

GENETIC DISEASE SCREENING  
PROGRAM  
May 2013  
ESTIMATE  
*for*  
FISCAL YEARS  
2012-13 *and* 2013-14



CALIFORNIA  
DEPARTMENT OF PUBLIC HEALTH

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## **APPENDICES**

### **DEPARTMENT OF FINANCE DEMOGRAPHIC DATA**

**EXECUTIVE SUMMARY**

The Genetic Disease Screening Program (GDSP) proposes no additional authority for either the Current or Budget Year. The May 2013 Revision Estimate CY and BY budget remains at approximately \$87.7 million.

GDSP expenditure forecast is based upon the following factors:

- Updated birth data for years 2011 through 2014, derived from Department of Finance's Demographic Research Unit (see Table 1 and 2 on page 24);
- The average cost per case based on past year actuals and vendor prices for the Cost Centers, which are described in further detail in the Estimate package.
  - Cost Centers are strictly caseload and price driven, with the exception of System Project & Maintenance and Result Reporting & Fee Collection, which are impacted by other factors such as IT system maintenance, billing and shipping costs.

**NEWBORN SCREENING PROGRAM (NBS)**

|  | FY 13/14<br>BUDGET | FY 13/14<br>MAY REVISE | %<br>Change |
|--|--------------------|------------------------|-------------|
| Contract Laboratories                            | \$ 14.71           | \$ 14.29               | -3%         |
| Tech & Sci.                                      | \$ 39.68           | \$ 39.68               | 0%          |
| System Project & Maintenance (Total Budget)      | \$ 3,007,000       | \$ 3,007,000           | 0%          |
| Case Management & Coordination Services          | \$ 416.42          | \$ 416.42              | 0%          |
| Reference Laboratories                           | \$ 209.43          | \$ 209.43              | 0%          |
| Follow-up Diagnostic Services                    | \$ 883.75          | \$ 883.75              | 0%          |
| Result Reporting & Fee Collection (Total Budget) | \$ 1,575,000       | \$ 1,575,000           | 0%          |

The May 2013 Revise Estimate contains no changes other than a reduction to the NBS Contract Laboratories budget in the amount of \$221,778 as a result of ongoing rate and laboratory consolidation negotiations between the Department and the regional laboratories; negotiated BY rates are assumed in this budget and expected to remain at the CY rate level.

**PRENATAL SCREENING PROGRAM (PNS)**

|  | FY 13/14<br>BUDGET | FY 13/14<br>MAY REVISE | %<br>Change |
|--|--------------------|------------------------|-------------|
| Contract Laboratories                            | \$ 11.81           | \$ 13.70               | 14%         |
| Tech & Sci.                                      | \$ 32.33           | \$ 32.33               | 0%          |
| System Project & Maintenance (Total Budget )     | \$ 3,130,000       | \$ 3,130,000           | 0%          |
| Case Management & Coordination Services          | \$ 45.22           | \$ 45.22               | 0%          |
| Prenatal Diagnostic Services                     | \$ 894.58          | \$ 864.57              | -3%         |
| Result Reporting & Fee Collection (Total Budget) | \$ 2,027,000       | \$ 2,027,000           | 0%          |

The May 2013 Revise Estimate contains two changes to the budget; as a result of 1<sup>st</sup> Trimester and 2<sup>nd</sup> Trimester rate and laboratory consolidation negotiations between the Department and the regional laboratories, an increase to the Contract Laboratories of \$780,945 is expected. This increase is offset in part by savings to the Prenatal Diagnostic Services budget, which resulted because of improved detection and reduced utilization of costly invasive prenatal diagnostic services.

