

GENETIC DISEASE SCREENING PROGRAM
November 2011
ESTIMATE
for
FISCAL YEARS
2011-12 *and* 2012-13



CALIFORNIA
DEPARTMENT OF PUBLIC HEALTH

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Management Summary

The Genetic Disease Screening Program (GDSP) November 2011 Estimate aligns the Current Year (FY 2011-12) and Budget Year (FY 2012-13) budget in accordance to a reduced newborn caseload. Birth rates are projected to remain flat at a marginal .5% growth in FY 2012-13.

INFORMATION ONLY

In accordance with the legislative mandate pursuant to Chapter 461, Statutes of 2011 (hereafter referred to as Assembly Bill 395) CDPH must add Severe Combined Immunodeficiency (SCID) to the panel of disorders screened for by the GDSP Newborn Screening Program (NBS). GDSP would also be required to screen for related T-cell lymphopenias, provided there is no additional cost. By incorporating SCID screening into the newborn panel, California will meet the current national standard of care as recommended by the federal Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC).

SCID is considered to be one of the most serious primary immunodeficiency disorders. The defining characteristic is the absence of T-cells and, as a result, lack of B-cell function, the specialized white blood cells made in the bone marrow to fight infection. These genetic defects lead to extreme susceptibility to serious illness. Without appropriate treatment, babies require expensive medical treatments and still suffer from chronic lifelong conditions. Early detection of SCID by newborn screening can minimize and even prevent the medical complications as well as the financial burden to the family and the public health-care system. Screening for SCID is expected to begin January 1, 2012.

BUDGET ESTIMATE OVERVIEW

GENETIC DISEASE SCREENING PROGRAM
November 2011 Estimate

GENETIC DISEASE SCREENING PROGRAM NOV 2011 BUDGET ESTIMATE	2011-2012			2012-2013		
	BUDGET ACT	REVISED November 2011 Estimate	DIFFERENCE	BUDGET ACT	REVISED November 2011 Estimate	DIFFERENCE
LOCAL ASSISTANCE						
NBS						
Contract Laboratories:	\$7,177,000	\$7,094,000	(\$83,000)	\$7,177,000	\$7,206,000	\$29,000
Tech & Sci:	\$23,165,000	\$21,912,000	(\$1,253,000)	\$23,165,000	\$21,077,000	(\$2,088,000)
System Project & Maintenance:	\$3,773,000	\$2,873,000	(\$900,000)	\$3,773,000	\$2,864,000	(\$909,000)
Case Management & Coordination Services:	\$4,575,000	\$4,436,000	(\$139,000)	\$4,575,000	\$4,478,000	(\$97,000)
Reference Laboratories:	\$2,491,000	\$2,456,000	(\$35,000)	\$2,491,000	\$2,456,000	(\$35,000)
Follow-up Diagnostic Services:	\$2,500,000	\$2,279,000	(\$221,000)	\$2,500,000	\$2,279,000	(\$221,000)
Result Reporting & Fee Collection	\$1,500,000	\$1,500,000	\$0	\$1,500,000	\$1,500,000	\$0
	\$45,181,000	\$42,550,000	(\$2,631,000)	\$45,181,000	\$41,860,000	(\$3,321,000)
PNS						
Contract Laboratories:	\$5,122,000	\$4,655,000	(\$467,000)	\$5,122,000	\$4,860,000	(\$262,000)
Tech & Sci:	\$13,300,000	\$13,029,000	(\$271,000)	\$13,300,000	\$13,136,000	(\$164,000)
System Project & Maintenance:	\$4,803,000	\$4,264,000	(\$539,000)	\$4,803,000	\$2,955,000	(\$1,848,000)
Case Management & Coordination Services:	\$6,242,000	\$5,593,000	(\$649,000)	\$6,242,000	\$5,761,000	(\$481,000)
Prenatal Diagnostic Services:	\$17,411,000	\$16,735,000	(\$676,000)	\$17,411,000	\$17,237,000	(\$174,000)
Result Reporting & Fee Collection	\$1,942,000	\$1,942,000	\$0	\$1,942,000	\$1,942,000	\$0
	\$48,820,000	\$46,218,000	(\$2,602,000)	\$48,820,000	\$45,891,000	(\$2,929,000)
LOCAL ASSISTANCE, TOTAL	\$94,001,000	\$88,768,000	(\$5,233,000)	\$94,001,000	\$87,751,000	(\$6,250,000)

