About the Data

Data Source

Demographic and specimen information pertaining to newborns participating in newborn screening (NBS) is documented on the NBS test request form by the birth provider. Outcome data is collected from special care centers via an online form for all newborns who are screen positive and referred to their centers. Data was extracted on 6/15/2023.

The number of births were estimated from provisional statewide live birth files (data.chhs.ca.gov/dataset/test-cdph-statewide-live-birth-profiles).

Date Ranges

Date ranges are noted in chart titles. The date ranges refer to when specimens are accessioned by the NBS laboratory and are inclusive. For example, "8/1/2018-12/31/2022" includes specimens accessioned on both 8/1/2018 and 12/31/2022 as well as all dates in-between.

Indicators and Denominators

Number of births – number of live births in California

Number of newborns screened – number of newborns who received dried blood spot screening through the California NBS Program

Number of screen-positive newborns – number of newborns who received a positive screening result through the California NBS Program

Number of newborns identified with a condition – newborns who were diagnosed with a condition screened for by the California NBS Program. A full list of conditions screened for by the California NBS Program can be found on the Genetic Disease Screening Program website (https://www.cdph.ca.gov/Programs/CFH/DGDS/Pages/nbs/NBS-Disorders-Detectable.aspx).

Category and Subcategory Definitions

Race/Ethnicity – "Asian" includes Asian East Indian, Chinese, Japanese, Korean, Cambodian, Laos, Vietnamese, Filipino, and Other Southeast Asian. "Other" includes Middle Eastern, Hawaiian, Guamanian, Samoan, and Other. Black, Hispanic, Native American, and White are distinct categories. When multiple races/ethnicities are selected for each newborn a single race/ethnicity is assigned according to the following priority: Native American first, then Black, Hispanic, Asian, White, then Other.

Special Care Center – approved California Children's Services special care centers that provide follow-up testing and care for newborns who screen positive through the California NBS Program

Condition - conditions screened for by the California NBS Program

- **Biotinidase deficiency (BD)** – includes:
  - Profound biotinidase deficiency
  - Partial biotinidase deficiency
- **Cystic fibrosis (CF)** – does not include CRMS
- **Endocrine (ENDO)** – includes:
  - Primary congenital hypothyroidism (PCH)
  - Congenital adrenal hyperplasia (CAH):
    - Salt wasting CAH
    - Simple virilizing CAH
- **Galactosemia (GAL)** – includes:
  - Classic galactosemia
  - Duarte (D/G) galactosemia
- **Hemoglobin (HB)** – When noted, HB conditions are separated into the following categories:
  - **Sickle cell disease (HB SCD)** – includes:
    - HB S/S (Sickle S/S disease)
    - HB S/C (sickle S/C disease)
    - HB S/D (sickle S/D disease)
    - HB S/E (sickle S/E disease)
    - HB S/V (sickle cell disease variant)
    - HB S/beta0 (sickle beta0 thalassemia)
    - HB S/beta+ (sickle beta+ thalassemia)
  - **Other hemoglobinopathies (HB other)** – includes:
    - Alpha thalassemia major
    - Alpha thalassemia trait
    - Beta thalassemia major
    - Beta thalassemia intermedia
    - Beta thalassemia trait
    - HB S/HPFH (sickle S/hereditary persistence of fetal hemoglobin)
    - HB E/beta0 thalassemia
    - HB E/beta+ thalassemia
    - HB E/delta beta thalassemia
    - HB C/beta0 thalassemia
    - HB C/beta+ thalassemia
    - HB D/beta0 thalassemia
    - HB D/beta+ thalassemia
    - HB variant/beta0 thalassemia
    - HB variant/beta+ thalassemia
    - HPFH/HPFH (Hereditary persistence of fetal hemoglobin, homozygous)
    - HB H disease
    - HB H/constant spring disease
    - HB H/other variant point mutations
    - HB E/E homozygous
    - HB C/C (HB C disease)
    - HB D/D (HB D disease)

        - HB variant/variant
        - HB FS confirmed but not differentiated
        - HB other disease
        - HB C/variant
        - HB D/variant
        - HB E/variant
- **Lysosomal storage disorders (LSD)** – includes:
    - Mucopolysaccharidosis:
        - Severe mucopolysaccharidosis type I (Hurlers)
        - Attenuated mucopolysaccharidosis type I (Scheie and Hurler-Scheie)
        - Mucopolysaccharidosis type I - not otherwise specified
    - Pompe disease:
        - Classic infant onset Pompe disease with cardiac involvement
        - Classic infant onset Pompe disease without cardiac involvement
        - Late onset Pompe disease
        - Pompe disease not otherwise specified
- **Metabolic disorders detected through tandem mass spectrometry (MS/MS)** – includes:
    - Phenylketonuria (PKU)
    - 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG CoA lyase deficiency/glutaric aciduria)
    - 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)
    - Argininosuccinyl-CoA lyase deficiency (ASAL deficiency)
    - Beta-ketothiolase deficiency (BKT)
    - Carnitine transporter deficiency (CTD)/carnitine uptake defect (CUD)
    - Citrullinemia type I (argininosuccinic acid synthetase deficiency)/CIT-1
    - Glutaric acidemia type I (GA1)
    - Homocystinuria (cystathionine beta-synthase deficiency, HCY)
    - Isovaleric acidemia (IVA)
    - Long chain hyroxy acyl-CoA dehydrogenase deficiency (LCHAD deficiency)
    - Maple syrup urine disease (MSUD)
    - Medium chain acyl-CoA dehydrogenase deficiency (MCAD Deficiency)
    - Methylmalonic acidemia, Cbl C, D, F (MMA)
    - Methylmalonic acidemia mut 0 (MMA)
    - Multiple carboxylase deficiency (MCD)
    - Propionic acidemia (PA)
    - Trifunctional protein deficiency (TFP Deficiency)
    - Tyrosinemia type I
    - Very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)
- **Severe combined immunodeficiency (SCID)** – includes:
    - SCID
    - Leaky SCID
    - Omenn syndrome
- **Spinal muscular atrophy (SMA)** - Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1
- **X-linked adrenoleukodystrophy (ALD)** – Males with ALD

Data Accuracy

Due to the small number of newborns affected by some conditions, caution should be exercised when making conclusions about the data.