

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



CALIFORNIA
DEPARTMENT OF PUBLIC HEALTH

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DEPARTMENT OF FINANCE DEMOGRAPHIC DATA

Management Summary

The Genetic Disease Screening Program (GDSP) November Estimate includes adjustments in caseload, expenditures, and revenues for the Genetic Disease Testing Fund.

Several factors impact the GDSP Estimate in both the current year and budget year. Historically, the birthrate in California has been on a steady incline; however the birthrate began declining during the later part of 2009. The current economic climate is suspected to have had a significant impact on births. Evaluation and review of caseload data done internally during the prior fiscal year (FY 2009-10) noted the decline; GDSP was unable to implement changes to its birth projections before confirming with external sources, such as the updated Department of Finance Birth projections, released at the end of September 2010.

Offsetting the decrease in birthrates, GDSP is experiencing increased participation in the Prenatal Screening Program (PNS) as more pregnant women become aware of the benefits of 1st Trimester screening and the PNS program continues to grow. Furthermore, the full repayment of the General Fund loan in FY 2009-10 left the program with a less than favorable start for the current fiscal year and created pressure on GDSP's reserves. Consequently, the effect of these factors impacts current year and also extends into the budget year.

Current Year (CY) 2010-11

The November 2010 Estimate projects \$93.2 million in local assistance expenditures in the current year, a 2.1 percent decrease over the Budget Act 2010 appropriation of \$95.2 million. The department projects the total GDSP caseload in the current year to be 986,013 cases, a 2.6-percent decrease from the prior approved forecast of 1,012,111.

2010-11 Adjustments

Newborn Screening (NBS) Program – Local Assistance

The November 2010 Estimate projects the Newborn Screening (NBS) caseload to decrease by 29,304 or 5.2 percent to 532,980 from the prior approved caseload projection of 562,284 thus resulting in a net reduction of \$803,000 or 1.8 percent to the NBS budget.

In order to provide more detail in the fiscal display, some categories have been renamed and/or split into separate categories as explained in the narrative section of the Estimate.

Several budget reductions and shifts taken across the NBS program are reflected in the following adjustments:

Contract Laboratories:

A \$572,000 reduction due to the decrease in NBS caseload and associated costs for processing laboratory specimens; \$116,000 of the \$572,000 is shifted to Reference Laboratories as it is the appropriate cost center for the specified laboratory funding.

Technical and Scientific:

A \$347,000 reduction for expenses such as reagent kits and associated costs, resulting from the decrease in NBS caseload.

System Project & Maintenance:

(Previously labeled as *Systems Dedicated Equipment, System Development & Maintenance*) A reduction of \$234,000 is shifted to the Reference Laboratories cost center.

Case Management & Coordination Services:

Previously, this category was labeled 'Follow-up Costs' as all costs associated with following-up, tracking and diagnostic services were at this cost center. This line item has now been more appropriately identified and split into two categories of services: *Case Management & Coordination Services* and *Reference Laboratories*, which resulted in a shift of \$1,334,000 to the new Reference Laboratories cost center.

Reference Laboratories:

A new cost center budgeted at \$2,550,000 reflects funding that was previously under several categories: \$116,000 from Contract Laboratories, \$1,334,000 from Case Management and Coordination Services, \$866,000 from Follow-up Diagnostic Services and \$234,000 as a budgetary shift from System Project & Maintenance cost center make up this budget.

Follow-up Diagnostic Services:

A shift of \$866,000 of Reference Laboratories funding is transferred to its appropriate cost center, as a portion was previously budgeted in Follow-up Diagnostic Services.

Prenatal Screening (PNS) Program – Local Assistance

The November 2010 Estimate projects the Prenatal Screening (PNS) caseload to slightly increase by 3,206 or less than 1 percent to 453,033 from the prior approved caseload projection of 449,827. As a result of the 1st Trimester expansion, more women are participating in the program than previously expected. Although the NBS caseload decreased, the PNS program is experiencing unforeseen growth, reaching to an expected 85 percent participation rate. Budget line item shifts as well as minor reductions result in a net decrease of \$1,175,000 or approximately 2.4 percent to the PNS budget as the program concludes its initial expansion stage and commences operations at the new level.

Several budget reductions and shifts taken across the PNS program are reflected in the following adjustments:

Contract Laboratories:

A reduction of \$75,000 to the contract laboratories budget is reflected in the current year.

