.13%	95%	5.06%	87%	6.70%	97%
< 35	0.13%	55%	0.11%	75%	0.05%	36%	0.12%	76%
>=35	1.38%	77%	0.80%	91%	0.82%	72%	1.41%	91%
All Ages	0.31%	67%	0.21%	79%	0.16%	59%	0.31%	81%

(*) Second Trimester rates assume patients with positive results in first trimester accept referral and all patients with preliminary (negative) risk assessments return for second trimester screening

Midtrimester Risk for Chromosome Abnormalities ¹

Age	Down Syndrome Term Risk	Down Syndrome Mid-Trimester Risk	Trisomy 18 Mid-Trimester Risk
20	1:1480	1:1140	1:4430
21	1:1460	1:1130	1:4380
22	1:1440	1:1110	1:4320
23	1:1420	1:1090	1:4250
24	1:1380	1:1060	1:4150
25	1:1340	1:1030	1:4020
26	1:1290	1:990	1:3860
27	1:1220	1:940	1:3660
28	1:1140	1:880	1:3420
29	1:1050	1:810	1:3140
30	1:940	1:720	1:2820
31	1:820	1:630	1:2460
32	1:700	1:540	1:2090
33	1:570	1:441	1:1720
34	1:456	1:351	1:1370
35	1:353	1:272	1:1060
36	1:267	1:205	1:800
37	1:199	1:153	1:600
38	1:148	1:114	1:444
39	1:111	1:85	1:333
40	1:85	1:65	1:255
41	1:67	1:51	1:200
42	1:54	1:42	1:162
43	1:45	1:35	1:136
44	1:39	1:30	1:117
45	1:35	1:27	1:104
46	1:31	1:24	1:94
47	1:29	1:22	1:87
48	1:27	1:21	1:82
49	1:26	1:20	1:79
50	1:25	1:19	1:76

The numbers provided in this table are approximate risks based on data currently available. These numbers are **population-based** risk estimates, and should not be presented as a woman's individual risk.

These numbers represent the estimated risk for a fetus with Down syndrome or Trisomy 18 at midtrimester. Approximately 23% of Down syndrome fetuses², and 70% of Trisomy 18 fetuses³ will be lost between midtrimester and term.

¹ Morris JK et al.: Revised estimates of the maternal age specific live birth prevalence of Down's syndrome. *J Med Screen* 2002; 9:2-6

² Hook, E.B.: Chromosome abnormalities and spontaneous fetal deaths following amniocentesis: further data and associations with maternal age. *Am J Hum Genet* 1983 35:110-116.

³ Hook, E.B., Cross, P.K., Schreinemachers, D.M.: Chromosomal abnormality rates at amniocentesis and in live-born infants. *J Am Med Assoc.* 1983, 249: 2034-2038.

Communicating Risks (Proportions, Percents, and Rates)

The California Expanded AFP Screening Program provides an individualized risk for a pregnancy to be affected with a fetus with Down syndrome or trisomy 18. This risk is presented as a proportion, such as 1 in 100. However, some individuals have difficulty in comprehending risk and using it in their decision-making. During genetic counseling, some individuals may understand the risk more clearly if the proportion is converted into a percent or a rate. One study indicates that converting the proportion risk into a rate (number of affected per 1000 persons) helps the patient to better understand the magnitude of their risk. (1) Also, some genetic counselors convert the proportion risk to a percent risk and then use this to explain the difference between the likelihood of an affected pregnancy versus an unaffected pregnancy.

Still, some people may prefer to state a proportion risk so that it may be compared to the risk of procedural complications or to the population risk. To facilitate the genetic counselor in communicating risk, this table is provided to assist in the conversion between proportions, percents and rates.

(1) Grimes H, Snively G: Patients' Understanding of Medical Risks: Implications for Genetic Counseling. *Obstet Gynecol* 1999; 93: 910-4.

Proportion (1 in ...)	Percent	Rate (...in 1000)
1 in 10	10.0%	100/1000
1 in 11	9.0%	90/1000
1 in 12	8.3%	83/1000
1 in 13	7.6%	76/1000
1 in 14	7.1%	71/1000
1 in 15	6.6%	66/1000
1 in 16	6.2%	62/1000
1 in 17	5.8%	58/1000
1 in 18	5.5%	55/1000
1 in 19	5.2%	52/1000
1 in 20	5.0%	50/1000
1 in 21	4.7%	47/1000
1 in 22	4.5%	45/1000
1 in 23	4.3%	43/1000
1 in 24	4.1%	41/1000
1 in 25	4.0%	40/1000
1 in 26	3.8%	38/1000
1 in 27	3.7%	37/1000
1 in 28	3.5%	35/1000
1 in 29	3.4%	34/1000
1 in 30	3.3%	33/1000
1 in 31	3.2%	32/1000
1 in 32	3.1%	31/1000
1 in 33	3.0%	30/1000
1 in 34	2.9%	29/1000
1 in 35	2.8%	28/1000

Proportion (1 in ...)	Percent	Rate (...in 1000)
1 in 37	2.7%	27/1000
1 in 38	2.6%	26/1000
1 in 40	2.5%	25/1000
1 in 41	2.4%	24/1000
1 in 43	2.3%	23/1000
1 in 45	2.2%	22/1000
1 in 47	2.1%	21/1000
1 in 50	2.0%	20/1000
1 in 52	1.9%	19/1000
1 in 55	1.8%	18/1000
1 in 58	1.7%	17/1000
1 in 62	1.6%	16/1000
1 in 66	1.5%	15/1000
1 in 71	1.4%	14/1000
1 in 76	1.3%	13/1000
1 in 83	1.2%	12/1000
1 in 90	1.1%	11/1000
1 in 100	1.0%	10/1000
1 in 111	0.9%	9/1000
1 in 125	0.8%	8/1000
1 in 142	0.7%	7/1000
1 in 166	0.6%	6/1000
1 in 200	0.5%	5/1000
1 in 250	0.4%	4/1000
1 in 333	0.3%	3/1000
1 in 500	0.2%	2/1000
1 in 1000	0.1%	1/1000

CALIFORNIA PRENATAL SCREENING PROGRAM
PNS SURVEILLANCE REPORT: DATA FOR GENERAL RELEASE

12:30 Wednesday, April 30, 2008 1

TIME PERIOD: CALENDAR 2007

The FREQ Procedure

ACC_DT	Frequency	Percent	Cumulative Frequency	Cumulative Percent
2007:01	33971	8.92	33971	8.92
2007:02	31282	8.21	65253	17.13
2007:03	36234	9.51	101487	26.64
2007:04	33553	8.81	135040	35.45
2007:05	33902	8.90	168942	44.35
2007:06	33126	8.70	202068	53.05
2007:07	30493	8.01	232561	61.06
2007:08	31921	8.38	264482	69.44
2007:09	29791	7.82	294273	77.26
2007:10	30214	7.93	324487	85.19
2007:11	28102	7.38	352589	92.57
2007:12	28301	7.43	380890	100.00