## BUDGET ESTIMATE OVERVIEW

GENETIC DISEASE SCREENING PROGRAM  
May 2013 Estimate

| GENETIC DISEASE SCREENING PROGRAM<br>MAY 2013 BUDGET ESTIMATE |                                   |                         |            | 2013-2014                         |                         |                  |
|---|-----------------------------------|-------------------------|------------|-----------------------------------|-------------------------|------------------|
| PROGRAM   | 2012-2013                         |                         |            | 2013-2014                         |                         |                  |
|   | 2012-2013<br>NOVEMBER<br>ESTIMATE | 2012-2013 MAY<br>REVISE | DIFFERENCE | 2013-2014<br>GOVERNOR'S<br>BUDGET | 2013-2014 MAY<br>REVISE | DIFFERENCE       |
| <b>LOCAL ASSISTANCE</b>                                       |                                   |                         |            |                                   |                         |                  |
| <b>NBS</b>  |                                   |                         |            |                                   |                         |                  |
| Contract Laboratories:  | \$7,285,874                       | \$7,285,874             | \$0        | \$7,614,377                       | \$7,392,600             | (\$221,778)      |
| Tech & Sci:   | \$20,238,041                      | \$20,238,041            | \$0        | \$20,534,492                      | \$20,534,492            | \$0              |
| System Project & Maintenance:                                 | \$3,312,000                       | \$3,312,000             | \$0        | \$3,007,000                       | \$3,007,000             | \$0              |
| Case Management & Coordination Services:                      | \$4,478,262                       | \$4,478,262             | \$0        | \$4,543,861                       | \$4,543,861             | \$0              |
| Reference Laboratories:                                       | \$2,464,521                       | \$2,464,521             | \$0        | \$2,125,528                       | \$2,125,528             | \$0              |
| Follow-up Diagnostic Services:                                | \$2,057,078                       | \$2,057,078             | \$0        | \$2,087,211                       | \$2,087,211             | \$0              |
| Result Reporting & Fee Collection                             | \$1,650,000                       | \$1,650,000             | \$0        | \$1,575,000                       | \$1,575,000             | \$0              |
|   | \$41,485,776                      | \$41,485,776            | \$0        | \$41,487,469                      | \$41,265,691            | (\$221,778)      |
| <b>PNS</b>  |                                   |                         |            |                                   |                         |                  |
| Contract Laboratories:  | \$4,680,166                       | \$4,680,166             | \$0        | \$4,891,184                       | \$5,672,129             | \$780,945        |
| Tech & Sci:   | \$13,190,493                      | \$13,190,493            | \$0        | \$13,383,711                      | \$13,383,711            | \$0              |
| System Project & Maintenance:                                 | \$3,474,825                       | \$3,474,825             | \$0        | \$3,130,000                       | \$3,130,000             | \$0              |
| Case Management & Coordination Services:                      | \$5,999,993                       | \$5,999,993             | \$0        | \$6,087,882                       | \$6,087,882             | \$0              |
| Prenatal Diagnostic Services:                                 | \$16,499,746                      | \$16,499,746            | \$0        | \$16,741,438                      | \$16,179,808            | (\$561,630)      |
| Result Reporting & Fee Collection                             | \$2,402,000                       | \$2,402,000             | \$0        | \$2,027,000                       | \$2,027,000             | \$0              |
|   | \$46,247,224                      | \$46,247,224            | \$0        | \$46,261,215                      | \$46,480,530            | \$219,315        |
| <b>LOCAL ASSISTANCE, TOTAL</b>                                | <b>\$87,733,000</b>               | <b>\$87,733,000</b>     | <b>\$0</b> | <b>\$87,748,684</b>               | <b>\$87,746,221</b>     | <b>(\$2,463)</b> |
|   |                                   |                         |            |                                   |                         |                  |

**Genetic Disease Screening Program – Newborn Screening Testing  
BUDGET DETAIL – May 2013<sup>1</sup>**

**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> |
|--------------------|-------------------|------------------------------|
| 2011/2012          | 503,525           | \$ 13.87                     |
| 2012/2013          | 510,028           | \$ 14.29                     |
| 2013/2014          | 517,499           | \$ 14.29                     |

**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> |
|--------------------|-------------------|------------------------------|
| 2011/2012          | 503,525           | \$ 39.68                     |
| 2012/2013          | 510,028           | \$ 39.68                     |
| 2013/2014          | 517,499           | \$ 39.68                     |

**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 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> |
|--------------------|-------------------|-----------------------|------------------------------|
| 2011/2012          | 10,617            | 2.11%                 | \$ 416.42                    |
| 2012/2013          | 10,754            | 2.11%                 | \$ 416.42                    |
| 2013/2014          | 10,912            | 2.11%                 | \$ 416.42                    |

