

GENETIC DISEASE SCREENING PROGRAM  
MAY 2011  
ESTIMATE  
*for*  
FISCAL YEARS  
2010-11 *and* 2011-12



CALIFORNIA  
DEPARTMENT OF PUBLIC HEALTH

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**APPENDICES**

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## **Management Summary**

The Genetic Disease Screening Program (GDSP) May Estimate makes no changes from the November 2010 Estimate.

BUDGET ESTIMATE OVERVIEW

GENETIC DISEASE SCREENING PROGRAM

May 2011 Estimate

GENETIC DISEASE SCREENING PROGRAM MAY 2011 BUDGET ESTIMATE	2010-2011			2011-2012		
	2010-2011 NOVEMBER ESTIMATE	2010-2011 MAY REVISE	DIFFERENCE	2011 2012 GOVERNOR'S BUDGET	2011-2012 MAY REVISE	DIFFERENCE
<b>LOCAL ASSISTANCE</b>						
<b>NBS</b>						
Contract Laboratories:	\$6,857,000	\$6,857,000	\$0	\$7,177,000	\$7,177,000	\$0
Tech & Sci:	\$23,150,000	\$23,150,000	\$0	\$23,165,000	\$23,165,000	\$0
System Project & Maintenance:	\$3,988,000	\$3,988,000	\$0	\$3,773,000	\$3,773,000	\$0
Case Management & Coordination Services:	\$4,500,000	\$4,500,000	\$0	\$4,575,000	\$4,575,000	\$0
Reference Laboratories:	\$2,550,000	\$2,550,000	\$0	\$2,491,000	\$2,491,000	\$0
Follow-up Diagnostic Services:	\$2,500,000	\$2,500,000	\$0	\$2,500,000	\$2,500,000	\$0
Result Reporting & Fee Collection	\$1,290,000	\$1,290,000	\$0	\$1,500,000	\$1,500,000	\$0
	\$44,835,000	\$44,835,000	\$0	\$45,181,000	\$45,181,000	\$0
<b>PNS</b>						
Contract Laboratories:	\$5,015,000	\$5,015,000	\$0	\$5,122,000	\$5,122,000	\$0
Tech & Sci:	\$13,146,000	\$13,146,000	\$0	\$13,300,000	\$13,300,000	\$0
System Project & Maintenance:	\$4,803,000	\$4,803,000	\$0	\$4,803,000	\$4,803,000	\$0
Case Management & Coordination Services:	\$6,110,000	\$6,110,000	\$0	\$6,242,000	\$6,242,000	\$0
Prenatal Diagnostic Services:	\$17,376,000	\$17,376,000	\$0	\$17,411,000	\$17,411,000	\$0
Result Reporting & Fee Collection	\$1,942,000	\$1,942,000	\$0	\$1,942,000	\$1,942,000	\$0
	\$48,392,000	\$48,392,000	\$0	\$48,820,000	\$48,820,000	\$0
<b>LOCAL ASSISTANCE, TOTAL</b>	<b>\$93,227,000</b>	<b>\$93,227,000</b>	<b>\$0</b>	<b>\$94,001,000</b>	<b>\$94,001,000</b>	<b>\$0</b>

**Genetic Disease Screening Program – Newborn Screening Testing  
BUDGET DETAIL – May 2011**

**COST CENTER: Contract Laboratories**

Laboratory testing of specimens is performed at regional screening laboratories contracted by the State to screen newborns for 75 specific genetic disorders. Costs include laboratory services for performing genetic screening tests. Screening laboratories ascertain the possible presence of a birth defect or a congenital disorder; a screening test is not diagnostic, additional follow up is likely to be required for a case that has an initial positive or questionable screening test result. The State contracts with several regional contract laboratories that are paid on a per screening test basis.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>Average Cost Per Case</u>
2009/2010	526,934	\$ --
2010/2011	532,980	\$ 12.87
2011/2012	540,442	\$ 13.28

**COST CENTER: Technical & Scientific**

Costs are associated with specimen screening and include reagents, supplies and processing, limited maintenance and support (as it directly relates to the reagents) of laboratory equipment that is with the contract laboratories. In addition, there are costs associated with specimen screening including: laboratory supplies, blood specimen filter paper, blood specimen storage and costs for special packaging for blood specimen transport. Reagents, which are the majority of the Technical & Scientific costs, are purchased in lots based on anticipated caseload. Reagents vary in costs depending on the type of screening performed.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>Average Cost Per Case</u>
2009/2010	526,934	\$ --
2010/2011	532,980	\$ 43.44
2011/2012	540,442	\$ 42.86

**COST CENTER: Case Management and Coordination Services**

Services provided to infants that screen initial positive or have questionable screening test results for the 75 genetic disorders screened. These services include specific confirmatory services, family consultation – including consultation with the infant's Pediatrician, genetic disease counseling, family educational services and coordinated care referrals to specialized care medical institutions such as university medical centers and hospitals that specialize in those particular genetic disorders. The NBS Area Service Centers (ASC) provide critical coordination and tracking services to ensure appropriate diagnostic measures are completed and that affected infants are provided with appropriate medical care and receive treatment within a life-critical timeframe. Costs are fixed for a required core team of clinical professionals. Costs vary by ASC dependent upon the geographical location as well as the volume of caseload served.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>% of NBS Cases</u>	<u>Average Cost Per Case</u>
2009/2010	9,169	1.74%	\$ --
2010/2011	9,274	1.74%	\$ 485.23
2011/2012	9,404	1.74%	\$ 486.50