System Project & Maintenance:

(Previously labeled as *Systems Dedicated Equipment, System Development & Maintenance*). As the expansion stage of the 1st Trimester roll-out begins to subside and the internal state-wide IT infrastructure is updated to collect, retain and process the inflow of new data associated with new screening, a reduction of \$1,682,000 to System Project & Maintenance is maintained; \$632,000 of this funding is shifted to the Result Reporting & Fee Collection cost center.

Prenatal Diagnostic Services:

A reduction of \$50,000 the Prenatal Diagnostic Centers is reflected in the current year.

Result Reporting & Fee Collection:

The initial stages of the 1st Trimester expansion begin to subside and the PNS program ramps up to reach more participants, it becomes evident that the Result Reporting & Fee Collection cost center is lacking sufficient funding to keep up with the increased number of women accessing the program for prenatal testing. A funding shift from the System Project & Maintenance of \$632,000 is added to this cost center.

Budget Year (BY) 2011-12

The November 2010 Estimate projects \$94.0 million in local assistance expenditures in the budget year, a 1.3 percent decrease over the Budget Act 2010 appropriation of \$95.2 million. The department projects GDSP's total caseload in the budget year to be 999,818 cases, a 1.4 percent increase from the current year caseload forecast of 986,013.

2011-12 Adjustments**Newborn Screening (NBS) Program – Local Assistance**

In comparison to the caseload projection for the current year of 532,980, the November 2010 Estimate projects the NBS budget year caseload to increase by 7,462 or 1.4 percent to 540,442, thus resulting in a net budget increase of \$346,000 compared to the current year NBS projected expenditures.

Several budget reductions and shifts taken across the NBS program are reflected in the following adjustments

Contract Laboratories:

A \$252,000 reduction from Budget Act 2010 appropriation due to the revised NBS caseload as well as associated increases in costs for processing laboratory specimens.

Technical and Scientific:

A \$332,000 reduction for expenses such as reagent kits and associated costs, resulting from the decrease in NBS caseload to the previously approved in the Budget Act 2010.

System Project & Maintenance:

(Previously labeled as *Systems Dedicated Equipment, System Development & Maintenance*) A reduction of \$449,000; \$366,000 is shifted to the Reference Laboratories cost center.

Case Management & Coordination Services:

Previously, this category was labeled 'Follow-up Costs' as all costs associated with following-up, tracking and diagnostic services were at this cost center. This line item has now been more appropriately identified and split into two categories of services: Case Management & Coordination Services and Reference Laboratories, which resulted in a shift of \$1,259,000 to the Reference Laboratories cost center.

Reference Laboratories:

A newly identified cost center budgeted at \$2,491,000 reflects funding that was previously under several categories: \$1,259,000 from Case Management and Coordination Services, \$866,000 from Follow-up Diagnostic Services, \$366,000 as a budgetary shift from System Project & Maintenance cost centers make up this budget.

Follow-up Diagnostic Services:

A shift of \$866,000 of Reference Laboratories funding is transferred to its appropriate cost center, as a portion was previously budgeted in the Follow-up Diagnostic Services.

Result Reporting & Fee Collection:

An increase of \$210,000 is requested to meet production expenses and make this cost center whole.

Prenatal Screening (PNS) Program – Local Assistance

In comparison to the caseload projection for the current year, the November 2010 Estimate projects the Prenatal Screening (PNS) caseload to increase by 6,343 or approximately 1.4 percent to 459,376 from the caseload projection of 453,033 in the current year. As a result of the 1st Trimester expansion, more women are participating in the program than previously expected. Although the NBS caseload decreased, the PNS program is experiencing unforeseen growth, reaching to an expected 85 percent participation rate.

Several budget line item shifts as well as minor reductions result in a net increase of \$428,000 to the PNS budget compared to the current year PNS projected expenditures.

Contract Laboratories:

A \$32,000 increase to the Budget Act 2010 appropriation due to the increase in the PNS caseload as well as associated increases in costs for processing laboratory specimens.

Technical and Scientific:

A \$154,000 increase for expenses such as reagent kits and associated costs, resulting from the increase in the PNS caseload.

System Project & Maintenance:

As the expansion phase of the 1st Trimester roll-out begins to subside and the internal state-wide IT infrastructure is updated to collect, retain and process the inflow of new data associated with new screening, a reduction of \$1,682,000 to System Project & Maintenance is maintained; several funding shifts from this category are utilized for other cost centers in the prenatal program.

Case Management & Coordination Services:

An increase of \$132,000 to support clinical teams across the state that provide case management as part of the PNS program.

Prenatal Diagnostic Services

A reduction of \$15,000 the Prenatal Diagnostic Centers is reflected in the budget year.