**Genetic Disease Screening Program – Newborn Screening Testing
BUDGET DETAIL – November 2011¹**

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 processing 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>
2010/2011	510,908	\$ 13.49
2011/2012	512,901	\$ 13.83
2012/2013	515,290	\$ 13.98

COST CENTER: Technical & Scientific

Costs are associated with specimen screening; include reagents kits, 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 fixed costs associated with specimen screening including: laboratory supplies, blood specimen filter paper, blood specimen storage and costs for special packaging for blood specimen transport, etc. Reagent kits, which are the majority of the Technical & Scientific costs, are purchased in lots based on anticipated caseload volume. 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>
2010/2011	510,908	\$ 44.66
2011/2012	512,901	\$ 42.72
2012/2013	515,290	\$ 40.90

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 time-sensitive coordination for specific confirmatory testing, family consultation – including consultation with the infant's Pediatrician, genetic disease counseling, family educational services and coordinated care referrals to specialized medical institutions. 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. The ASCs are reimbursed based on caseload and the type of service performed; this funding supports 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>
2010/2011	10,241	2.00%	\$ 427.59
2011/2012	10,474	2.04%	\$ 423.52
2012/2013	10,707	2.08%	\$ 418.23

**Genetic Disease Screening Program – Newborn Screening Testing
Budget Detail – November 2011¹**

COST CENTER: Reference Laboratories

Cases that result in a positive screening test are referred for diagnostic testing at various confirmatory laboratories. Costs include medical and confirmatory diagnostic tests, as well as fixed costs for lab technical support and expert medical consultation services for rare genetic abnormalities. 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>
2010/2011	9,979	1.92%	\$ 236.28
2011/2012	9,855	1.92%	\$ 249.21
2012/2013	9,922	1.93%	\$ 247.53

COST CENTER: Follow-up Diagnostic Services

Follow-up Diagnostic Services are for infants that require extended monitoring while undergoing confirmatory testing and diagnosis. Clinical outcome data is collected on infants once diagnosis is made as a means of tracking, confirmation, evaluation and refinement of program standards. Services include coordination with the NBS ASC and the GDSP for ongoing medical care, ensuring the establishment of infant treatment plans through specialty care hospitals and university medical centers specializing in the specific genetic disorder such as sickle cell anemia, cystic fibrosis, PKU, beta thalassemia, alpha thalassemia, and various neurologic, metabolic and endocrine disorders, etc. 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>
2010/2011	2,477	.48%	\$ 920.01
2011/2012	2,486	.48%	\$ 916.73
2012/2013	2,495	.48%	\$ 913.43

¹ Beginning in FY 2011-12, the following Cost Centers do not include SCID related expenditures as these costs are budgeted in State Operations: Technical & Scientific, Case Management and Coordination Services, Reference Laboratories and Follow-up Diagnostic Services. Therefore, the average cost per case excludes SCID.

**Genetic Disease Screening Program - Prenatal Testing
BUDGET DETAIL – November 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.

Fiscal Year	Total # of Cases	Average Cost Per Case	1st Trimester Screens	Average Cost Per Case	2nd Trimester Screens	Average Cost Per Case
2010/2011	408,726	\$10.81	280,293	\$4.42	370,528	\$8.70
2011/2012	410,321	\$11.34	294,308	\$4.53	372,010	\$8.93
2012/2013	412,232	\$11.79	309,023	\$4.65	373,870	\$9.16

COST CENTER: Technical & Scientific

Costs associated with screening services provided at the laboratory and include reagent kits, 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. Reagent kits, 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 Per Case	1st Trimester Cases	Average Cost Per Case	2nd Trimester Cases	Average Cost Per Case
2010/2011	408,726	\$31.30	280,293	\$13.26	370,528	\$24.49
2011/2012	410,321	\$31.75	294,308	\$13.19	372,010	\$24.59
2012/2013	412,232	\$31.87	309,023	\$12.73	373,870	\$24.61

**Genetic Disease Screening Program - Prenatal Testing
Budget Detail – November 2011**

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, id patients whose blood specimens were drawn too early or were inadequate, requiring additional blood draws. The PNS 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. Coordinators 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 PNS 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>
2010/2011	112,319	27.48%	\$ 48.62
2011/2012	115,689	28.19%	\$ 48.35
2012/2013	119,159	28.91%	\$ 48.35

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 reimbursed per service type.