**Genetic Disease Screening Program – Newborn Screening Testing  
Budget Detail – November 2010**

**COST CENTER: Reference Laboratories**

Cases that result in a positive screening test are referred for diagnostic testing at a confirmatory laboratory. Confirmatory testing is carried out at designated reference laboratories. Costs include medical and confirmatory diagnostic tests, as well as fixed costs for lab technical support. Reference Laboratories are reimbursed on a cost per test basis, with one laboratory doing all confirmatory testing for a particular genetic disorder.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>% of NBS Cases</u>	<u>Average Cost Per Case</u>
2009/2010	14,543	2.76%	\$ --
2010/2011	14,710	2.76%	\$ 173.35
2011/2012	14,916	2.76%	\$ 167.00

**COST CENTER: Follow-up Diagnostic Services**

Services are for infants with positive or questionable genetic screening results which require extended monitoring while undergoing confirmatory testing and diagnosis. Services include coordination with the NBS ASC and the Program for ongoing medical care, ensuring the establishment of infant treatment plans through specialty care hospitals and university medical centers specializing in the specific diagnoses, such as sickle cell anemia, cystic fibrosis, PKU, beta thalassemia, alpha thalassemia, and various neurologic, metabolic and endocrine disorders. Case data is provided to the Program on infants with a disorder as a means of tracking as well as confirmation, evaluation and refinement of Program standards. Services are provided through Special Care Centers, which are composed of highly specialized medical teams and cost is based on per case reimbursement.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>% of NBS Cases</u>	<u>Average Cost Per Case</u>
2009/2010	2,740	.52%	\$ --
2010/2011	2,772	.52%	\$ 901.88
2011/2012	2,810	.52%	\$ 889.68

**Genetic Disease Screening Program - Prenatal Testing  
BUDGET DETAIL – May 2011**

**COST CENTER: Contract Laboratories**

Laboratory testing to screen pregnant women for genetic and congenital disorders, such as Trisomy 21, Trisomy 18, Smith-Lemli-Opitz Syndrome (SLOS) and Neural Tube Defects. Costs include laboratory services for performing prenatal genetic screening tests. The screening test estimates the chance or risk that the fetus has a certain birth defect; the screening provides a Risk Assessment and not a diagnosis. The State contracts with several regional contract laboratories that are paid on a per screening test basis.

<u>Fiscal Year</u>	<u>Total # of Cases</u>	<u>Average Cost Per Case</u>	<u>1st Trimester Screens</u>	<u>Average Cost Per Case</u>	<u>2nd Trimester Screens</u>	<u>Average Cost Per Case</u>
2009/2010	437,345	\$ --	--	\$ --	--	\$ --
2010/2011	453,033	\$11.07	255,599	\$4.44	443,150	\$8.76
2011/2012	459,376	\$11.15	263,267	\$4.45	450,000	\$8.78

**COST CENTER: Technical & Scientific**

Costs associated with screening services provided at the laboratory and include reagents, limited maintenance and support (as it directly relates to the reagents) of laboratory equipment, supplies and processing. In addition, there are several costs associated with screening including: blood specimen tubes and laboratory supplies blood specimen storage, and costs for special packaging for blood specimen transport. Reagents, which are the majority of the Technical & Scientific costs, are purchased in lots based on anticipated caseload. Reagents vary in costs depending on the type of screening performed.

<u>Fiscal Year</u>	<u>Total # of Cases</u>	<u>Average Cost Per Case</u>	<u>1st Trimester Cases</u>	<u>Average Cost Per Case</u>	<u>2nd Trimester Cases</u>	<u>Average Cost Per Case</u>
2009/2010	437,345	\$ --	--	\$ --	--	\$ --
2010/2011	453,033	\$29.02	255,599	\$13.35	443,150	\$21.96
2011/2012	459,376	\$28.95	263,267	\$13.13	450,000	\$21.87

**Genetic Disease Screening Program - Prenatal Testing  
Budget Detail – November 2010**

**COST CENTER: Case Management and Coordination Services**

Services provided to pregnant women that screen positive or have questionable results. Includes coordination of first and second trimester screens and NT Ultrasounds, identify patients whose blood specimens were drawn too early or were inadequate, requiring additional blood draws. The NBS Area Service Centers (ASC) provide clinician and patient education and consultations; make referrals to Prenatal Diagnostic Centers for diagnostic and confirmatory tests, and genetic counseling, and track patients to ensure appointments are kept and patients seen within prescribed timeframes. Confirm and verify specific patient information as needed with the treating physician offices, and the Prenatal Diagnostic Centers. Costs are fixed for a required core team of medical professionals for the NBS ASC to ensure adequate personnel and infrastructure needs are always in place to provide for all cases referred. Costs vary by ASC dependent upon the geographical location as well as the distribution of caseload.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>% of Prenatal Cases</u>	<u>Average Cost Per Case</u>
2009/2010	106,712	24.4%	\$ --
2010/2011	110,885	24.5%	\$ 55.10
2011/2012	111,994	24.4%	\$ 55.74

**COST CENTER: Prenatal Diagnostic Services**

Women with positive results are provided additional services which include confirmatory and diagnostic prenatal testing, genetic counseling, education, and coordinated medical care referrals. Coordination and consultation with patient's physician, and specialty care providers. Services are provided through Prenatal Diagnostic Centers and are paid on the basis of services provided.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>% of Prenatal Cases</u>	<u>Average Cost Per Case</u>
2009/2010	19,364	4.43%	\$ --
2010/2011	20,119	4.44%	\$ 863.66
2011/2012	20,320	4.42%	\$ 856.84