Result Reporting & Fee Collection:

The initial stages of the 1st Trimester expansion begin to subside and the PNS program ramps up to reach more participants, it becomes evident that the Result Reporting & Fee Collection cost center is lacking sufficient funding to keep up with the increased number of women accessing the program for prenatal testing. A funding shift from the System Project & Maintenance of \$632,000 is added to this cost center.

GENETIC DISEASE SCREENING PROGRAM NOV 2010 BUDGET ESTIMATE		2010-2011			2011-2012	
PROGRAM	2010 BUDGET ACT	REVISED 2010-2011 ESTIMATE	DIFFERENCE FROM APPROPRIATION	2010 BUDGET ACT	PROPOSED 2011-2012 GOVERNOR'S BUDGET	DIFFERENCE FROM APPROPRIATION
LOCAL ASSISTANCE						
NBS						
Contract Laboratories:	\$7,429,000	\$6,857,000	(\$572,000)	\$7,429,000	\$7,177,000	(\$252,000)
Tech & Sci:	\$23,497,000	\$23,150,000	(\$347,000)	\$23,497,000	\$23,165,000	(\$332,000)
System Project & Maintenance:	\$4,222,000	\$3,988,000	(\$234,000)	\$4,222,000	\$3,773,000	(\$449,000)
Case Management & Coordination Services:	\$5,834,000	\$4,500,000	(\$1,334,000)	\$5,834,000	\$4,575,000	(\$1,259,000)
Reference Laboratories:	\$3,366,000	\$2,550,000	\$2,550,000	\$3,366,000	\$2,491,000	\$2,491,000
Follow-up Diagnostic Services:	\$1,290,000	\$2,500,000	(\$866,000)	\$1,290,000	\$2,500,000	(\$866,000)
Result Reporting & Fee Collection	\$1,290,000	\$1,290,000	\$0	\$1,290,000	\$1,500,000	\$210,000
	\$45,638,000	\$44,835,000	(\$803,000)	\$45,638,000	\$45,181,000	(\$457,000)
PNS						
Contract Laboratories:	\$5,090,000	\$5,015,000	(\$75,000)	\$5,090,000	\$5,122,000	\$32,000
Tech & Sci:	\$13,146,000	\$13,146,000	\$0	\$13,146,000	\$13,300,000	\$154,000
System Project & Maintenance:	\$6,485,000	\$4,803,000	(\$1,682,000)	\$6,485,000	\$4,803,000	(\$1,682,000)
Case Management & Coordination Services:	\$6,110,000	\$6,110,000	\$0	\$6,110,000	\$6,242,000	\$132,000
Prenatal Diagnostic Services:	\$17,426,000	\$17,376,000	(\$50,000)	\$17,426,000	\$17,411,000	(\$15,000)
Result Reporting & Fee Collection	\$1,310,000	\$1,942,000	\$632,000	\$1,310,000	\$1,942,000	\$632,000
	\$49,567,000	\$48,392,000	(\$1,175,000)	\$49,567,000	\$48,820,000	(\$747,000)
LOCAL ASSISTANCE, TOTAL	\$95,205,000	\$93,227,000	(\$1,978,000)	\$95,205,000	\$94,001,000	(\$1,204,000)

**Genetic Disease Screening Program – Newborn Screening Testing
BUDGET DETAIL – November 2010**

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 – November 2010

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.

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

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

November 2010
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 November 2010 Estimate.**GDSP: OLD ASSUMPTIONS**Applicable F/Y
C/Y B/Y