<u>Fiscal Year</u>	<u># of Cases</u>	<u>% of Prenatal Cases</u>	<u>Average Cost Per Case</u>
2010/2011	18,175	4.45%	\$ 894.58
2011/2012	18,699	4.69%	\$ 894.98
2012/2013	19,260	4.69%	\$ 894.98

GENETIC DISEASE TESTING FUND
FUND CONDITION REPORT
DOLLARS IN THOUSANDS

	2010-11	2011-12	2012-13
RESOURCES			
BEGINNING BALANCE	\$1,413	\$6,502	\$7,310
Prior Year Adjustment	5,640		-
<i>Adjusted Beginning Balance</i>	<u>7,053</u>	<u>6,502</u>	<u>7,310</u>
REVENUES			
Genetic Disease Testing Fees	109,958	111,860	114,893
Income from Surplus Investments	9	50	50
Escheat of Unclaimed Checks & Warrants	8	10	10
TOTALS, REVENUES	<u>109,975</u>	<u>111,920</u>	<u>114,953</u>
TOTALS, REVENUES AND TRANSFERS	<u>\$109,975</u>	<u>\$111,920</u>	<u>\$114,953</u>
TOTAL RESOURCES	<u>\$117,028</u>	<u>\$118,422</u>	<u>\$122,263</u>

EXPENDITURES AND EXPENDITURE ADJUSTMENTS			
STATE OPERATIONS			
2011-12 Budget Act Appropriation	21,735	22,232	21,838
GDSP Administration	(18,503)	(19,583)	(19,189)
Lease Revenue Debt Service	(2,093)	(2,098)	(2,098)
HIPAA	(551)	(551)	(551)
Adjustments to State Operations:			
Lease Revenue Debt Service	-		
NBS SCID Expansion			5,296
<i>Subtotal, State Operations</i>	<u>21,147</u>	<u>22,232</u>	<u>27,134</u>
LOCAL ASSISTANCE			
2011-12 Budget Act Appropriation	93,227	94,001	94,001
Adjustments to Local Assistance:			
November 2011 Estimate Base	-	88,768	87,751
<i>Subtotal, Local Assistance</i>	<u>89,265</u>	<u>88,768</u>	<u>87,751</u>
State Controller	44	42	37
Financial Information System for California (FI\$Cal)	70	70	19
TOTAL EXPENDITURES AND EXPENDITURE ADJUSTMENTS	<u>110,526</u>	<u>111,112</u>	<u>114,941</u>

FUND BALANCE	6,502	7,310	7,322
	6%	7%	6%

REVENUE PROJECTIONS

2011-12

2011-12 NBS FEES BASED ON	512,901	TESTS @	\$102.75	AND	98%	=	\$51,646,566
SCID 6 mo. fee increase \$9.95	512,901	TESTS @	\$9.95	AND	98%	=	\$2,500,649
2011-12 PNS FEES BASED ON	205,160	TESTS @	\$152.00	AND	93%	=	\$29,001,474
2011-12 PNS FEES BASED ON	205,160	TESTS @	\$150.48	AND	93%	=	\$28,711,459
	<u>410,321</u>						<u>\$57,712,934</u>

\$111,860,149

2012-13

2012-13 NBS FEES BASED ON	515,290	TESTS @	\$102.75	AND	98%	=	\$51,887,127
SCID fee increase \$9.95	515,290	TESTS @	\$9.95	AND	98%	=	\$5,024,593
2012-13 PNS FEES BASED ON	206,116	TESTS @	\$152.00	AND	93%	=	\$29,136,558
2012-13 PNS FEES BASED ON	206,116	TESTS @	\$150.48	AND	93%	=	\$28,845,192
	<u>412,232</u>						<u>\$57,981,750</u>

