About the Data – Newborn Screening Conditions

Indicator Description

An infant is considered diagnosed with a newborn screening (NBS) condition if they received a positive result on their NBS test and were later confirmed with an NBS-detectable condition. A full list of conditions screened for by the California NBS Program can be found on the Genetic Disease Screening Program website.

Data Source

California Department of Public Health. Screening Information System. Compiled by the Genetic Disease Screening Program. Data Extracted: 1/16/2024.

California Department of Public Health. California Comprehensive Birth File (Dynamic), 2023–2024. Compiled by Center for Health Statistics and Informatics. Date extracted: 1/16/2024.

Data Analysis

The diagnosed rate shown in this dashboard is the number of newborns diagnosed with a screened condition per 100,000 newborns screened. The screening percentage is the number of newborns screened among those born in California. The 95% confidence interval indicates there is a 95% chance that the range contains the true rate in the population. Rates or percentages with wide confidence intervals should be interpreted with caution. Dates are based on when the specimen was accessioned by the state laboratory.

Diagnoses were determined by a specialist from a contracted California Children's Services approved Special Care Center. Diagnoses were grouped into six broad categories, based on the type of Special Care Center the child was referred to: cystic fibrosis, endocrine, immunology, hemoglobin, metabolic, and neuromuscular. The diagnoses were grouped as follows:

- Cystic fibrosis:
    - Cystic fibrosis (does not include cystic fibrosis transmembrane conductance regulator related metabolic syndrome (CRMS))
- Endocrine:
    - Congenital adrenal hyperplasia (includes Salt wasting and Simple virilizing)
    - Congenital hypothyroidism
- Hemoglobin:

- Sickle cell disease (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 hemoglobinopathy (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)

- Immunology
  - Severe combined immunodeficiency (SCID) (includes classic SCID, Leaky SCID, Omenn syndrome)
- Metabolic
  - Amino acid disorder (includes Phenylketonuria (PKU) , Hyperphenylalaninemia, variant, Biopterin disorders, co-factor biosynthesis, Hyperphenylalaninemia, benign, Argininemia/arginase deficiency (ARG), Argininosuccinyl-CoA lyase deficiency (ASAL deficiency), Biopterin disorders, co-factor regeneration, Citrullinemia type I (argininosuccinic acid synthetase deficiency (ASAS deficiency)/CIT-1), Citrullinemia type II (Citrin deficiency/CIT-II), Homocystinuria (cystathionine beta-synthase deficiency, HCY), Homocitrulinuria, hyperornithinenia, hyperammonemia (HHH), Hypermethioninemia (MET/MAT deficiency), Maple syrup urine disease (MSUD), Tyrosinemia type I, Tyrosinemia type II, Tyrosinemia type III, Tyrosinemia, transient, Gyrate atrophy of the choroid and retina, Prolinemia type-I, Prolinemia type-II, Prolinemia, unclassified, Remethylation Defects (MTHFR, MTR, MTRR, Cbl D v1, Cbl G deficiencies), Ornithine transcarbamylase deficiency (OTC deficiency))
  - Biotinidase deficiency (includes Profound biotinidase deficiency and Partial biotinidase deficiency)
  - Fatty acid disorder (includes Carnitine palmitoyl transferase deficiency type I (CPT1 deficiency), Carnitine palmitoyl transferase deficiency type II (CPT2 deficiency), Carnitine transporter deficiency (CTD)/carnitine uptake defect (CUD)

(deep dive), Carnitine-acylcarnitine translocase deficiency (CAT/CACT deficiency), Long chain hyroxy acyl-CoA dehydrogenase deficiency (LCHAD deficiency, Medium chain acyl-CoA dehydrogenase deficiency (MCAD Deficiency), Multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/glutaric acidemia type II (GA2), Short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency), Trifunctional protein deficiency (TFP Deficiency), Very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency) (deep dive), Medium/short chain L-3 hydroxy acyl-CoA dehydrogenase deficiency (M/SCHADD))

- Galactosemia (includes Classic galactosemia and Duarte (D/G) galactosemia)
- Lysosomal storage disorder (includes Severe mucopolysaccharidosis type I (Hurlers), Attenuated mucopolysaccharidosis type I (Scheie and Hurler-Scheie), Mucopolysaccharidosis type I - not otherwise specified, 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)
- Organic acid disorder (includes 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBCD), 3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG CoA lyase deficiency/glutaric aciduria), 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency), 3-Methylglutaconyl-CoA hydratase deficiency type I (MGA), 3-Methylglutaconyl-CoA hydratase deficiency type II (MGA), 3-Methylglutaconyl-CoA hydratase deficiency type III (MGA), 3-Methylglutaconyl-CoA hydratase deficiency type IV (MGA), Beta-ketothiolase deficiency (BKT), Glutaric acidemia type I (GA1), Isobutyryl-CoA dehydrogenase deficiency (IBD/IBG deficiency), Isovaleric acidemia (IVA), Methylmalonic acidemia Cbl A, B (MMA), Methylmalonic acidemia, Cbl C, D, F (MMA), Methylmalonic acidemia mut 0 (MMA), Methylmalonic acidemia mut- (MMA), Multiple carboxylase deficiency (MCD), Propionic acidemia (PA), Malonic aciduria (MA)/malonic acidemia (MAL), 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency (2M3HBA), Ethylmalonic encephalopathy (EE))
- X-linked adrenoleukodystrophy (ALD) (includes males with ALD)

- Neuromuscular
  - Spinal muscular atrophy (includes Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1)

Denominators include all live births that occurred in California. See Category and Subcategory Definitions below for additional inclusion/exclusion criteria.

Data Suppression

The numerator, rate, and confidence interval are not shown if the numerator is less than 6.

Category and Subcategory Definitions

Year: Year in which the newborn's dried blood spot specimen was accessioned by the Genetic Disease Laboratory.

Geography: State or county where the patient's newborn dried blood spot specimen was collected. In the years shown in this dashboard, the following counties did not collect newborn dried blood spot specimens: Alpine, Calaveras, Colusa, Glenn, Modoc, Sierra, Sutter, and Trinity.

Race/ethnicity: Asian/Pacific Islander includes Asian East Indian, Chinese, Japanese, Korean, Cambodian, Laotian, Vietnamese, Filipino, Other Southeast Asian, Hawaiian, Guamanian, and Samoan. When multiple races/ethnicities are selected for each newborn a single race/ethnicity is assigned according to the following priority: American Indian or Alaska Native (AIAN) first, then Black, Hispanic, Asian/Pacific Islander, White, then Other.

Type: The subtypes or subcategories of a newborn screening condition. Endocrine includes congenital adrenal hyperplasia and congenital hypothyroidism. Hemoglobin includes sickle cell disease and other hemoglobinopathy. Immunology includes severe combined immunodeficiency. Metabolic includes amino acid disorder, biotinidase deficiency, fatty acid disorder, galactosemia, lysosomal storage disorder, organic acid disorder, and x-linked adrenoleukodystrophy. Neuromuscular includes spinal muscular atrophy.

Suggested Citation

California Department of Public Health, Center for Family Health, Maternal, Child and Adolescent Health Division in collaboration with the Genetic Disease Screening Program, Newborn Screening Conditions Dashboard, Last Modified June 2024. go.cdph.ca.gov/Newborn-Screening-Conditions-Dashboard