**GENETIC DISEASE TESTING FUND**  
**FUND CONDITION REPORT**  
**DOLLARS IN THOUSANDS**

<b>RESOURCES</b>	<b>2009-10</b>	<b>2010-11</b>	<b>2011-12</b>
<b>BEGINNING BALANCE</b>	\$5,912	\$1,413	\$3,796
Prior Year Adjustment	(1,945)	-	-
<i>Adjusted Beginning Balance</i>	<u>3,967</u>	<u>1,413</u>	<u>3,796</u>
<b>REVENUES</b>			
Genetic Disease Testing Fees	114,536	117,389	119,032
Income from Surplus Investments	27	50	50
Escheat of Unclaimed Checks & Warrants	1	-	-
<b>TOTALS, REVENUES</b>	<u>114,564</u>	<u>117,439</u>	<u>119,082</u>
<b>TRANSFERS</b>			
To General Fund for Loan Repayment per Item 4260-011-0001, Budget Act of 2003	-4,240	-	-
<b>TOTALS, TRANSFERS</b>	<u>-4,240</u>	<u>0</u>	<u>0</u>
<b>TOTALS, REVENUES AND TRANSFERS</b>	<u>\$110,324</u>	<u>\$117,439</u>	<u>\$119,082</u>
<b>TOTAL RESOURCES</b>	<u>\$114,291</u>	<u>\$118,852</u>	<u>\$122,878</u>

<b>EXPENDITURES AND EXPENDITURE ADJUSTMENTS</b>			
<b>STATE OPERATIONS</b>			
2010-11 Budget Act Appropriation	18,625	20,723	20,723
GDSP Administration	(16,382)	(18,480)	(18,480)
Lease Revenue Debt Service	(1,692)	(1,692)	(1,692)
HIPAA	(551)	(551)	(551)
Adjustments to State Operations:			
BCP: FH-02 Blood Specimen Repositories		576	566
BCP: FH-03 Business System Upgrade Project		1,385	901
AB 2300/GLPC			67
Employee Compensation Adjustments		-2,114	-897
Lease Revenue Debt Service		408	405
Miscellaneous Baseline Adjustments		737	683
<i>Subtotal, State Operations</i>	<u>18,625</u>	<u>21,715</u>	<u>22,448</u>
<b>LOCAL ASSISTANCE</b>			
2010-11 Budget Act Appropriation	94,235	95,205	95,205
Adjustments to Local Assistance:			
November 2010 Estimate		-1,978	-1,204
<i>Subtotal, Local Assistance</i>	<u>94,235</u>	<u>93,227</u>	<u>94,001</u>
State Controller	18	44	42
Financial Information System for California (FI\$Cal)	-	70	97
<b>TOTAL EXPENDITURES AND EXPENDITURE ADJUSTMENTS</b>	<u>112,878</u>	<u>115,056</u>	<u>116,588</u>

<b>FUND BALANCE</b>	1,413	3,796	6,290
	1%	3.3%	5.4%

**REVENUE PROJECTIONS**

**2010-11**

2010-11 NBS FEES BASED ON	532,980	TESTS @	\$102.75	AND	98%	=	\$53,668,421	
2010-11 PNS FEES BASED ON	226,517	TESTS @	\$152.00	AND	93%	=	\$32,020,372	
2010-11 PNS FEES BASED ON	226,517	TESTS @	\$150.48	AND	93%	=	\$31,700,169	
	<u>453,033</u>						<u>\$63,720,541</u>	
								<b>\$117,388,962</b> Estimated

**2011-12**

2011-12 NBS FEES BASED ON	540,442	TESTS @	\$102.75	AND	98%	=	\$54,419,807	
2011-12 PNS FEES BASED ON	229,688	TESTS @	\$152.00	AND	93%	=	\$32,468,674	
2011-12 PNS FEES BASED ON	229,688	TESTS @	\$150.48	AND	93%	=	\$32,143,988	
	<u>459,376</u>						<u>\$64,612,662</u>	
								<b>\$119,032,469</b> Estimated

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## **GENETIC DISEASE SCREENING PROGRAM ASSUMPTIONS**

### **May 2011**

### **FISCAL YEARS 2010-11 & 2011-12**

#### **INTRODUCTION**

The Genetic Disease Screening Program (GDSP) Estimate is based upon the information outlined in the following pages. The Estimate includes the costs of all major components necessary to administer the program except State Operations. The Estimate is presented in two sections: (1) the base and (2) the adjustments to the base. The base estimate is the anticipated level of program expenditures assuming that there will be no changes in program direction and is derived from prior year actual caseload and expenditures. Adjustments to the base reflect the expected impacts of program changes which are either anticipated to occur at some point in the future or have recently occurred and are not fully reflected in the base estimate. The combination of these two estimate components produces the final Genetic Disease Screening Program Estimate for the Newborn Screening Program (NBS) and the Prenatal Screening Program (PNS).

#### Genetic Disease Screening Program

GDSP provides screening of all newborns for genetic and congenital disorders that are preventable or remediable by early intervention. GDSP also provides screening of all pregnant women who consent to screening for serious birth defects. The screening programs provide public education, laboratory, and diagnostic clinical services through contracts with private vendors meeting state standards. The program is fully supported through fees collected from screening participants through the hospital of birth, third party payers, or private parties and are deposited into the Genetic Disease Testing Fund (GDTF). The Medi-Cal Program funds screening services for the eligible population.

#### **BASE ESTIMATE**

Actual caseload and expenditures for the prior year for both the newborn and prenatal screening programs are used to construct the base estimate and to establish trend data and adjustments to the base.

The base level for newborn screening workload is established as follows:

- Number of tests performed by contract laboratories X per test reimbursement.
- Number of reagent kits used X cost per kit.