CALIFORNIA PRENATAL SCREENING PROGRAM
PNS SURVEILLANCE REPORT: DATA FOR GENERAL RELEASE

12:30 Wednesday, April 30, 2008 2

TIME PERIOD: CALENDAR 2007

ESTIMATED UTILIZATION RATE

(# IN PNC BY END OF 2ND TRIMESTER: CA VITAL STATS, 2005 DATA)
(# SCREENED: CA XAFP SCREENING PROGRAM DATA)

AGE GROUP	MATERNAL AGE	# WOMEN IN PNC BY END OF 2ND TRIMESTER	# WOMEN IN PRENATAL SCREENING	APPROX % UTILIZATION IN PNC	# MOTHERS IN NEWBORN SCREENING (+6 MOS)	APPROX % UTILIZATION BY NBS MOTHERS
<= 34	12	7	13	.	21	61.9
	13	87	44	50.6	66	66.7
	14	501	281	56.1	301	93.4
	15	1,953	1,261	64.6	1,260	.
	16	4,573	3,331	72.8	3,650	91.3
	17	8,272	5,914	71.5	7,161	82.6
	18	12,641	9,399	74.4	11,018	85.3
	19	17,800	13,255	74.5	16,888	78.5
	20	20,446	14,789	72.3	20,862	70.9
	21	21,519	15,481	71.9	22,428	69.0
	22	24,050	16,290	67.7	23,947	68.0
	23	25,023	17,144	68.5	25,299	67.8
	24	26,308	17,906	68.1	26,788	66.8
	25	27,056	19,199	71.0	27,379	70.1
	26	26,888	19,810	73.7	28,125	70.4
	27	27,152	19,985	73.6	27,944	71.5
	28	27,938	19,923	71.3	28,464	70.0
	29	28,144	19,839	70.5	28,927	68.6
	30	27,811	19,643	70.6	28,589	68.7
	31	27,368	19,124	69.9	28,058	68.2
	32	26,062	17,941	68.8	26,973	66.5
	33	24,795	16,484	66.5	25,501	64.6
	34	23,708	15,132	63.8	23,843	63.5
-----				-----	-----	-----
<= 34		430,102	302,188	70.3	433,492	69.7

CALIFORNIA PRENATAL SCREENING PROGRAM
PNS SURVEILLANCE REPORT: DATA FOR GENERAL RELEASE

12:30 Wednesday, April 30, 2008 3

TIME PERIOD: CALENDAR 2007

ESTIMATED UTILIZATION RATE

(# IN PNC BY END OF 2ND TRIMESTER: CA VITAL STATS, 2005 DATA)
(# SCREENED: CA XAFP SCREENING PROGRAM DATA)

AGE GROUP	MATERNAL AGE	# WOMEN IN PNC BY END OF 2ND TRIMESTER	# WOMEN IN PRENATAL SCREENING	APPROX % UTILIZATION IN PNC	# MOTHERS IN NEWBORN SCREENING (+6 MOS)	APPROX % UTILIZATION BY NBS MOTHERS
>= 35	35	21,348	11,392	53.4	22,472	50.7
	36	17,730	9,495	53.6	19,922	47.7
	37	14,069	7,673	54.5	16,185	47.4
	38	11,318	5,931	52.4	12,775	46.4
	39	8,966	4,488	50.1	10,112	44.4
	40	6,736	3,111	46.2	7,915	39.3
	41	4,672	2,161	46.3	5,754	37.6
	42	3,015	1,423	47.2	3,990	35.7
	43	1,852	860	46.4	2,512	34.2
	44	1,038	477	46.0	1,469	32.5
	45	603	252	41.8	813	31.0
	46	296	126	42.6	422	29.9
	47	189	82	43.4	214	38.3
	48	94	45	47.9	135	33.3
	49	64	28	43.8	87	32.2
	50	56	13	23.2	56	23.2
	51	23	12	52.2	35	34.3
	52	13	11	84.6	26	42.3
53	9	7	77.8	24	29.2	
54	12	9	75.0	19	47.4	
	UNKNOWN	11	7	63.6	15	46.7
-----		-----	-----	-----	-----	-----
>= 35		92,114	47,603	51.7	104,952	45.4

CALIFORNIA PRENATAL SCREENING PROGRAM
 PNS SURVEILLANCE REPORT: DATA FOR GENERAL RELEASE

12:30 Wednesday, April 30, 2008 4

TIME PERIOD: CALENDAR 2007

ESTIMATED UTILIZATION RATE

(# IN PNC BY END OF 2ND TRIMESTER: CA VITAL STATS, 2005 DATA)
 (# SCREENED: CA XAFP SCREENING PROGRAM DATA)

AGE GROUP	MATERNAL AGE	# WOMEN IN PNC BY END OF 2ND TRIMESTER	# WOMEN IN PRENATAL SCREENING	APPROX % UTILIZATION IN PNC	# MOTHERS IN NEWBORN SCREENING (+6 MOS)	APPROX % UTILIZATION BY NBS MOTHERS
UNKNOWN	UNKNOWN	0	31,099	.	0	.
		=====	=====	=====	=====	=====
		522,216	380,890	72.9	538,444	70.7

TIME PERIOD: CALENDAR 2007

RACE/ETHNICITY SUMMARY

The FREQ Procedure

RACGP

RACGP	Frequency	Percent
HISPANIC	213805	56.13
WHITE	78254	20.55
BLACK	19304	5.07
ASIAN	33767	8.87
OTHER	16676	4.38
MULTIPLE	9821	2.58
UNKNOWN	9263	2.43

RACGP	RACSNGL	ETH_MULT	Frequency	Percent
HISPANIC	HISPANIC		213805	56.13
WHITE	WHITE		78254	20.55
BLACK	BLACK		19304	5.07
ASIAN	CHINESE		8276	2.17
ASIAN	FILIPINA		9985	2.62
ASIAN	KOREAN		3604	0.95
ASIAN	JAPANESE		1423	0.37
ASIAN	VIETNAMESE		6323	1.66
ASIAN	LAOTIAN		656	0.17
ASIAN	CAMBODIAN		1157	0.30
ASIAN	OTHER SE ASIAN		1717	0.45
ASIAN	MULTIPLE	ALL ASIAN	626	0.16
OTHER	SAMOAN		404	0.11
OTHER	HAWAIIAN		152	0.04
OTHER	GUAMANIAN		111	0.03
OTHER	NAT AMERICAN		878	0.23
OTHER	MIDDLE EASTERN		2668	0.70
OTHER	ASIAN INDIAN		6879	1.81
OTHER	OTHER		5571	1.46
OTHER	MULTIPLE	ALL PAC ISL	13	0.00
MULTIPLE	MULTIPLE	OTHER	9821	2.58
UNKNOWN	UNKNOWN		9263	2.43