**Genetic Disease Screening Program – Newborn Screening**

**Genetic Disease Screening Program – Newborn Screening Testing  
Budget Detail – May 2013**

**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.

| <u>Fiscal Year</u> | <u># of Cases</u> | <u>% of NBS Cases</u> | <u>Average Cost Per Case</u> |
|--------------------|-------------------|-----------------------|------------------------------|
| 2011/2012          | 9,875             | 1.96%                 | \$ 246.39                    |
| 2012/2013          | 10,003            | 1.96%                 | \$ 246.39                    |
| 2013/2014          | 10,149            | 1.96%                 | \$ 209.43                    |

**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 genetic disorders 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> |
|--------------------|-------------------|-----------------------|------------------------------|
| 2011/2012          | 2,298             | .46%                  | \$ 883.75                    |
| 2012/2013          | 2,486             | .46%                  | \$ 883.75                    |
| 2013/2014          | 2,495             | .46%                  | \$ 883.75                    |

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<sup>1</sup> These cost estimates do not include SCID expenditures, as all costs related to this disorder are budgeted under State Operations.

**Genetic Disease Screening Program - Prenatal Testing**  
**BUDGET DETAIL – May 2013**

**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 Tests | Average Cost Per Test | 2nd Trimester Tests | Average Cost Per Test |
|-------------|------------------|-----------------------|---------------------|-----------------------|---------------------|-----------------------|
| 2011/2012   | 402,820          | \$11.14               | 286,800             | \$4.54                | 354,622             | \$8.98                |
| 2012/2013   | 408,022          | \$11.47               | 293,458             | \$4.65                | 355,000             | \$9.16                |
| 2013/2014   | 413,999          | \$13.70               | 299,327             | \$8.66                | 356,000             | \$8.66                |

**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 Tests | Average Cost Per Test | 2nd Trimester Tests | Average Cost Per Test |
|-------------|------------------|-----------------------|---------------------|-----------------------|---------------------|-----------------------|
| 2011/2012   | 402,820          | \$31.40               | 286,800             | \$13.22               | 354,622             | \$24.98               |
| 2012/2013   | 408,022          | \$32.33               | 293,458             | \$13.53               | 355,000             | \$25.97               |
| 2013/2014   | 413,999          | \$32.33               | 299,327             | \$13.53               | 356,000             | \$25.97               |



**Genetic Disease Screening Program - Prenatal Testing  
Budget Detail – May 2013**

**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> |
|--------------------|-------------------|----------------------------|------------------------------|
| 2011/2012          | 130,995           | 32.52%                     | \$ 43.59                     |
| 2012/2013          | 132,687           | 32.52%                     | \$ 45.22                     |
| 2013/2014          | 134,630           | 32.52%                     | \$ 45.22                     |

**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> |
|--------------------|-------------------|----------------------------|------------------------------|
| 2011/2012          | 18,209            | 4.52%                      | \$ 894.58                    |
| 2012/2013          | 18,444            | 4.52%                      | \$ 894.58                    |
| 2013/2014          | 18,714            | 4.52%                      | \$ 864.57                    |

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

|  | 2011-12          | 2012-13          | 2013-14          |
|--|------------------|------------------|------------------|
| <b>RESOURCES</b>                       |                  |                  |                  |
| BEGINNING BALANCE                      | \$6,502          | \$4,517          | \$925            |
| Prior Year Adjustment                  | 539              | 0                | -                |
| Adjusted Beginning Balance             | 7,041            | 4,517            | 925              |
| <b>REVENUES</b>                        |                  |                  |                  |
| Genetic Disease Testing Fees           | 108,600          | 111,125          | 115,679          |
| Income from Surplus Investments        | 7                | 7                | 7                |
| Escheat of Unclaimed Checks & Warrants | 4                | 4                | 4                |
| TOTALS, REVENUES                       | 108,611          | 111,136          | 115,690          |
| <b>TOTALS, REVENUES AND TRANSFERS</b>  | <b>\$108,611</b> | <b>\$111,136</b> | <b>\$115,690</b> |
| <b>TOTAL RESOURCES</b>                 | <b>\$115,652</b> | <b>\$115,653</b> | <b>\$116,615</b> |