- ❑ Number of tests requiring follow-up, referral, and counseling X cost of follow-up for these tests.
- ❑ Number of referrals to special centers for clinical diagnostic services X cost of follow-up at special centers.

The base level for prenatal screening workload is established as follows:

- ❑ Number of tests by contract laboratories X per test reimbursement.
- ❑ Number of reagent kits used X cost per kit.
- ❑ Number of tests requiring follow-up, referral, and counseling X cost of follow-up for these tests.
- ❑ Number of women referred to Prenatal Diagnostic Centers (PDC) X cost per PDC referral.

The base estimate is the anticipated level of program expenditures assuming there will be no changes in the program as approved in the Governor's Budget. The base estimate is adjusted by projected utilization rates and projected changes in the associated costs of contracts for the laboratory tests, follow up services, counseling, and diagnostic services. Any increased costs will be reflected in the fiscal estimates that follow.

Expenditures are those reflected in CALSTARS.

**ADJUSTMENTS TO THE LOCAL ASSISTANCE BASE****GDSP: NEW ASSUMPTIONS**

Applicable F/Y

C/Y B/Y

**X    No New Assumptions are proposed for the May 2011**  
**Revise.**

**GDSP: OLD ASSUMPTIONS**

Applicable F/Y

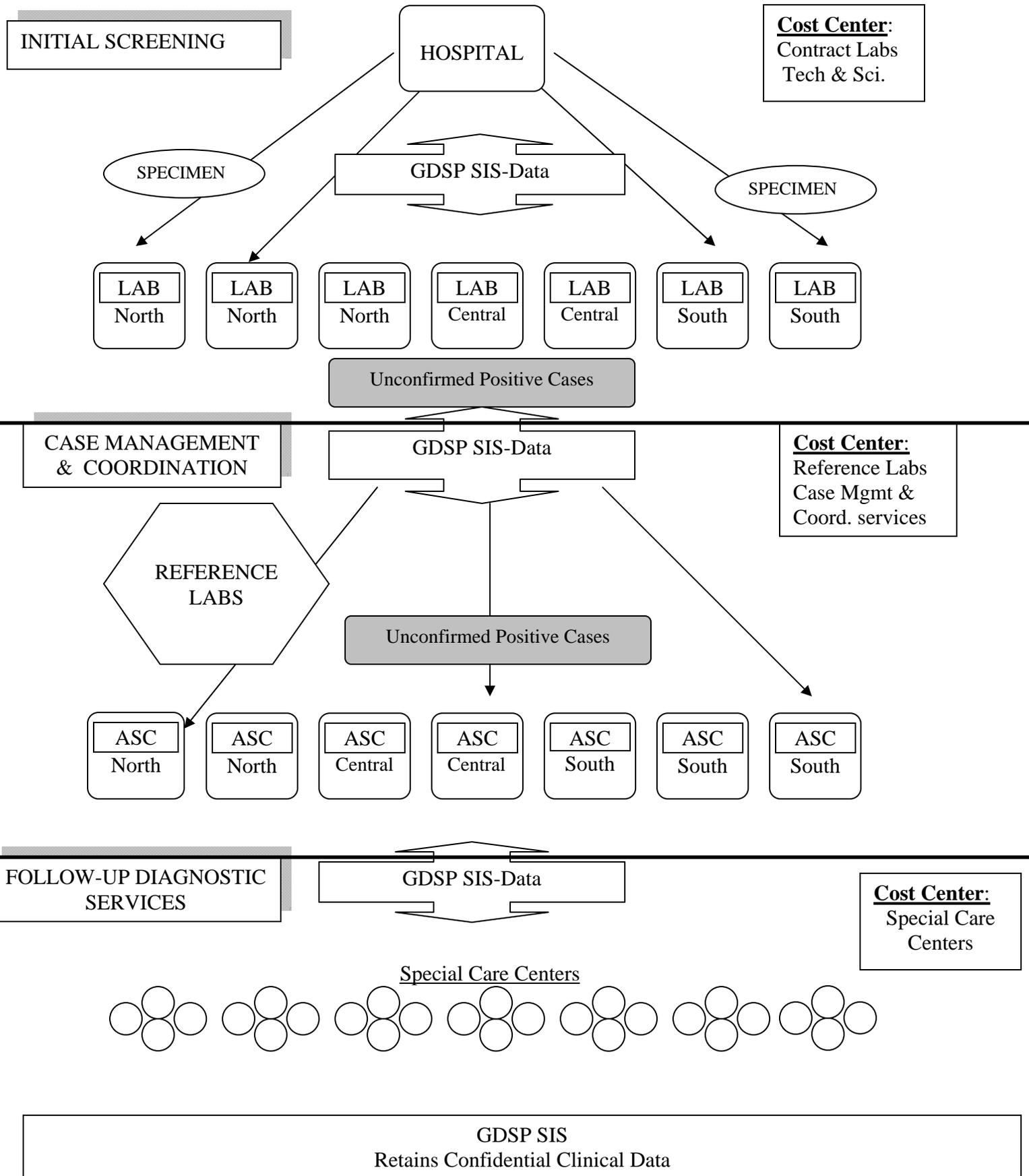
C/Y B/Y

1.    ✕            Prenatal Screening (PNS) Program Increased Costs:
- With the First Trimester Expansion, costs associated with providing additional testing, follow up, and diagnostic services are expected to increase. Additionally, with the First Trimester expansion, participation rates also are expected to increase, thus resulting in additional costs for testing, follow-up and diagnostic services. Currently, the PNS Program provides a blood screen and associated follow-up and diagnostic services for blood screening done in the Second Trimester of a pregnancy. With the addition of First Trimester, women will be able to receive screening services in both trimesters, including a second ultrasound during the First Trimester. Combining both screens will result in what is referred to as Integrated Screening, an approach that improves detection rates. (Expansion occurred April 2009).