RACE DESIGNATION IS DERIVED FROM CHECK BOXES ON THE EXPANDEDAPP TEST REQUEST FORM AS FOLLOWS:
 HISPANIC = HISPANIC ONLY, OR HISPANIC +: WHITE, OTHER AND/OR UNKNOWN
 WHITE = WHITE ONLY

REFERENCE: GEN_REL.sas DATABASE: SIS CRDB

TIME PERIOD: CALENDAR 2007

INITIAL INTERPRETATION

The FREQ Procedure

OVRSTGP

OVRSTGP	Frequency	Percent
INAD SPECIMEN	1346	0.35
INSUFF PAT INFO	46480	12.20
DRAWN TOO EARLY	5745	1.51
DRAWN TOO LATE	7591	1.99
INCONSIST VALUES	54	0.01
SCREEN NEGATIVE	291662	76.57
SCREEN POSITIVE	23544	6.18
PART PNL, SPC CIR	1715	0.45
UNAUTHORIZED RDRW	2131	0.56
NOT SCREENABLE	448	0.12
NULL	174	0.05

CALIFORNIA PRENATAL SCREENING PROGRAM
 PNS SURVEILLANCE REPORT: DATA FOR GENERAL RELEASE

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TIME PERIOD: CALENDAR 2007

MONITORING # AND % SCREEN-POSITIVE: BY AGE GROUP

AGE GROUP	# INC RISK: ALL	% INC RISK: ALL	# INC RISK: NTD	% INC RISK: NTD	# INC RISK: T21	% INC RISK: T21	# INC RISK: T18	% INC RISK: T18	# INC RISK: SLOS	% INC RISK: SLOS
<= 34	13855	5.36	3036	1.17	10262	4.01	513	0.20	611	0.23
>= 35	8432	19.60	587	1.36	7567	19.56	599	1.57	155	0.36
UNKNOWN	1227	8.99	153	1.12	1051	7.99	44	0.33	31	0.23
=OVERALL=	23514	7.46	3776	1.19	18880	6.14	1156	0.38	797	0.25

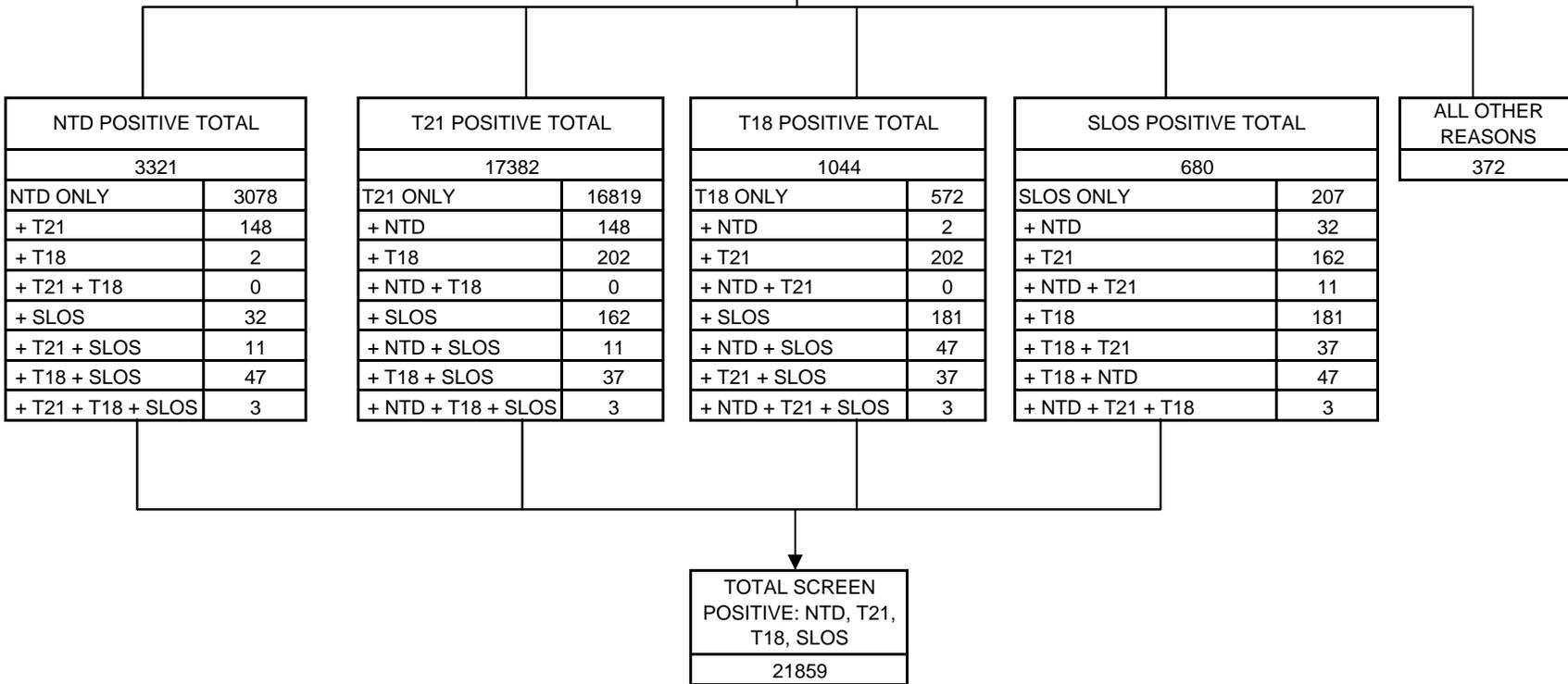
THE OVERALL SCREEN POSITIVE RATE IS CALCULATED FROM SCREEN POSITIVE AND SCREEN NEGATIVE RESULTS ONLY:
 OVERALL SCREEN POSITIVE RATE = 100 X (SCREEN POSITIVE/(SCREEN POSITIVE + SCREEN NEGATIVE))
 BECAUSE SOME WOMEN ARE DOUBLY SCREEN POSITIVE, (% INC RISK: ALL) IS SLIGHTLY LESS THAN
 (% INC RISK: NTD + % INC RISK: T21 + % INC RISK: T18)