|   |                |                |                |
|---|----------------|----------------|----------------|
| <b>EXPENDITURES AND EXPENDITURE ADJUSTMENTS</b>       |                |                |                |
| <b>STATE OPERATIONS</b>                               |                |                |                |
| Budget Act Appropriation/Budget Bill                  | 22,232         | 27,134         | 27,134         |
| GDSP Administration                                   | (17,811)       | (24,486)       | (24,486)       |
| Lease Revenue Debt Service                            | (2,091)        | (2,097)        | (2,097)        |
| HIPAA   | (297)          | (551)          | (551)          |
| Adjustments to State Operations:                      |                |                |                |
| Lease Revenue Debt Service                            | -              | -40            | -121           |
| NBS SCID Expansion                                    |                |                |                |
| Subtotal, State Operations                            | 20,199         | 26,803         | 27,985         |
| <b>LOCAL ASSISTANCE</b>                               |                |                |                |
| Budget Act Appropriation/Budget Bill                  | 94,001         | 87,751         | 87,751         |
| Adjustments to Local Assistance:                      |                |                |                |
| November 2012 Estimate Base                           |                | 87,751         | 87,749         |
| May 2013 Adjustments to Local Assistance:             |                |                | -3             |
| Subtotal, Local Assistance                            | 90,842         | 87,733         | 87,746         |
| State Controller                                      | 42             | 47             | 0              |
| Financial Information System for California (FI\$Cal) | 52             | 145            | 125            |
| <b>TOTAL EXPENDITURES AND EXPENDITURE ADJUSTMENTS</b> | <b>111,135</b> | <b>114,728</b> | <b>115,856</b> |

|                     |       |     |     |
|---------------------|-------|-----|-----|
| <b>FUND BALANCE</b> | 4,517 | 925 | 759 |
|                     | 4%    | 1%  | 1%  |

**REVENUE PROJECTIONS**

**2012-13**

|                           |         |                            |              |
|---------------------------|---------|----------------------------|--------------|
| 2012-13 NBS FEES BASED ON | 510,028 | TESTS @ \$112.70 AND 98% = | \$56,330,552 |
| 2012-13 PNS FEES BASED ON | 204,011 | TESTS @ \$152.00 AND 93% = | \$28,839,023 |
| 2012-13 PNS FEES BASED ON | 204,011 | TESTS @ \$136.80 AND 93% = | \$25,955,121 |
|                           | 408,022 |                            | \$54,794,144 |

**\$111,124,697**

**2013-14**

|                           |         |                            |              |
|---------------------------|---------|----------------------------|--------------|
| 2013-14 NBS FEES BASED ON | 517,499 | TESTS @ \$112.70 AND 98% = | \$57,155,695 |
| 2013-14 PNS FEES BASED ON | 207,000 | TESTS @ \$152.00 AND 93% = | \$29,261,463 |
| 2013-14 PNS FEES BASED ON | 207,000 | TESTS @ \$152.00 AND 93% = | \$29,261,463 |
|                           | 413,999 |                            | \$58,522,927 |

\*FY 2013-14 PNS FEE assumes full payment from Medi-Cal.

**\$115,678,621**

# **GENETIC DISEASE SCREENING PROGRAM ASSUMPTIONS**

## **November 2012**

### **FISCAL YEARS 2012-13 & 2013-14**

#### **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****FUTURE FISCAL ISSUES**

CDPH/GDSP contracts with seven laboratories around the state to provide Newborn screening and Prenatal screening services for the newborns and pregnant women of California. GDSP is in the process of analyzing the viability of consolidating seven regional laboratories to four. This transition is projected to occur during Fiscal Year 2014/15 unless it is calculated to be cost prohibitive.

**NEW ASSUMPTIONS**

Applicable F/Y

|            |            |
|------------|------------|
| <u>C/Y</u> | <u>B/Y</u> |
|            | X          |

The Prenatal Screening Program provides genetic screening services during the 1st and 2<sup>nd</sup> Trimester of a woman's pregnancy. The existing contracted laboratory screening rates for 1st Trimester testing will expire June 30, 2013. The introductory 1st Trimester screening rates GDSP received during the 2009 expansion are expected to increase.