**GDSP: INFORMATION ONLY**

1.                    Repayment of the General Fund Loan
- The GDSP fully repaid two prior General Fund loans during Fiscal Year 2009-10. The first repayment was in June 30, 2008. The second repayment was complete on June 30, 2010. The original loan amount was \$10.3 million; payment of the outstanding principal balance of \$4.24 million is reflected in the Fiscal Year 2009-10.

NEWBORN SCREENING PROGRAM



**BACKGROUND****THE GENETIC DISEASE SCREENING PROGRAM:  
NEWBORN SCREENING PROGRAM**

The mandatory Newborn Screening Program tests nearly every baby born in California for over 75 different congenital and genetic disorders. These disorders cause disability and even death if left undiagnosed and untreated.

**Contract Laboratories:**

- The newborn's blood sample (specimen) is collected at the hospital prior to discharge on special filter paper, dried, and mailed to a pre-assigned regional screening laboratory contracted by the State.
- Screening tests are carried out at seven (7) contract laboratories located throughout the State. Each specimen is subject to the same routine set of screening panels at all of the contract laboratories.
- Screening laboratories ascertain the *possibility* of a birth defect or a congenital disorder; a screening test is not diagnostic, therefore additional follow up may be required for a case that has an initial positive or a questionable screening test result.
- Each contract laboratory serves certain County jurisdiction with no duplication and all counties are served.
- Contract laboratories are compensated on a per screening panel set basis that is a contract negotiated rate and varies from laboratory to laboratory.
- Laboratory rates vary due to geographical lab locations, Union/non-Union laboratory agreements, as well as the volume of screens performed.

<b>COST CENTER:</b> Expenditures under <b>Contract Laboratories</b> reflect the cost of services performed by the contract laboratories to process initial specimen screening.
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**Technology & Scientific Supplies:**

- Screening for genetic abnormalities requires the use of testing reagents to analyze blood specimens.
- GDSP purchases and supplies reagents, test kits, chemicals and other supplies to the 7 contract laboratories, thereby securing best negotiated price based on large volume purchases.
  - GDSP approximates 3-5% of shelf life expiration, spills, and other wastage (varies depending on testing equipment and reagent type).
  - Laboratory standard of practice requires regular scheduled standardization of the test and the equipment (positive and negative controls, and spiked test specimens (unknowns) provided by GDSP, tested in contract laboratories under real conditions, and reported back to GDSP). This requires approximately 15-20% additional reagent use for standardization testing above and beyond routine specimen testing.
- Reagent costs vary depending on the type of screening performed. Purchase prices are actively negotiated to secure best value for the State.

- GDSP maintains inventories that can be used to supply the 7 contract laboratories in the event of unforeseen shortages.
- Additional costs associated with specimen screening include laboratory supplies (test tubes, pipettes, etc), blood specimen storage, as well as costs for special packaging for blood specimen transport.
- The Technology & Science budget also includes fixed costs such as limited maintenance and support of laboratory equipment provided to the seven contract laboratories for required repairs, maintenance and upgrades in the event the equipment can be serviced and full replacement may be avoided.

**COST CENTER:** Expenditures under **Technology & Scientific Supplies** reflect costs associated with reagents/supplies necessary to analyze blood specimens.

#### **System Project & Maintenance:**

- GDSP maintains a highly complex IT system, the Screening Information System (SIS), which is a web-based application that serves as a tracking mechanism of confidential clinical data for the NBS Program, as well as follow-up services for multiple statewide partners.
- Multiple technical resources are required to assist GDSP with ongoing maintenance and system operations.
- Support of GDSP's IT infrastructure is critical to Program operations; any technical disruptions may bring the Program to a halt.
- Acquisition of information technology projects may be reflected in this cost center.

**COST CENTER:** Expenditures in the System Project & Maintenance are for ongoing maintenance and operation costs for the existing IT infrastructure.

#### **Case Management and Coordination Services**

- Diagnosis, management, follow-up and counseling are critical components of the Program.
- Positive or equivocal results for newborns with inadequate or untimely specimens are reported to regional NBS Area Service Center (ASC) contractors, which are strategically located throughout the State within seven regions and are linked electronically to the NBS Program via the highly technical computer system, Screening Information System.
- The ASC Coordinators provide time critical case management so that short term follow-up is done as quickly as possible, sometimes within a life-threatening time frame.
- The ASC Coordinators are responsible for notifying the newborn's physician of all questionable results and tracking the cases until follow-up is completed and the case is either ruled out or transferred to a specialized treatment center.
- The ASC is composed of a core team of medical professionals and the cost for each ASC varies depending upon the geographical location as well as the range in volume of caseload served.

**COST CENTER:** Expenditures in **Case Management & Coordinating Services** reflect costs for a core team of clinical personnel.

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### **Reference Laboratories**

- When a *screening* test result is questionable or positive, the patient is referred for diagnostic testing at a confirmatory laboratory.
- Expert genetic diseases laboratories are contracted by GDSP to perform reference and confirmatory testing for screening positive or equivocal tests.
- Reference Laboratories are reimbursed on a per test compensation basis, with one laboratory doing all confirmatory testing for a particular genetic disorder.

**COST CENTER:** Expenditures in **Reference Laboratories** reflect costs associated with confirmatory diagnostic testing.