1. X Prenatal Screening (PNS) Program Increased Costs:

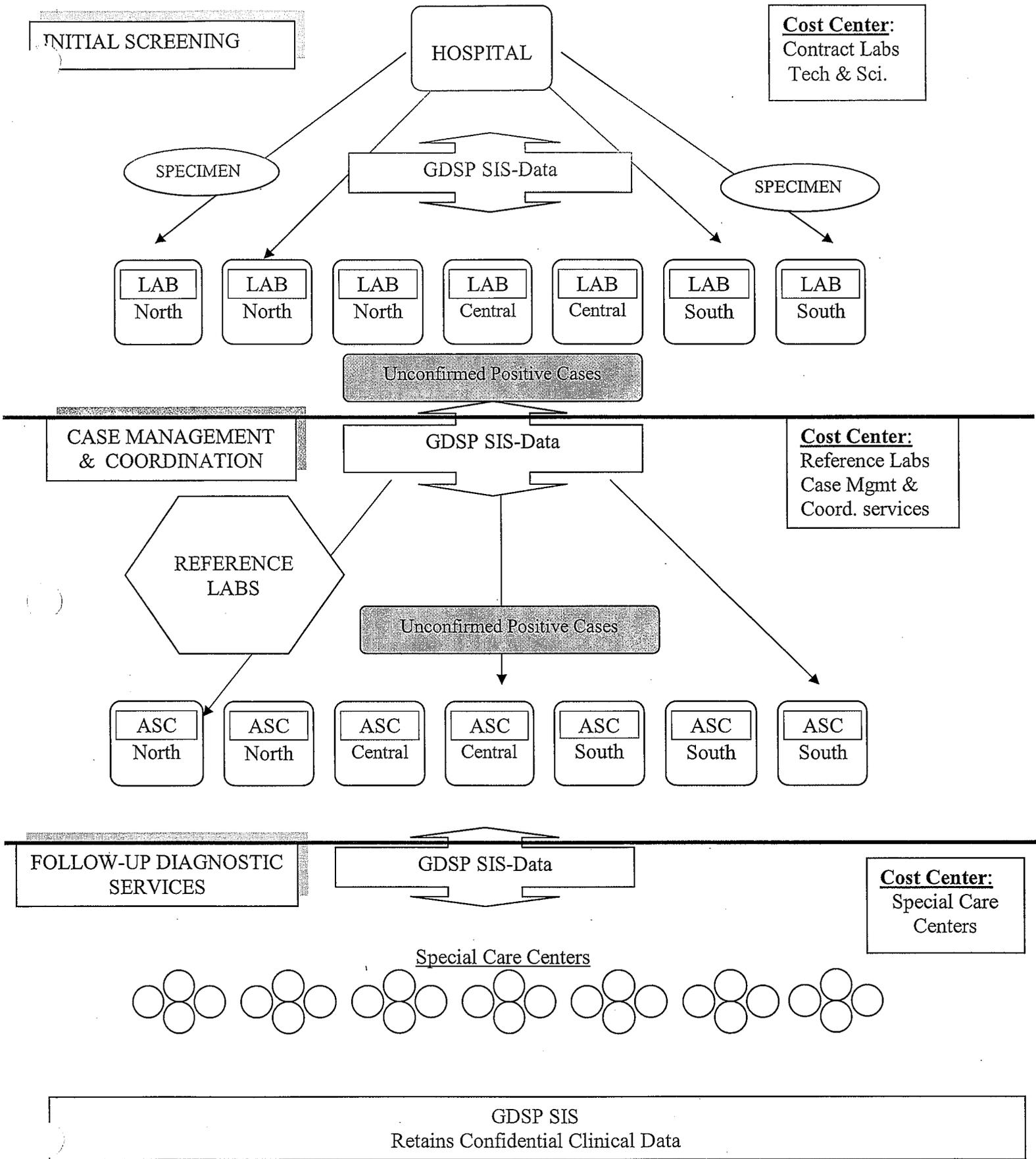
With the First Trimester Expansion, costs associated with providing additional testing, follow up, and diagnostic services are expected to increase. Additionally, with the First Trimester expansion, participation rates also are expected to increase, thus resulting in additional costs for testing, follow-up 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.

COST CENTER: Expenditures under **Contract Laboratories** reflect the cost of services performed by the contract laboratories to process initial specimen screening.

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.

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.

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.