**OLD ASSUMPTIONS**

Applicable F/Y

|            |            |
|------------|------------|
| <u>C/Y</u> | <u>B/Y</u> |
|------------|------------|

None

**GDSP: INFORMATION ONLY****1. Newborn Screening Program (NBS) Program Expansion FY 2011-12**

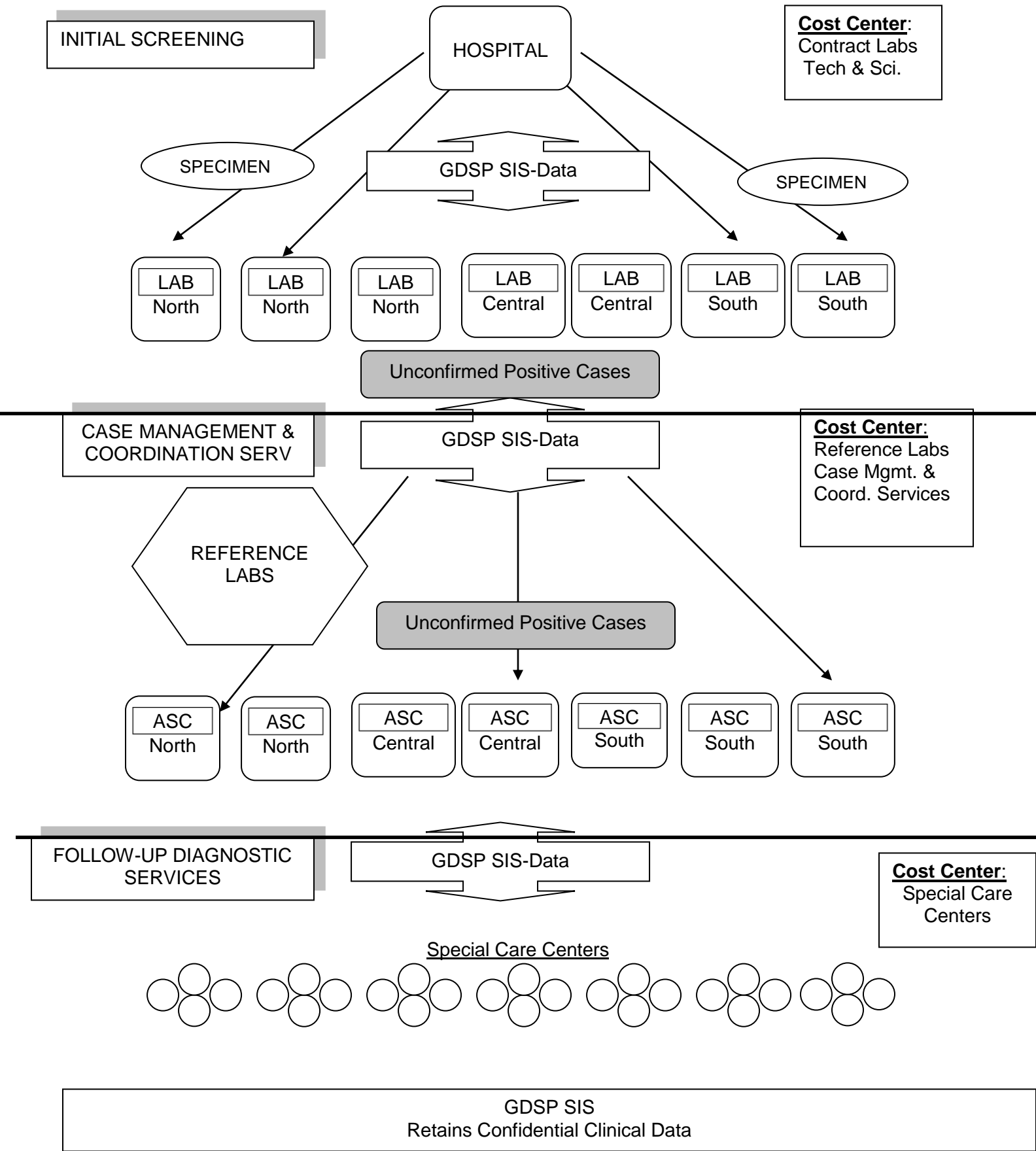
In accordance with the legislative mandate pursuant to Chapter 461, Statutes of 2011 (hereafter referred to as Assembly Bill 395) requires CDPH to add Severe Combined Immunodeficiency (SCID) to the panel of disorders screened for by the GDSP Newborn Screening Program (NBS). CDPH would also be required 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 80 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. |
|--|

**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.

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#### **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.