\$114,893,469

GENETIC DISEASE SCREENING PROGRAM ASSUMPTIONS

November 2011

FISCAL YEARS 2011-12 & 2012-13

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

None

GDSP: OLD ASSUMPTIONS

Applicable F/Y

C/Y B/Y

None

GDSP: INFORMATION ONLY

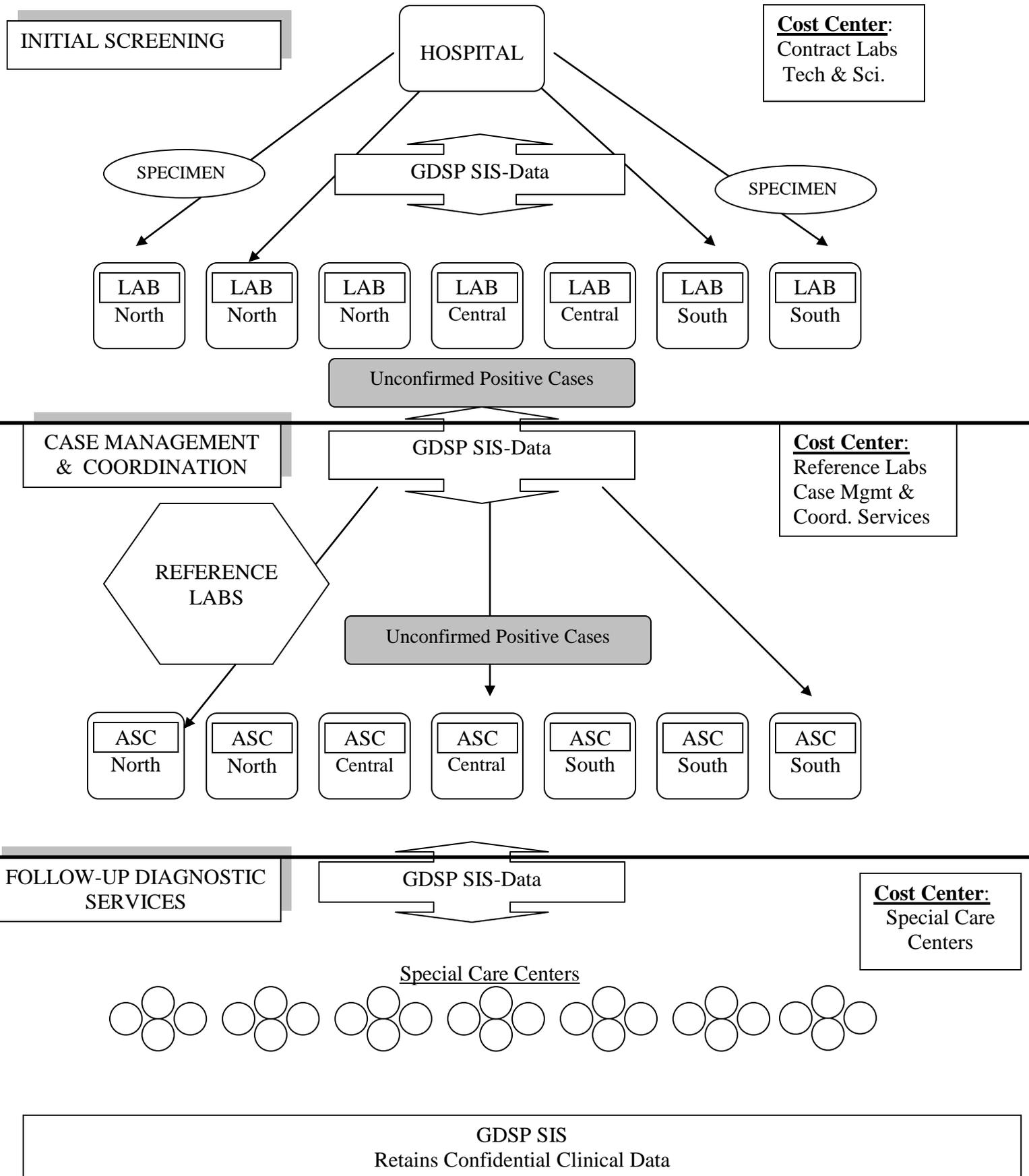
1. Newborn Screening Program (NBS) Program Expansion FY 2011-12

In accordance with the legislative mandate pursuant to Chapter 461, Statutes of 2011 (hereafter referred to as Assembly Bill 395) requires CDPH to add Severe Combined Immunodeficiency (SCID) to the panel of disorders screened for by the GDSP Newborn Screening Program (NBS). CDPH would also be required to screen for related T-cell lymphopenias, provided there is no additional cost. By incorporating SCID screening into the newborn panel, California will meet the current national standard of care as recommended by the federal Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) and bring the NBS Program into alignment with the most up-to-date research, technology, laboratory, public health standards and practices.