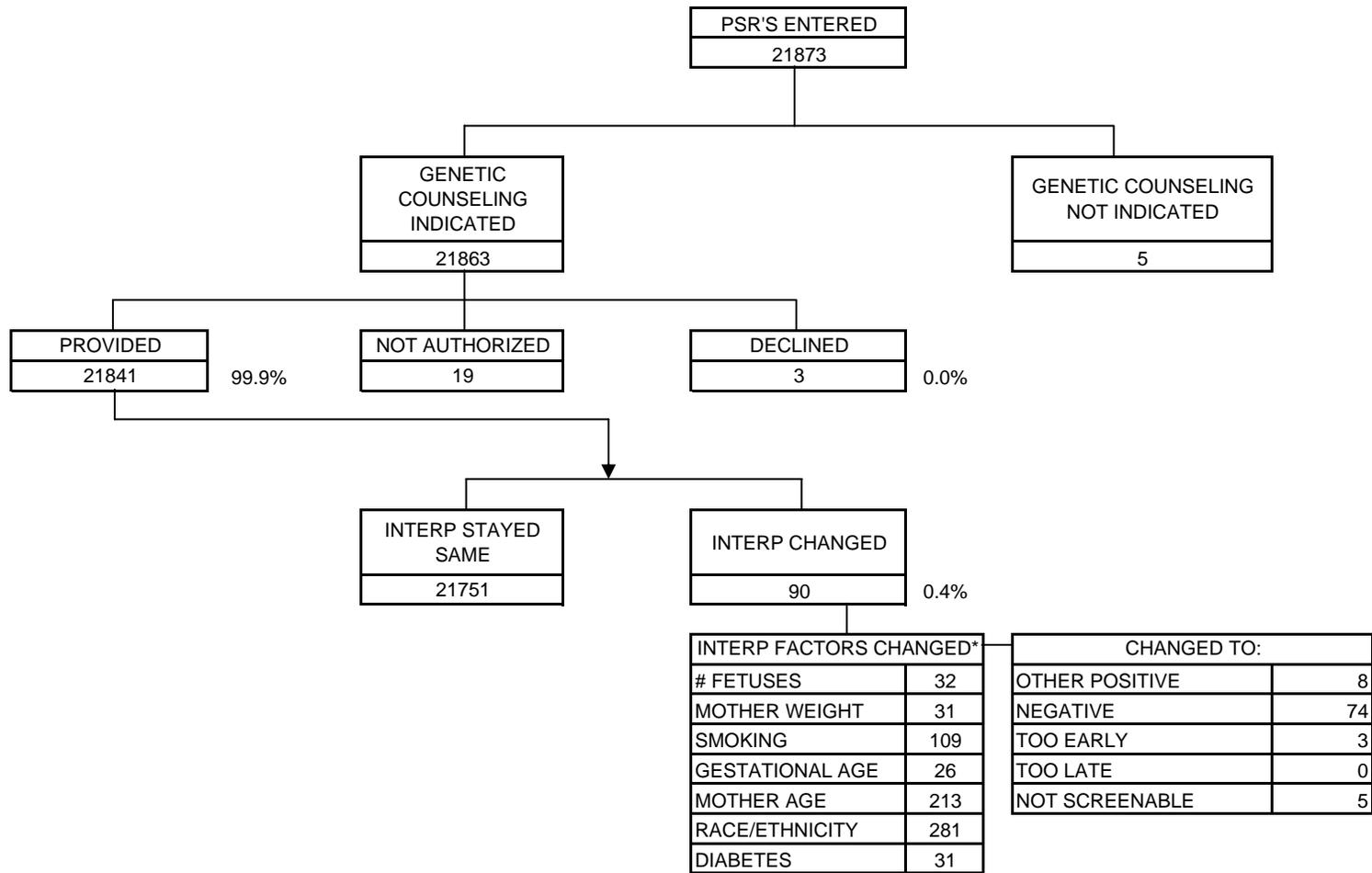
NOTE: INITIAL INTERPRETATION

REFERENCE: GEN_REL.sas DATABASE: SIS CRDB

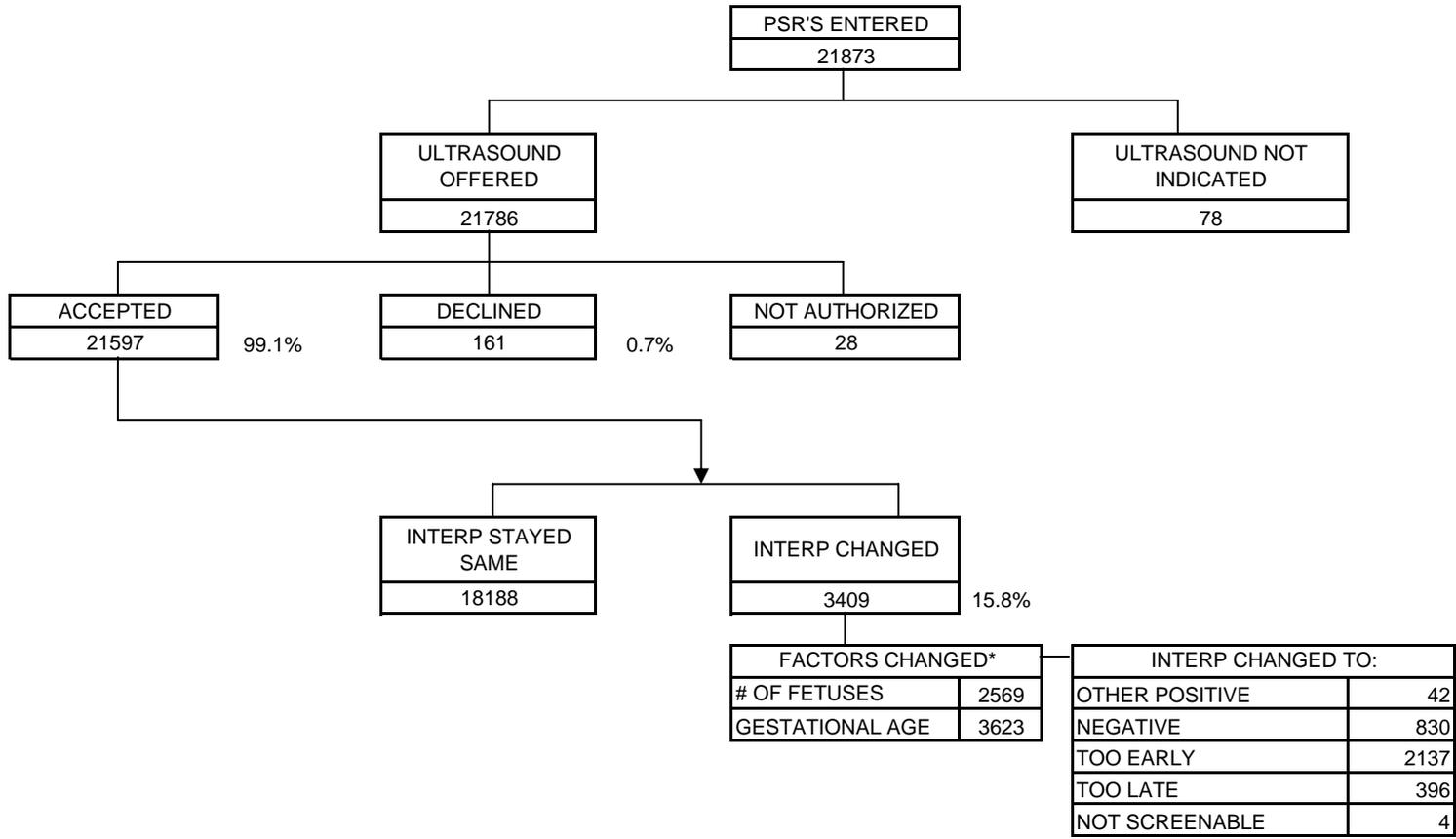
PDC APPOINTMENT KEPT
21927

PSR'S ENTERED