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### **Follow-up Diagnostic Services**

- Services are for infants with positive or questionable genetic screening results which require additional and extensive monitoring while undergoing confirmatory testing and diagnosis.
- Services are provided through Special Care Centers; these Centers are experts in the specific area of the genetic abnormality and cost is based on a per case reimbursement.
- Includes coordination with the NBS ASC as well as GDSP for ongoing medical care, establishment of infant treatment plans through specialty care hospitals and university medical centers specializing in a particular diagnoses, such as sickle cell anemia, cystic fibrosis, PKU, beta thalassemia, alpha thalassemia, and various neurologic, metabolic, endocrine and immune disorders.
- Multiple regional contracted specialty centers provide annual reporting for 5 years on diagnosed cases medically managed through the specialty center.
- Annual case data provided to GDSP on infants with a diagnosed disorder is a critical mechanism of tracking as well as confirmation, evaluation and refinement of program standards.

**COST CENTER:** Expenditures in **Follow-up Diagnostic Services** reflect costs for services on a per case reimbursement.

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### **Result Reporting & Fee Collection:**

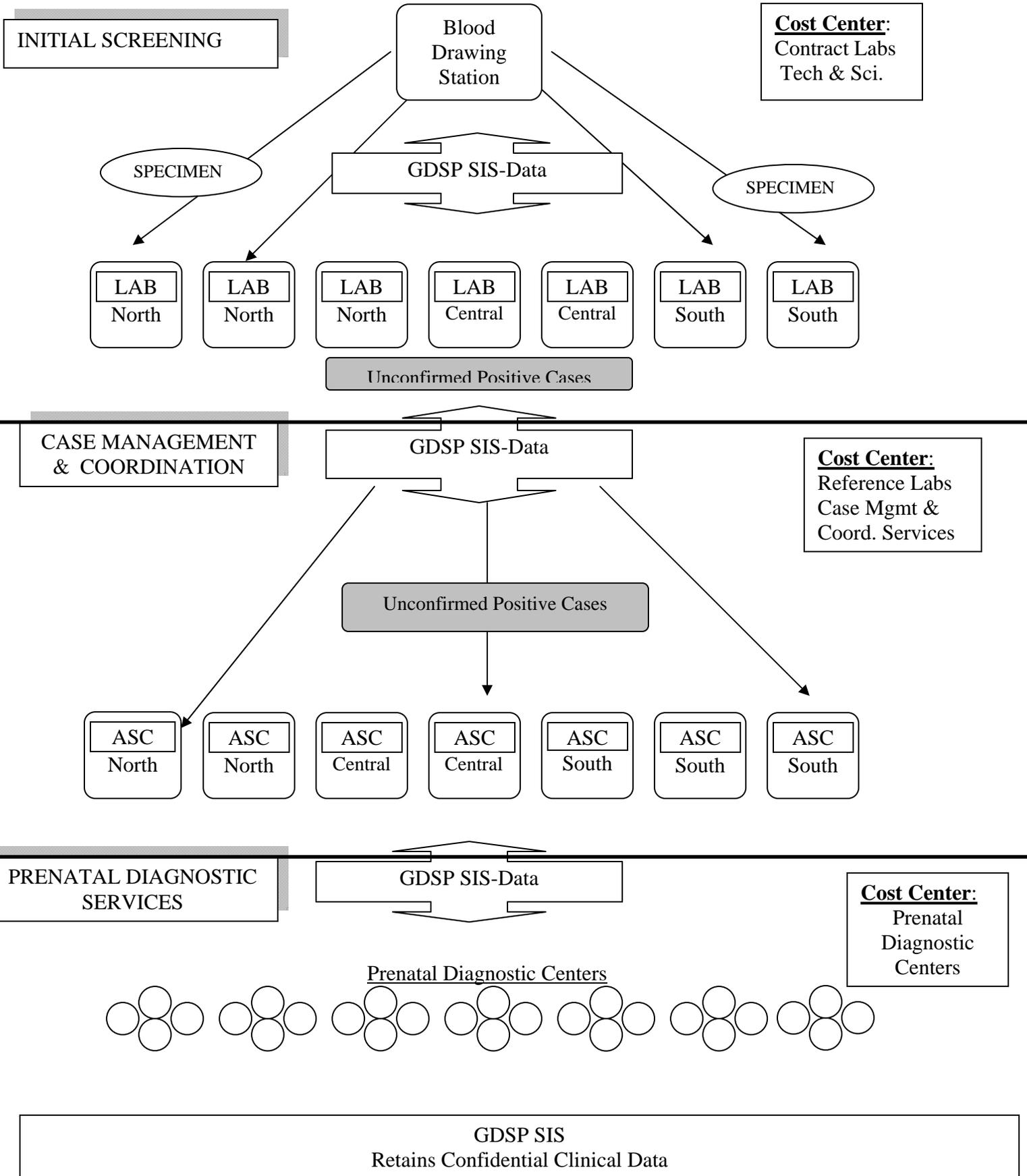
- Production expenses associated with communicating results of genetic screens, educational materials, etc. For example, a report of each baby's initial test results, called the Newborn Screening Results Mailer, is mailed to the hospital that drew the specimen as well as to the newborn's physician. If the initial screening test is positive, or if the sample is not adequate for testing, the mailer provides information on follow-up procedures.
- Costs associated with tracking and processing revenue from hospitals and other birthing locations.
- Educational materials developed by the Program and distributed through health care clinics which provides practical information and support to parents. Materials are

available at no cost to health care providers, hospitals, clinics and local health departments.

- Costs related to invoicing and collecting payment from the hospitals and birthing facilities is categorized under this line item.