2. Prenatal Screening (PNS) Program Expansion FY 2008-09

The GDSP successfully implemented First Trimester screening beginning in April 2009. With the addition of First Trimester testing, the GDSP became current with the most up-to-date recommendations and public health standards in prenatal screening. Currently women receive screening services in both trimesters, including a second ultrasound during the First Trimester. Combining both screens results in what is referred to as Integrated Screening, an approach that improves detection rates. As a result of available First Trimester screening, PNS program participation rate increased by 10% from FY 2008-09 through FY 2010-11 and is expected to remain steady at approximately 80% of the NBS birth caseload.

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.
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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 or result in unacceptable reporting errors.
- 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 the initial 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 conducted at multiple regional Special Care Centers; these Centers are experts in the specific area of the genetic abnormality and provide extensive monitoring, diagnosis and treatment. Specialty Centers provide case reporting and annual 5 year follow-up data to GDSP on diagnosed cases medically managed through the specialty center.
- Health outcome data of infants detected through the program serves as a critical mechanism for tracking, confirmation, evaluation and refinement of program standards. This data is used to collaborate with other State genetic screening programs to refine identification and treatment as a national benefit.
- GDSP is data driven in its policy and funding allocations, the outcome data of cases initially identified through the program are critical in ensuring the CA screening program is a success.
- Having a contractual relationship with a network of specialized medical care centers assures the State that efforts in detecting genetic disorders are not wasted in that once a genetic disorder is detected, a timely and a precise referral process for medical intervention will be initiated to resolve avoidable medical problems as well as unnecessary costs to the public health care system if the case is not treated.
- 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 diagnosis, such as sickle cell anemia, cystic fibrosis, PKU, beta thalassemia, alpha thalassemia, and various neurologic, metabolic, endocrine and immune disorders.

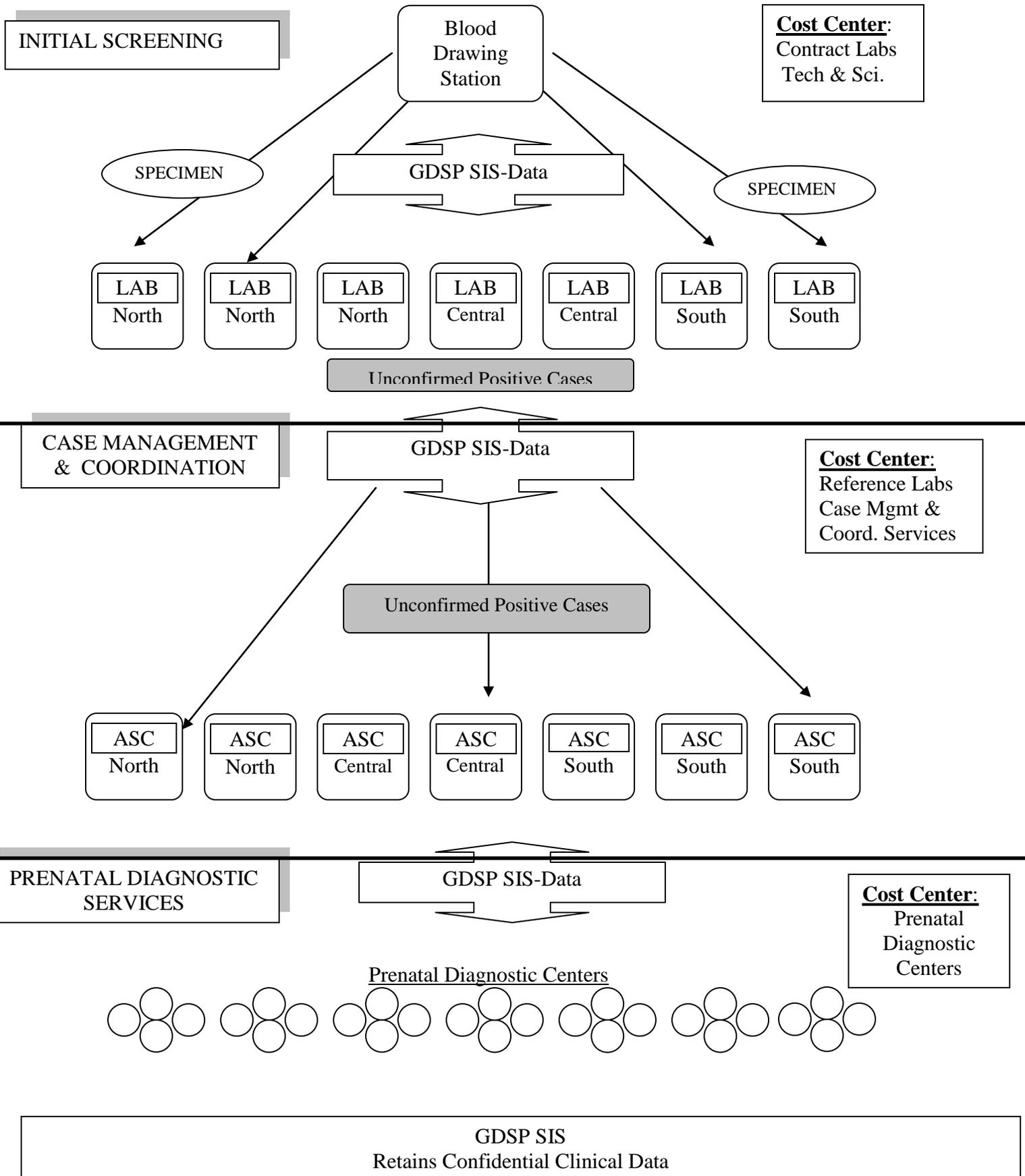
COST CENTER: Expenditures in **Follow-up Diagnostic Services** reflect costs for services on a per case reimbursement.