21873

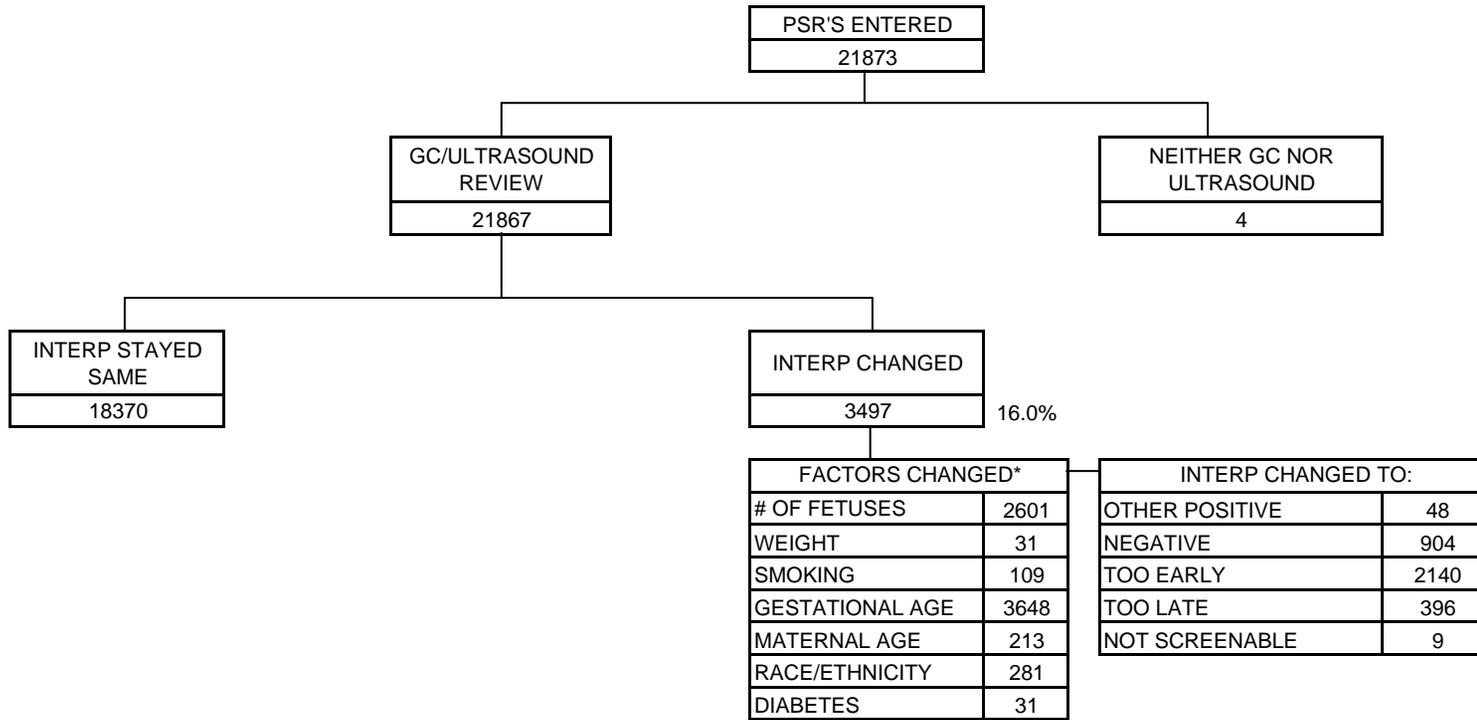




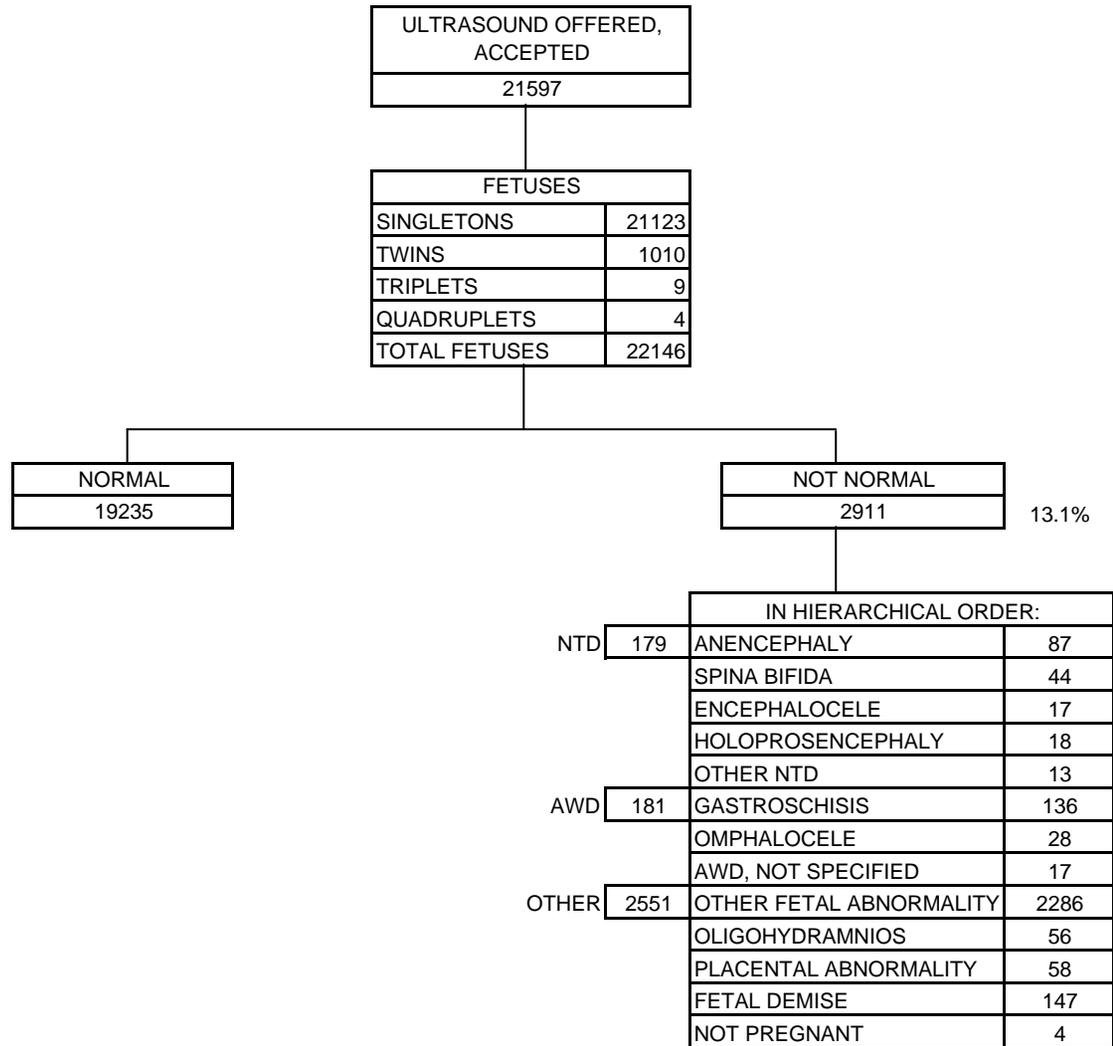
* ALL INTERP FACTOR CHANGES ARE COUNTED REGARDLESS OF THEIR EFFECT ON THE OVERALL INTERPRETATION

