

## Background

The California Department of Public Health, Center for Family Health (Department) maintains a large and diverse biobank, the California Biobank Program, which houses over 17.5 million prenatal serum samples and newborn leftover blood spots. These samples were collected for testing by the Prenatal (PNS) and Newborn (NBS) screening programs that are administered by the Genetic Disease Screening Program (GDSP) and are linked to GDSP data and California Birth Defects Monitoring Program (CBDMP) registry data. The samples and data are also linked to the State Registrar of Vital Statistics databases that include fetal death, live birth and death data. The California Biobank Program (CBP) is an internationally recognized public health asset because of its large size and culturally, geographically and genetically diverse population. The CBP is unique in that no other state or international effort approaches its scale in terms of the number and quality of specimens collected and preserved annually, the number of historic specimens, and existing linkage with statewide prenatal and newborn screening program data, birth defects data, CBDMP data and other population databases.

## History

The Genetic Disease Screening Program's (GDSP) Newborn and Prenatal Screening Programs are located within the Center for Family Health and represent one of the largest and most comprehensive screening programs in the world. Combined these programs screen approximately 500,000 newborns each year for over 80 genetic and congenital disorders and over 350,000 pregnant women for Down syndrome, trisomy 18 and neural tube defects. The screening programs provide testing and follow-up referral services. These referrals are critical for early diagnosis and treatment of disorders in an effort to prevent adverse outcomes, minimize the clinical effects of disorders, and improve health outcomes.

The California Birth Defects Monitoring Program is located within the Center for Family Health's, Maternal, Child and Adolescent Health Division. CBDMP is an active case finding, population based birth defects registry. The mission of the Division is to develop systems that protect and improve the health of California's women of reproductive age, infants, children, adolescents, and their families. The Birth Defects Monitoring Program was mandated in 1982 to find causes of birth defects. The Birth Defects Monitoring Program has collected information on children in California, up to one year of age, born with structural birth defects for over 30 years. Currently, data are collected from 10 counties in California. These include Fresno, Kern, Kings, Madera, Merced, Orange, San Diego, San Joaquin, Stanislaus and Tulare. Data collection specialists review hospital records and abstract information. This information is then linked to vital statistics (live birth or fetal death certificates) to provide demographic data. CDPH surveys more than 160,000 live births per year, of which approximately 3 - 5% have a birth defect.

Since the Newborn Screening Program began in 1980, the Genetic Disease Screening Program has banked leftover blood spots from screened newborns in freezer storage, but only those banked since 1982 are readily available. GDSP has banked over 16 million newborn specimens, which represents an entire generation of Californians. The GDSP began banking leftover maternal prenatal screening serum specimens in freezer storage from selected regions of the state in 2000. In 2003 the responsibility for banking was transferred to the California Birth Defects Monitoring Program (CBDMP) or designee under a memorandum of understanding with the Genetic Disease Screening Program. Over 1.5 million prenatal specimens have been banked so far, and approximately 100,000 specimens are added per year. In 2013 the Department combined these banking efforts under the California Biobank Program to comply with the 2008 mandate that the Department make the specimens available for research following approved purposes:

- **Identify risk factors for children's and women's diseases.**
- **Develop and evaluate screening tests.**
- **Develop and evaluate screening strategies.**
- **Develop and evaluate treatments.**

California laws require that the operation of the CBP be governed by strict protections for the confidentiality and security of the newborn and prenatal specimens and related data. These protections also apply to the researchers who receive such specimens and related data.

In the more than 30 years that GDSP and CBDMP have been storing specimens and data, there has never been a privacy breach involving the specimens and/or data. All specimens are stored without any identifying information. They are labeled only with a unique number.

Over the years, the newborn and prenatal specimens that have been banked have been used for a number of purposes. These include:

- 1) regular quality control and quality improvement efforts in the Genetic Disease Laboratory,
- 2) the timely evaluation, improvement and expansion of the newborn and prenatal screening programs,
- 3) investigation of the prenatal causes of diseases through collaborative research projects, with researchers in and out of state government,
- 4) the development of treatments for diseases effecting pregnant women and children and
- 5) providing specimens for individual requests of California families for further evaluation and testing and to investigate unexplained deaths or health impairments.