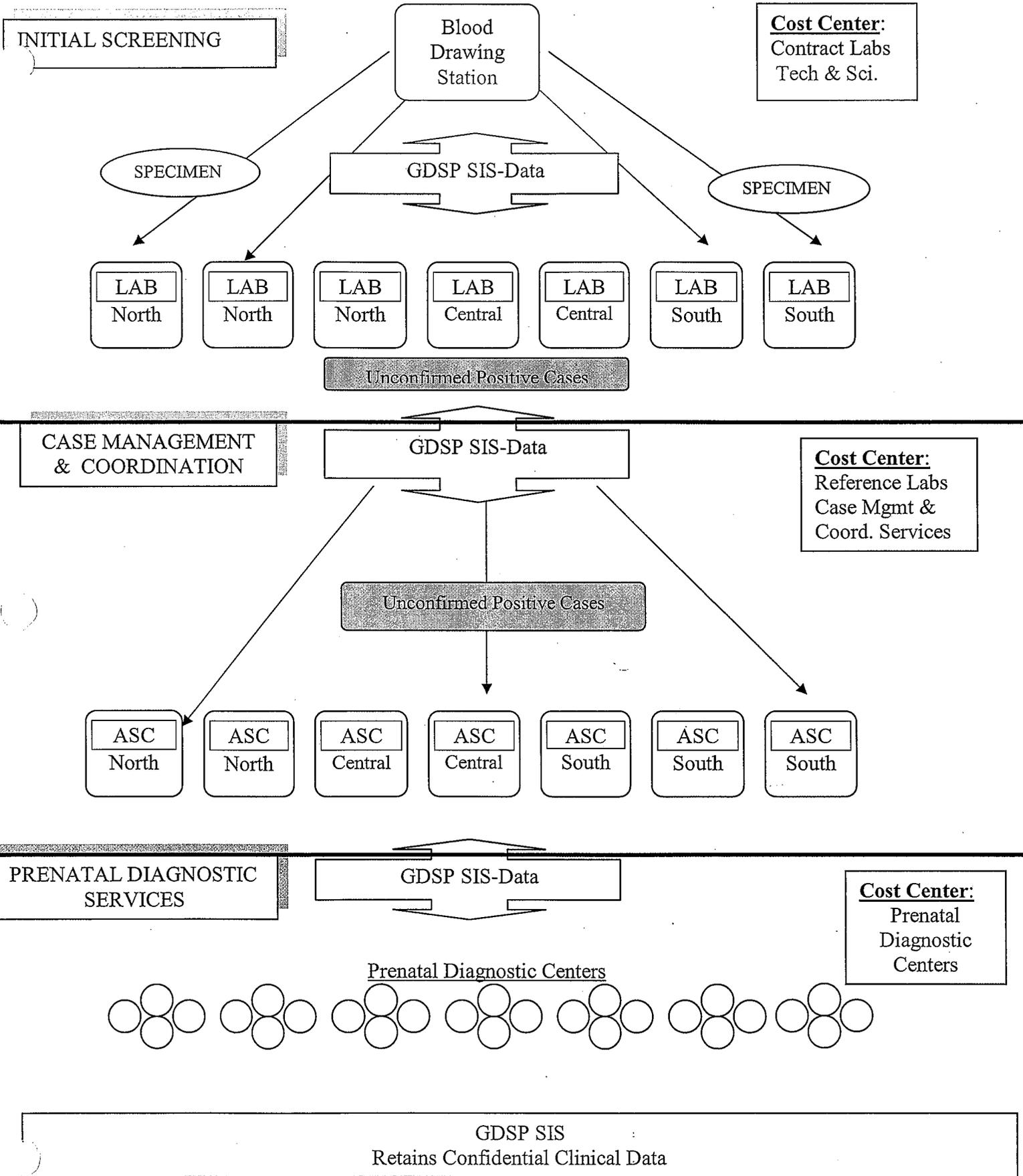
COST CENTER: Expenditures in **Follow-up Diagnostic Services** reflect costs for services on a per case reimbursement.

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.

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.

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.
 - **1st Trimester Screening:** Includes testing of human chorionic gonadotropin (HCG) and pregnancy-associated plasma protein A (PAPP-A).
 - **2nd 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 1st and 2nd 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.

COST CENTER: Expenditures under **Contract Laboratories** reflect the cost of services performed by the contract laboratories to process initial specimen screening.

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 time frames.

- 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						TABLE 2: AGE-SPECIFIC FERTILITY RATES							
	Age 15-19	Age 20-24	Age 25-29	Age 30-34	Age 35-39	Age 40-44	Total	Age 15-19	Age 20-24	Age 25-29	Age 30-34	Age 35-39	Age 40-44	TFR
1990	70,951	159,405	183,221	133,423	54,471	10,195	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	11,125	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	11,612	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	12,030	584,483	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	12,722	567,034	67.10	122.29	124.27	94.15	44.87	10.87	2.32
1995	68,284	132,607	148,663	127,853	60,577	13,252	551,226	64.47	118.38	120.18	94.15	45.38	11.15	2.27
1996	64,903	127,431	145,885	125,030	61,836	13,843	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	14,272	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	14,787	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	15,176	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	16,570	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	17,117	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	17,725	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	18,910	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	19,129	544,685	38.75	102.71	123.29	102.64	54.41	13.26	2.18
2005	50,777	125,541	143,463	133,760	75,740	19,418	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	20,007	562,157	38.32	102.18	127.12	107.25	56.12	14.03	2.23
2007	54,060	127,996	150,923	135,376	78,453	19,729	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	20,304	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	19,922	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	20,673	527,069	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	21,098	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	21,318	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	21,606	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	21,181	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	21,223	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	20,575	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	20,798	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	20,724	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	20,905	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.