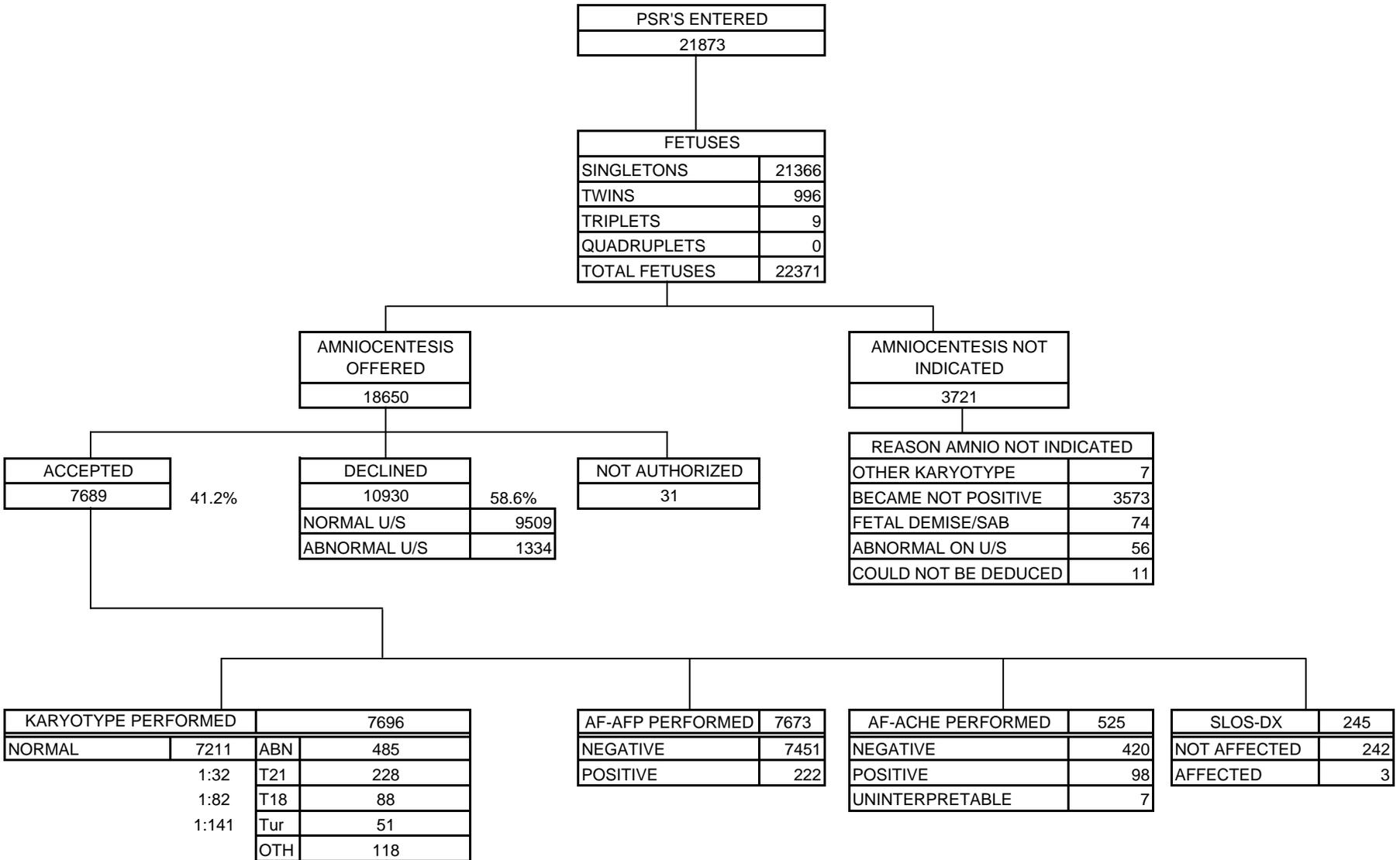


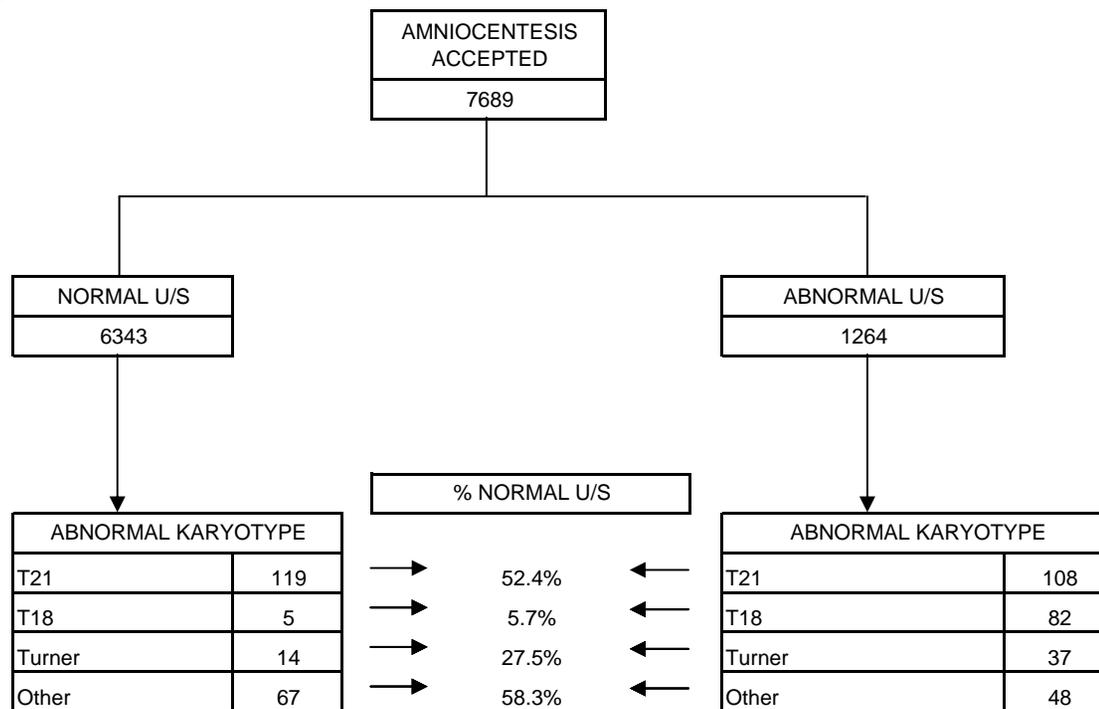
* ALL INTERP FACTOR CHANGES ARE COUNTED REGARDLESS OF THEIR EFFECT ON THE OVERALL INTERPRETATION