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#### **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.

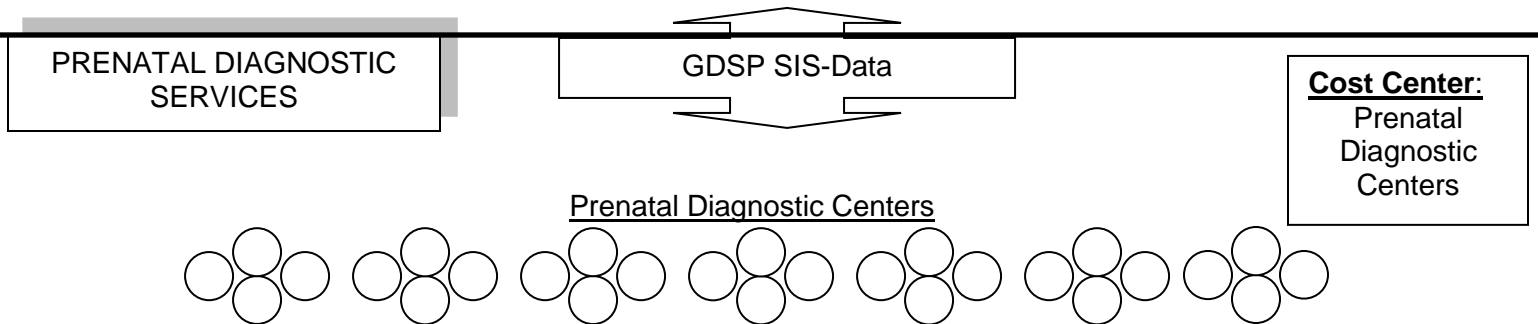
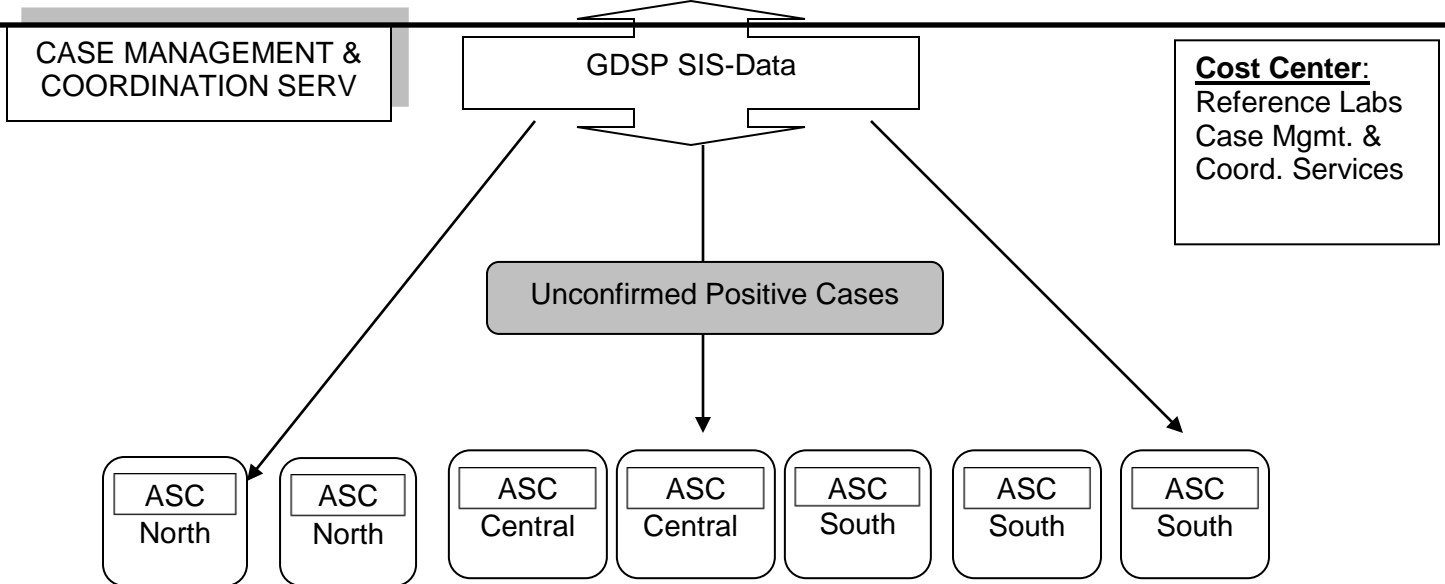