**COST CENTER:** Expenditures in **Result Reporting & Fee Collection** reflect costs for production of follow-up material to medical providers and families of children as well as costs related for the collection of payments.

PRENATAL SCREENING PROGRAM



**BACKGROUND****THE GENETIC DISEASE SCREENING PROGRAM:  
PRENATAL SCREENING PROGRAM**

The Prenatal Screening Program (PNS) screens for birth defects during pregnancy and provides risk assessment and follow-up services to all pregnant women in California.

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**Contract Laboratories:**

- GDSP contracts with seven (7) screening laboratories located throughout the State. Screening laboratories ascertain the *possibility* of a birth defect or a congenital disorder; a screening test is not diagnostic, therefore additional follow up may be required for a case that has an initial positive or a questionable screening test result.
- Laboratory testing screens pregnancies for genetic and congenital disorders, such as Trisomy 21, Trisomy 18, Smith-Lemli-Opitz Syndrome (SLOS) and Neural Tube Defects.
  - **1<sup>st</sup> Trimester Screening:** Includes testing of human chorionic gonadotropin (HCG) and pregnancy-associated plasma protein A (PAPP-A).
  - **2<sup>nd</sup> Trimester Screening:** Includes testing of HCG, alpha-fetoprotein (AFP), unconjugated estriol (uE3) and Inhibin.
- Contract laboratories are compensated on a per screening panel set basis that is a contract negotiated rate and varies from laboratory to laboratory as well as between 1<sup>st</sup> and 2<sup>nd</sup> Trimester screens.
- Each contract laboratory serves certain County jurisdiction with no duplication and all counties are served.
- Laboratory rates vary due to fluctuations in geographical areas, Union/non-Union laboratory agreements as well as the volume of screens performed.

<b>COST CENTER:</b> Expenditures under <b>Contract Laboratories</b> reflect the cost of services performed by the contract laboratories to process initial specimen screening.
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**Technology & Scientific Supplies:**

- Screening for genetic abnormalities requires the use of testing reagents to analyze blood specimens.
- GDSP purchases and supplies reagents, test kits, chemicals and other supplies to the 7 contract laboratories, thereby securing best negotiated price based on large volume purchases.

- GDSP approximates 3-5 % of shelf life expiration, spills, and other wastage (varies depending on testing equipment and reagent type).
- Laboratory standard of practice requires regular scheduled standardization of the test and the equipment (positive and negative controls, and spiked test specimens (unknowns) provided by GDSP, tested in contract laboratories under real conditions, and reported back to GDSP). This requires approximately 15-20% additional reagent use for standardization testing above and beyond routine specimen testing.
- Reagent costs vary depending on the type of screening performed. Purchase prices are actively negotiated to secure best value for the State.
- GDSP maintains inventories that can be used to supply the 7 contract laboratories in the event of unforeseen shortages.
- Additional costs associated with specimen screening include blood specimen tubes and laboratory supplies, blood specimen storage as well as costs for special packaging for blood specimen transport
- The Technology & Science budget also includes fixed costs such as limited maintenance and support of laboratory equipment provided to the 7 contract laboratories for required repairs, maintenance and upgrades in the event the equipment can be serviced and full replacement may be avoided.

**COST CENTER:** Expenditures under **Technology & Scientific Supplies** reflect costs associated with reagents/supplies necessary to analyze blood specimens.

#### **System Project & Maintenance:**

- GDSP maintains a highly complex IT system, the Screening Information System (SIS), which is a web-based application that serves as a tracking mechanism of confidential clinical data for the PNS Program, as well as follow-up services for multiple statewide partners.
- Multiple technical resources are required to assist GDSP with ongoing maintenance and system operations.
- Support of GDSP's IT infrastructure is critical to Program operations; any technical disruptions may bring the Program to a halt.
- Acquisition of information technology projects may be reflected in this cost center.

**COST CENTER:** Expenditures in the System Project & Maintenance are for ongoing maintenance and operation costs for the existing IT infrastructure.

#### **Case Management and Coordination Services**

- Diagnosis, management, follow-up and counseling are critical components of the Program.
- Services are provided by GDSP Area Service Centers (ASC) Coordinators to pregnant women include coordination of First and Second Trimester screens and NT Ultrasounds, identification of patients whose blood specimens was drawn too early or was inadequate and requires additional blood draws.
- Coordination and consultation with patient's physician and specialty care providers is done at this level.

- The ASC Coordinators provide clinician and patient education and consultations; make referrals to Prenatal Diagnostic Centers for confirmatory tests, provide some genetic counseling and track patients to ensure appointments are kept and patients are seen within prescribed timeframes.
- ASC contractors have projected caseloads based on the expected positive rates for various genetic screens for the population tested.
- The ASC is composed of a core team of medical professionals and the cost for each ASC varies depending upon the geographical location as well as the range in volume of caseload served.

**COST CENTER:** Expenditures in **Case Management & Coordinating Services** reflect costs for a core team of clinical personnel.

#### **Prenatal Diagnostic Services:**

- When a PNS screening test is positive, diagnostic services are offered at a State-approved Prenatal Diagnostic Centers (PDC).
- PDCs are composed of a core team of medical professionals and the cost for each PDC varies depending upon the geographical location as well as the volume in caseload served.
- Diagnostic services, such as comprehensive genetic counseling, Chronic Villus Sampling, Ultrasound, Amniocentesis, etc., are provided to women with positive results as a method of ruling out the estimated chance of a birth defect.
- The PDCs are reimbursed on the basis of services performed.

**COST CENTER:** Expenditures in **Prenatal Diagnostic Services** reflect costs for services performed for pregnant women with screen positive test results.

