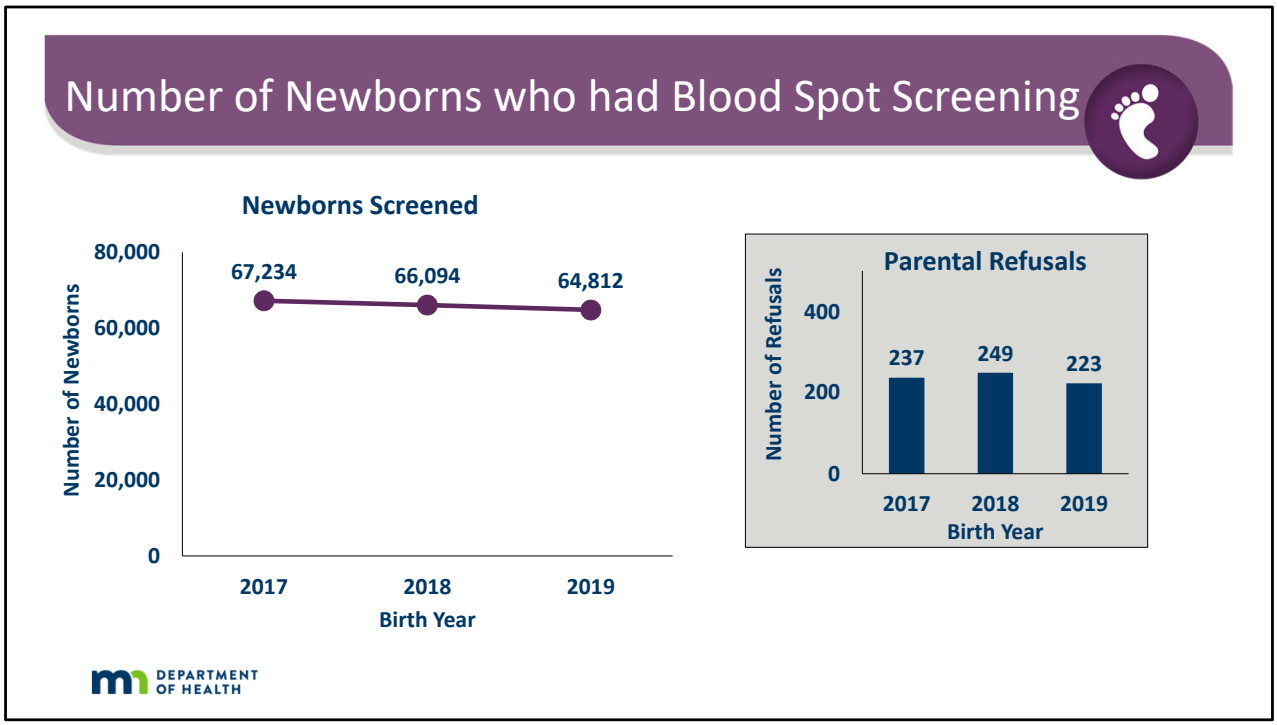
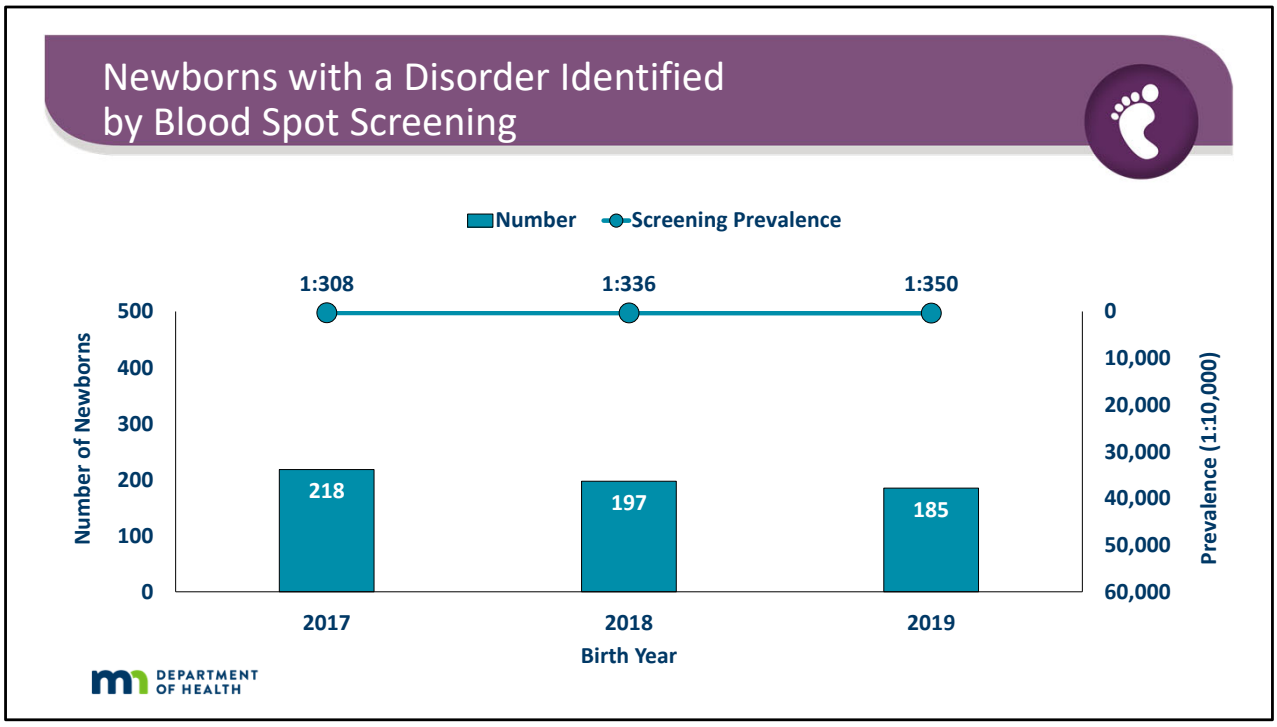
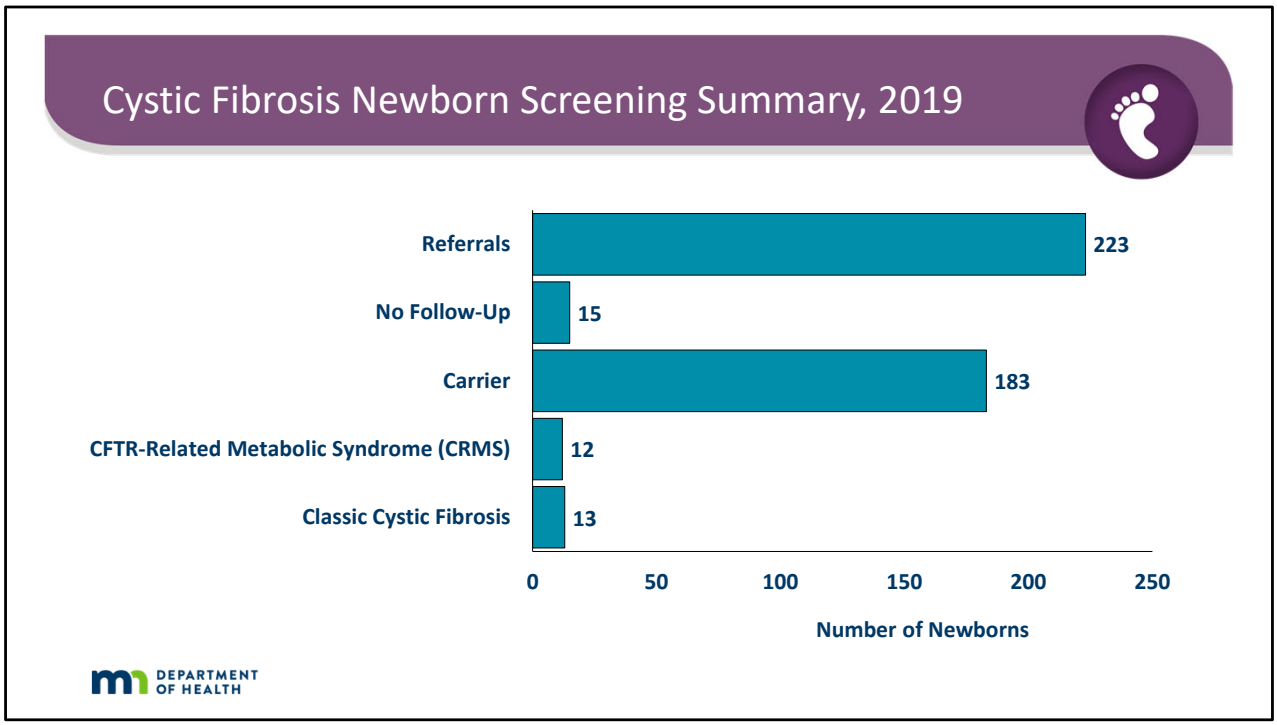
Minnesota Newborn Screening