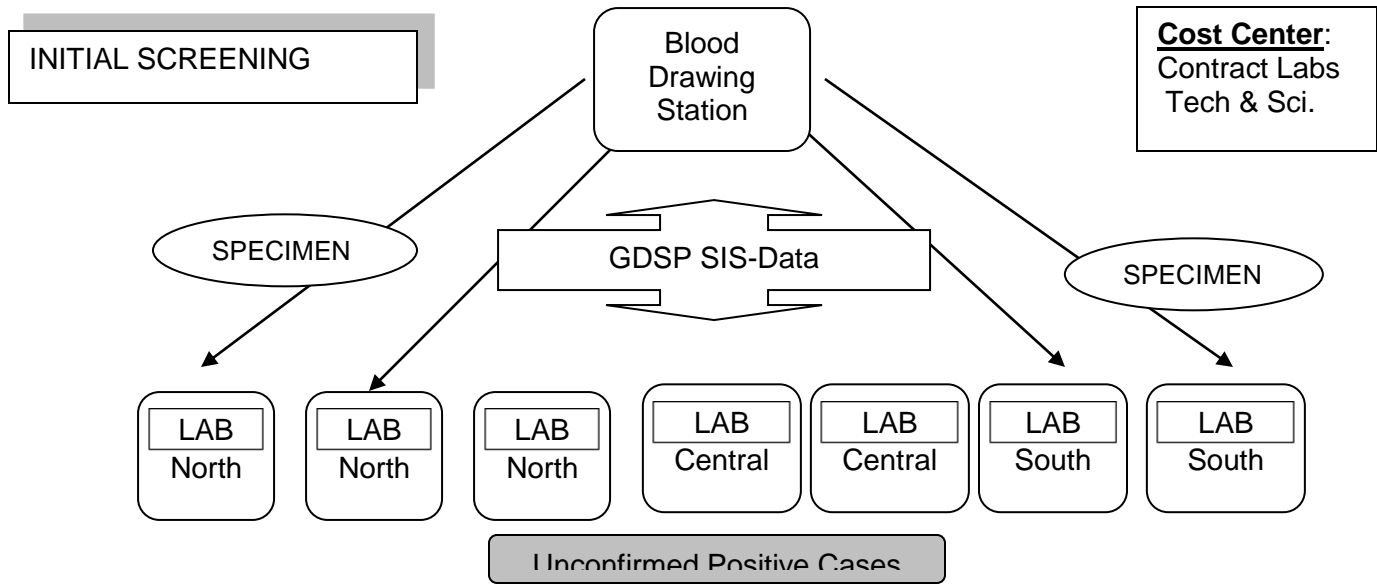
**COST CENTER:** Expenditures in **Follow-up Diagnostic Services** reflect costs for services on a per case reimbursement.