#### **Result Reporting & Fee Collection:**

- Production expenses associated with communicating results of the prenatal screens and educational materials. If the initial screening test is positive, or if the sample is not adequate for testing, a GDSP mailer provides detailed information on follow-up procedures.
- Costs associated with tracking and processing payment from women that participated in the Program.
- Educational materials developed by the Program and distributed through health care clinics provide practical information and guidance. Materials are available at no cost to health care providers, hospitals, clinics and local health departments.
- Costs related to invoicing and collecting payment from the hospitals and birthing facilities is also categorized under this line item.

**COST CENTER:** Expenditures in **Result Reporting & Fee Collection** reflect costs for production of follow-up material as well as resources for payment collection.

**TABLES 1 and 2: CALIFORNIA BIRTHS BY AGE OF MOTHER and AGE-SPECIFIC FERTILITY RATES**

**TABLE 1: CALIFORNIA BIRTHS BY AGE OF MOTHER**

	CALIFORNIA BIRTHS BY AGE OF MOTHER					Total	AGE-SPECIFIC FERTILITY RATES					TFR	
	Age 15-19	Age 20-24	Age 25-29	Age 30-34	Age 35-39		Age 40-44	Age 15-19	Age 20-24	Age 25-29	Age 30-34		Age 35-39
1990	70,951	159,405	183,221	133,423	54,471	611,666	70.77	134.71	134.91	96.93	44.26	9.48	2.46
1991	71,793	158,779	177,685	133,192	56,654	609,228	72.34	134.14	133.85	95.77	44.84	9.90	2.45
1992	70,867	155,065	171,429	133,205	58,660	600,838	70.20	131.42	131.12	95.59	45.08	10.22	2.42
1993	70,091	149,047	163,372	131,438	58,505	584,463	68.55	127.64	128.41	94.72	44.37	10.45	2.37
1994	69,885	140,172	154,779	129,926	59,550	567,034	67.10	122.29	124.27	94.15	44.87	10.87	2.32
1995	68,284	132,607	148,653	127,853	60,577	551,226	64.47	118.38	120.18	94.15	45.38	11.15	2.27
1996	64,603	127,431	145,885	125,030	61,836	538,628	59.62	115.39	117.08	93.94	45.99	11.41	2.22
1997	61,107	122,924	141,259	121,938	62,674	524,174	54.94	110.27	112.19	92.51	46.20	11.44	2.14
1998	59,207	121,317	140,418	121,326	64,210	521,265	51.83	107.90	110.77	92.91	47.05	11.59	2.11
1999	57,615	120,270	137,701	121,779	65,532	518,073	49.38	105.94	109.13	93.75	47.33	11.67	2.09
2000	56,273	122,604	139,629	127,516	68,693	531,285	47.41	107.80	114.08	97.24	49.52	12.35	2.14
2001	53,779	123,236	136,449	127,957	68,835	527,371	44.40	107.56	114.03	95.92	49.66	12.44	2.12
2002	50,947	123,065	137,250	130,379	69,879	529,245	41.32	106.17	116.86	97.26	50.62	12.64	2.12
2003	50,042	123,822	140,566	134,819	72,669	540,828	39.63	104.56	121.50	101.05	52.85	13.28	2.16
2004	50,436	124,318	141,621	134,592	74,589	544,685	38.77	102.71	123.29	102.64	54.41	13.26	2.18
2005	50,777	125,541	143,463	133,760	75,740	548,700	37.75	101.66	125.00	104.93	55.01	13.48	2.19
2006	53,455	129,153	148,287	133,462	77,793	562,157	38.32	102.18	127.12	107.25	56.12	14.03	2.23
2007	54,060	127,996	150,523	135,376	78,453	566,137	37.57	99.27	126.34	110.40	56.66	13.99	2.22
2008	52,332	122,281	147,071	132,616	76,962	551,567	35.59	92.70	120.09	109.32	56.08	14.55	2.14
2009	48,362	113,942	140,972	129,089	74,488	526,774	32.50	83.94	112.25	106.75	55.29	14.38	2.03
Projection:													
2010	46,841	116,013	140,613	129,899	73,030	527,059	31.47	82.67	109.54	107.15	55.64	14.86	2.01
2011	46,962	119,652	140,797	133,584	71,682	533,775	31.81	82.23	107.21	108.58	55.87	15.05	2.00
2012	45,514	123,466	142,334	138,227	71,119	541,978	31.31	82.23	106.06	109.94	56.19	15.22	2.00
2013	45,054	126,443	145,154	143,528	70,465	552,251	31.59	82.35	105.55	111.00	56.20	15.56	2.01
2014	43,947	126,937	148,227	148,546	70,681	559,520	31.39	81.60	104.62	111.94	56.49	15.53	2.01
2015	43,107	126,366	151,960	153,202	71,162	567,020	31.25	81.11	103.67	112.82	56.67	15.96	2.01
2016	42,843	123,109	155,802	157,284	72,802	572,415	31.03	79.65	102.69	113.36	57.16	15.81	2.00
2017	42,922	119,837	158,452	163,082	75,493	580,583	30.93	78.70	101.34	115.16	58.04	16.20	2.00
2018	43,451	116,058	160,263	167,701	78,682	586,878	30.95	77.60	100.32	115.71	58.88	16.29	2.00
2019	44,121	112,358	159,579	173,868	81,425	592,256	31.04	76.46	98.64	116.62	59.42	16.47	1.99

Source: Historical births through 2009, California Department of Public Health, Center for Health Statistics.

Projected births, California Department of Finance, Demographic Research Unit.

Rounding: Independent rounding may prevent the sum of selected data components from exactly matching the total.