* ALL INTERP FACTOR CHANGES ARE COUNTED REGARDLESS OF THEIR EFFECT ON THE OVERALL INTERPRETATION







STATUS OF FETUS AFTER PRENATAL DIAGNOSIS OF AN ABNORMALITY*								
ABNORMALITY (HIERARCHICAL)	TOTAL	FETAL DEMISE/SPONTANEOUS ABORTION	KNOWN ELECTIVE TERMINATION		INTENDS TO TERMINATE/ FETAL REDUCTION		CONT'G	UNKNOWN
DOWN SYNDROME	228	4	53	23.2%	39	17.1%	103	29
TRISOMY 18	88	4	33	37.5%	13	14.8%	31	7
TURNER SYNDROME	50	2	20	40.0%	1	2.0%	18	9
OTHER CHROME ABN	118	4	22	18.6%	3	2.5%	80	9
ALL CHROME ABNS	484	14	128	26.4%	56	11.6%	232	54
ANENCEPHALY	88	1	25	28.4%	16	18.2%	37	9
SPINA BIFIDA	42	1	10	23.8%	5	11.9%	24	2
ENCEPHALOCELE	19	5	4	21.1%	0	0.0%	7	3
HOLOPROSENCEPHALY	12	0	8	66.7%	1	8.3%	3	0
OTHER NTD	13	1	5	38.5%	2	15.4%	4	1
ALL NTDS	174	8	52	29.9%	24	13.8%	75	15
GASTROSCHISIS	137	1	1	0.7%	2	1.5%	130	3
OMPHALOCELE	21	2	5	23.8%	0	0.0%	12	2
AWD, NOT SPECIFIED	16	1	2	12.5%	2	12.5%	7	4
ALL AWDS	174	4	8	4.6%	4	2.3%	149	9
ALL TARGET ABNORMALITIES	832	26	188	22.6%	84	10.1%	456	78

* STATUS AT TIME OF PSR SUBMISSION

PDC Services by Calendar Year for All Centers Combined

Report Type	Non-XAFP Patient
Calendar Year	2007

Designed Using Fiscal Year Quarters (i.e., Qtr1=July-September) to Create Calendar Year

Section I: Indication for Prenatal Diagnosis

1. Age	Invasive Procedure Offered					Invasive Procedure Not Offered		
	CVS	EA	Amnio	PUBS	Declined at Visit	SAB	Couns. Only	Couns. & US
35	409	16	2,054	0	5,868	15	170	484
36	434	25	2,411	0	5,229	15	128	478
37	491	21	2,384	0	4,324	12	156	446
38	526	27	1,999	0	3,275	23	127	396
39	493	15	1,731	0	2,412	9	119	319
40	471	14	1,331	0	1,704	20	94	250
41	383	14	1,014	0	1,103	19	88	197
42	301	2	615	0	712	14	64	163
43	168	3	417	0	433	10	62	112
44	98	2	219	0	246	9	34	52
45	44	2	99	0	121	3	8	33
>45	46	2	96	0	150	3	8	28
Age Unknown	0	0	0	0	1	2	1	0
Sum:	3,864	143	14,370	0	25,578	154	1,059	2,958

2. No Other Indication	Invasive Procedure Offered					Invasive Procedure Not Offered		
	CVS	EA	Amnio	PUBS	Declined at Visit	SAB	Couns. Only	Couns. & US
Maternal Concern (<Age 35)	590	176	5,427	7	3,393	55	954	10,888
Sum:	590	176	5,427	7	3,393	55	954	10,888

3. Family History	Invasive Procedure Offered					Invasive Procedure Not Offered			
	CVS	EA	Amnio	PUBS	Declined at Visit	SAB	Couns. Only	Couns. & US	
Chromosome Abnormality	<i>Down Syndrome</i>	71	3	187	0	913	1	71	340
Chromosome Abnormality	<i>Trisomy 13</i>	9	0	16	0	38	0	5	8
Chromosome Abnormality	<i>Trisomy 18</i>	20	1	39	0	43	1	33	43
Chromosome Abnormality	<i>Chrom Abn. in Parent</i>	17	1	25	0	17	0	16	8
Chromosome Abnormality	<i>Other</i>	35	1	106	0	242	1	52	96
NTD (non-AFP referrals)	<i>Neural Tube Defects</i>	3	5	61	0	285	0	13	136
X-Linked	<i>Fragile X</i>	4	0	6	0	8	0	9	9
X-Linked	<i>Other</i>	7	2	10	0	37	0	15	30
Metabolic/Recessive or Dominant Disorders*	<i>Tay Sachs Disease</i>	3	0	3	0	9	0	10	14
Metabolic/Recessive or Dominant Disorders*	<i>Cystic Fibrosis</i>	27	0	58	0	231	0	38	78
Metabolic/Recessive or Dominant Disorders*	<i>Other</i>	29	3	39	0	231	2	48	229
Fam Hx or Risk for: Hemoglobinopathies	<i>Hemoglobinopathies</i>	19	2	64	0	303	2	35	146
Other Heritable Disorders	<i>Oth Heritable Disorder</i>	27	0	170	0	1,453	2	319	1,654
*=(other than Hemoglobinopathie)	Sum:	271	18	784	0	3,810	9	664	2,791

Report Type	Non-XAFP Patient
Calendar Year	2007

4. Other Indication for Counseling or Prenatal Services	Invasive Procedure Offered					Invasive Procedure Not Offered		
	CVS	EA	Amnio	PUBS	Declined at Visit	SAB	Couns. Only	Couns. & US
Abnormal Ultrasound	199	13	1,513	1	1,908	2	142	957
Teratogen Exposure	2	0	57	0	496	2	251	1,060
Multiple Miscarriages	11	1	15	0	196	8	144	251
Non-XAFP Multiple Marker	230	20	820	0	485	4	241	339
Other	68	2	287	0	1,242	4	138	1,264
Unknown	1	0	13	0	16	0	0	17
Sum:	511	36	2,705	1	4,343	20	916	3,888

GRAND TOTALS:	5,236	373	23,286	8	37,124	238	3,593	20,525
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5. Biochemical and DNA Family History		Biochem	DNA
Chromosome Abnormality	<i>Other</i>		
Chromosome Abnormality			
Fam Hx or Risk for: Hemoglobinopathies	<i>Hemoglobinopathies</i>	47	37
Metabolic/Recessive or Dominant Disorders*	<i>Cystic Fibrosis</i>	3	208
Metabolic/Recessive or Dominant Disorders*	<i>Other</i>	1	159
Metabolic/Recessive or Dominant Disorders*	<i>Tay Sachs Disease</i>	49	47
X-Linked	<i>Fragile X</i>	0	150
X-Linked	<i>Other</i>	2	10
	Sum:	102	611