**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 of reporting results and production of follow-up material to medical providers and families of children as well as costs related to the collection of payments.

# PRENATAL SCREENING PROGRAM



**GDSP SIS**  
Retains Confidential Clinical Data

## 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.

**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.

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#### **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.

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#### **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.

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#### **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.

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#### **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   | 148,653   | 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 | 47.43                                 | 106.97    | 111.44    | 95.86     | 49.41     | 12.40     | 2.12 |
| 2001  | 53,779    | 123,236   | 136,449   | 127,957   | 68,835    | 17,117    | 527,372 | 46.60                                 | 104.02    | 113.37    | 94.86     | 49.85     | 12.18     | 2.10 |
| 2002  | 50,947    | 123,065   | 137,250   | 130,379   | 69,879    | 17,725    | 529,245 | 43.84                                 | 101.96    | 111.92    | 95.27     | 50.65     | 12.45     | 2.08 |
| 2003  | 50,042    | 123,822   | 140,566   | 134,819   | 72,669    | 18,910    | 540,828 | 40.74                                 | 99.00     | 112.72    | 97.64     | 52.02     | 12.79     | 2.07 |
| 2004  | 50,436    | 124,318   | 141,621   | 134,592   | 74,589    | 19,129    | 544,685 | 39.25                                 | 97.64     | 115.04    | 102.43    | 54.77     | 13.59     | 2.11 |
| 2005  | 50,777    | 125,541   | 143,463   | 133,760   | 75,740    | 19,418    | 548,700 | 38.79                                 | 96.32     | 114.80    | 104.08    | 56.35     | 13.78     | 2.12 |
| 2006  | 53,455    | 1         | 148,287   | 133,462   | 77,793    | 20,007    | 562,157 | 38.50                                 | 96.20     | 114.37    | 105.21    | 56.99     | 14.16     | 2.13 |
| 2007  | 54,060    | 127,996   | 150,523   | 135,376   | 78,453    | 19,729    | 566,137 | 40.00                                 | 98.16     | 115.89    | 106.16    | 58.24     | 14.82     | 2.17 |
| 2008  | 52,332    | 122,281   | 147,071   | 132,616   | 76,962    | 20,304    | 551,567 | 39.88                                 | 96.98     | 115.12    | 107.64    | 58.67     | 14.80     | 2.17 |
| 2009  | 48,362    | 113,942   | 140,972   | 129,089   | 74,488    | 19,922    | 526,774 | 38.27                                 | 92.11     | 109.61    | 104.20    | 58.28     | 15.46     | 2.09 |
| 2010  | 43,584    | 107,664   | 136,837   | 128,895   | 72,962    | 20,031    | 509,974 | 35.34                                 | 85.44     | 103.52    | 99.32     | 57.72     | 15.27     | 1.98 |
| 2011  | 38,754    | 103,017   | 134,289   | 132,375   | 72,980    | 20,608    | 502,023 | 32.03                                 | 80.07     | 100.08    | 97.86     | 57.03     | 15.36     | 1.91 |
| Projection:                                 |           |           |           |           |           |           |         | Projection:                           |           |           |           |           |           |      |
| 2012  | 39,010    | 102,746   | 134,373   | 135,668   | 72,594    | 20,635    | 505,027 | 28.62                                 | 76.44     | 98.18     | 100.62    | 57.05     | 15.85     | 1.88 |
| 2013  | 39,801    | 106,850   | 136,273   | 136,380   | 74,742    | 20,984    | 515,029 | 29.91                                 | 73.45     | 97.11     | 103.17    | 56.74     | 15.92     | 1.88 |
| 2014  | 41,140    | 107,008   | 136,741   | 138,541   | 75,462    | 21,078    | 519,969 | 30.83                                 | 74.55     | 97.23     | 102.60    | 58.57     | 16.37     | 1.90 |
| 2015  | 41,645    | 103,820   | 137,785   | 141,015   | 77,855    | 21,057    | 523,178 | 32.11                                 | 74.67     | 96.08     | 103.24    | 59.11     | 16.66     | 1.91 |
| 2016  | 41,912    | 100,827   | 139,708   | 142,368   | 79,664    | 20,976    | 525,455 | 32.54                                 | 72.97     | 95.45     | 104.36    | 60.74     | 16.88     | 1.91 |
| 2017  | 41,929    | 98,816    | 144,557   | 146,254   | 82,586    | 21,391    | 535,534 | 32.71                                 | 71.84     | 93.57     | 103.31    | 61.01     | 16.62     | 1.90 |
| 2018  | 41,801    | 96,984    | 147,463   | 150,624   | 84,925    | 21,803    | 543,601 | 32.60                                 | 71.53     | 94.09     | 103.97    | 61.97     | 16.74     | 1.90 |
| 2019  | 42,308    | 95,438    | 146,682   | 153,093   | 86,022    | 21,808    | 545,351 | 32.38                                 | 71.09     | 95.72     | 105.99    | 63.21     | 17.16     | 1.93 |
| 2020  | 42,219    | 94,411    | 146,924   | 155,390   | 87,106    | 21,796    | 547,845 | 32.66                                 | 70.83     | 94.95     | 106.66    | 63.54     | 17.25     | 1.93 |
| 2021  | 41,988    | 93,457    | 146,071   | 157,832   | 88,176    | 21,780    | 549,304 | 32.48                                 | 70.93     | 94.83     | 107.21    | 63.86     | 17.33     | 1.93 |

Source: Historical births through 2011, 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.