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.
- 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.

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.
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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

	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	12,032	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	13,252	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	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	44.62	106.59	110.47	97.89	50.00	12.80	2.11
2002	50,947	123,065	137,250	130,379	69,879	17,725	529,245	41.64	104.73	110.11	100.06	51.15	13.30	2.11
2003	50,042	123,822	140,566	134,819	72,669	18,910	540,828	40.30	103.72	111.77	103.80	53.60	14.24	2.14
2004	50,436	124,318	141,621	134,592	74,589	19,129	544,685	40.03	102.51	111.61	103.96	55.44	14.46	2.14
2005	50,777	125,541	143,463	133,760	75,740	19,418	548,700	39.73	101.94	112.07	103.65	56.74	14.72	2.14
2006	53,455	129,153	148,287	133,462	77,793	20,007	562,157	41.23	103.29	114.83	103.75	58.73	15.22	2.19
2007	54,060	127,996	150,523	135,376	78,453	19,729	566,137	41.12	100.85	115.56	105.58	59.70	15.06	2.19
2008	52,332	122,281	147,071	132,616	76,962	20,304	551,567	39.26	94.94	111.94	103.77	59.04	15.55	2.12
2009	48,362	113,942	140,972	129,089	74,488	19,922	526,774	35.79	87.19	106.39	101.34	57.60	15.31	2.02
2010	43,584	107,664	136,837	128,895	72,962	20,031	509,974	31.78	81.08	102.24	101.36	56.79	15.43	1.94
Projection:								Projection:						
2011	41,816	108,194	137,380	129,752	74,451	20,248	511,842	30.52	79.16	101.30	102.05	57.99	15.57	1.93
2012	40,064	109,713	137,911	130,737	75,053	20,482	513,960	29.42	77.95	99.98	102.44	58.37	15.82	1.92
2013	38,203	111,175	139,406	131,870	75,298	20,666	516,619	28.32	77.11	99.08	102.79	58.38	16.05	1.91
2014	36,772	111,582	141,395	133,298	76,132	20,662	519,842	27.53	75.90	98.11	103.42	58.65	16.11	1.90
2015	35,501	111,208	144,294	136,093	76,740	20,686	526,521	26.82	74.80	97.31	104.55	58.74	16.34	1.89
2016	34,889	109,687	147,670	142,864	78,111	20,869	534,090	26.37	73.48	96.52	105.53	59.09	16.58	1.89
2017	34,399	108,186	150,973	148,191	80,500	21,101	543,350	25.95	72.66	95.74	106.69	60.03	16.82	1.89
2018	34,700	106,131	153,792	154,043	82,853	21,370	552,888	26.07	71.65	94.93	107.77	60.82	17.07	1.89
2019	34,900	104,188	156,203	159,276	84,877	21,672	561,116	26.09	70.74	94.40	107.87	61.43	17.35	1.89
2020	34,894	102,498	159,036	165,479	87,147	21,826	570,880	26.03	69.99	94.09	108.74	62.13	17.51	1.89

Source: Historical births through 2010, 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.