Section II: Repeat Samples

Number of Amnios for Non Growth	1
Num of Amnios for CVS Culture Fail	4
Number of Confirming PUBS	1
Num Repeat Samples for Other Indctn	19
Sum:	25

Section III: Abnormalities

Number of Chrom Abn	1,016
Number of NTD Abn	90
Number of Known Impact Abn	799
Number of Questionable Abn	1,107
Sum:	3,012

Report Type	Non-XAFP Patient	Non-XAFP Patient
Calendar Year	2007	0

Section IV: Demographic Information on All Patients Served

1) Race/Ethnicity

White	52,463
Black	3,139
Native American	1,110
Middle Eastern	1,308
Asian Indian	1,308
Cambodian	174
Laotian	128
Vietnamese	1,802
Other Southeast Asian	429
Filipino	1,711
Samoan	24
Hawaiian	62
Guamanian	33
Chinese	3,351
Japanese	1,115
Korean	668
Other Asian Pacific Islander	652
Other Race Ethnicity	1,382
Unknown Race Ethnicity	2,269
Sum:	73,128

2) Hispanic Origin

Hispanic	26,141
Non Hispanic	44,520
Unknown Hispanic	2,172
Sum:	72,833

3) Residence

County of Residence for California Patients:

County	Count
Alameda	3,972
Amador	26
Butte	205
Calaveras	30
Colusa	29
Contra Costa	2,000
Del Norte	3
El Dorado	226
Fresno	1,668
Glenn	27
Humboldt	113
Imperial	69
Inyo	8
Kern	221
Kings	128
Lake	64
Lassen	2
Los Angeles	21,646
Madera	226
Marin	1,446
Mariposa	10
Mendocino	103
Merced	137
Modoc	4
Mono	11
Monterey	759
Napa	217
Nevada	47
Orange	5,527
Placer	398
Plumas	2
Riverside	2,810
Sacramento	2,693
San Benito	40
San Bernardino	2,738
San Diego	4,389
San Francisco	4,888
San Joaquin	966
San Luis Obispo	242
San Mateo	3,287
Santa Barbara	465

Santa Clara	4,697
Santa Cruz	371
Shasta	158
Siskiyou	5
Solano	500
Sonoma	1,166
Stanislaus	505
Sutter	76
Tehama	45
Trinity	7
Tulare	458
Tuolumne	13
Unknown	1,070
Ventura	724
Yolo	415
Yuba	48
Sum:	72,100

California Urban/Rural Distribution:

Urban/Rural	Residents
Rural	1,254
Unknown	1,070
Urban	69,776
Sum:	72,100

In-State/Out-of-State Residence:

Out of State Residence	123
Non US Residence	17
Unknown Residence	1,727
Sum:	1,867

Information available through the Prenatal Screening Program
Website: www.cdph.ca.gov/programs/pns

Program Information for Patients

Patient Education Booklet and Consent

English
Spanish
Chinese
Korean
Vietnamese

Information for Providers

Prenatal Care Provider Handbook

Supply Order Form 2009

Two-sided Program Summary for Clinical Staff

Two-sided Program Summary for Support Staff

Time Window for the Prenatal Screening Program, First Trimester

Time Window for the Prenatal Screening Program, Second Trimester

Resources

Prenatal Diagnosis Centers by County

List of Registered NT Practitioners

Form for Reporting a Neural Tube Defect

Form for Reporting a Chromosomal Defect

News, Hot Topics & Information

Announcement: California Expanded AFP Screening Program Update – January, 2009

Announcement: Regional Forums For Licensed Clinicians on Prenatal Screening

Health Information

First Trimester

Screen Positive Increased Risk for T21

English
Spanish
Chinese
Korean
Vietnamese

Screen Positive Increased Risk for T18

English
Spanish
Chinese
Korean
Vietnamese

Second Trimester

Screen Positive Increased Risk for T21

English
Spanish
Chinese
Korean
Vietnamese

Screen Positive Increased Risk for T18

English
Spanish
Chinese
Korean
Vietnamese

Screen Positive Increased Risk for NTD

English
Spanish
Chinese
Korean
Vietnamese

Screen Positive Increased Risk for SLOS

English
Spanish
Chinese
Korean
Vietnamese

Program Supplies and Patient Education Materials

The following materials are available free of charge. Call (866) 718-7915 toll free to order.

Program Supplies

First Trimester Test Forms – Test Request Forms for drawing blood from 10 weeks 0 days to 13 weeks 6 days.

Second Trimester Test Forms – Test Request Forms, for drawing blood from 15 weeks 0 days to 20 weeks 0 days.

Blood Shipping Kits – each kit contains one serum separator tube, one tray, one absorbent pouch, and one box to mail the blood specimen. The kits meet the shipping requirements of the U.S. Postal Service.

Tubes – 3.5ml Beckton-Dickinson serum separator tubes with a yellow hemoguard. These tubes must be used for the Prenatal Screening Program. **No other tubes are allowed.**

All Prenatal Screening Program supplies are the property of the State of California. Other use is strictly prohibited.

Patient Education Materials

Patient Booklet – *The California Prenatal Screening Program.*

This booklet describes the Prenatal Screening Program, its purpose and cost. It also contains the consent form. It is available in English, Spanish, Chinese, Vietnamese, and Korean.

Important Information for Parents About the Newborn Screening Test

Provides a description of the Newborn Screening Tests. (Required by State law for prospective parents.)

Folate Pamphlet – *Folic Acid: Every Woman, Every Day.*

This brochure informs women about the benefits of folic acid particularly in preventing birth defects. It is available in English and Spanish.

Prenatal Diagnosis of Birth Defects

This brochure describes genetic counseling, ultrasound, amniocentesis, and CVS. It provides a checklist for women to use to see whether prenatal diagnosis may be recommended for them in this pregnancy. It is available in English, Spanish, and Chinese.

Screen Positive Booklets

These booklets are only for women with **Screen Positive** Prenatal Screening blood test results. They are given to women receiving follow-up services at a State-Approved Prenatal Diagnosis Center (PDC).

For **first trimester Screen Positive** results, there are separate booklets for trisomy 21 and for trisomy 18, in English and Spanish. Electronic copies for Chinese, Korean, and Vietnamese are available on the Prenatal Screening Program website.

For **second trimester Screen Positive** results, there are separate booklets each for trisomy 21, trisomy 18, SLOS, and neural tube defects (NTD), in English and Spanish. Electronic copies for Chinese, Korean, and Vietnamese are available on the Prenatal Screening Program website.

These and other Program materials, including Supplies Order Forms, are available on the Program website: www.cdph.ca.gov/programs/pns.