Summary 2017-2019



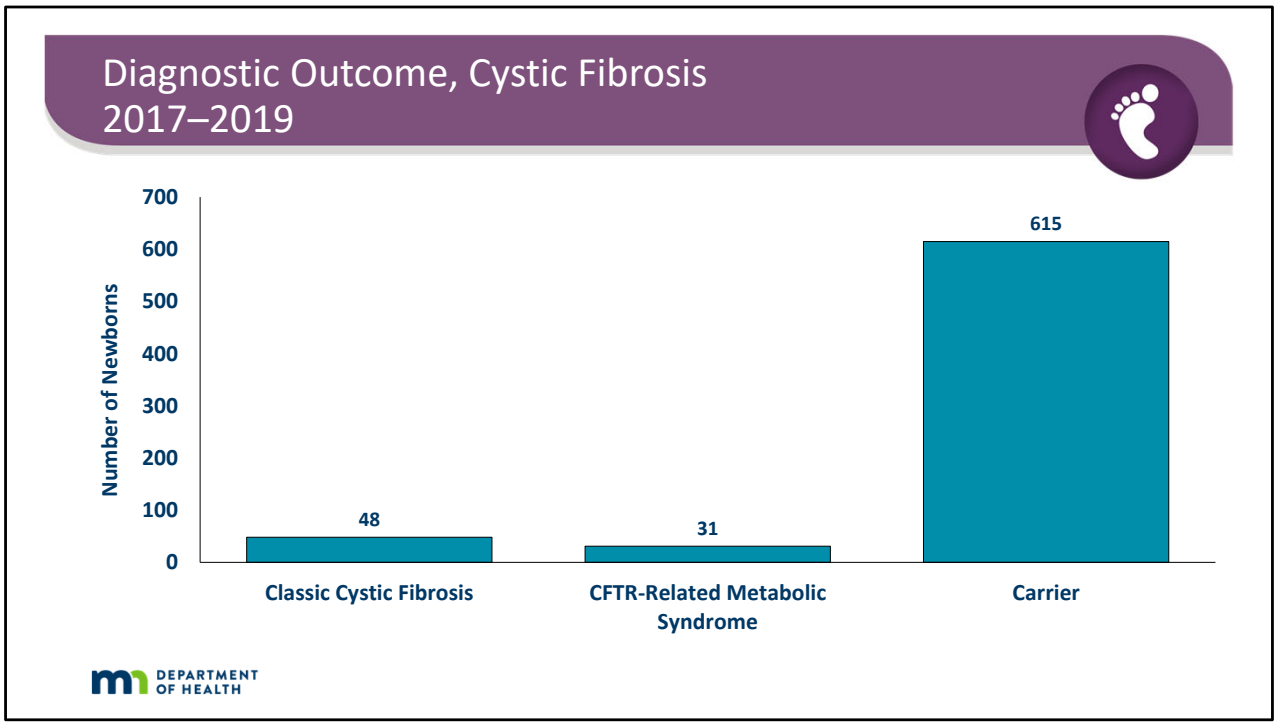
The Minnesota Newborn Screening program screens an average of 66,000 newborns each year. Nearly all (99.6%) infants born in the state are screened. Approximately 236 (<0.4%) parents choose to opt out of screening annually.



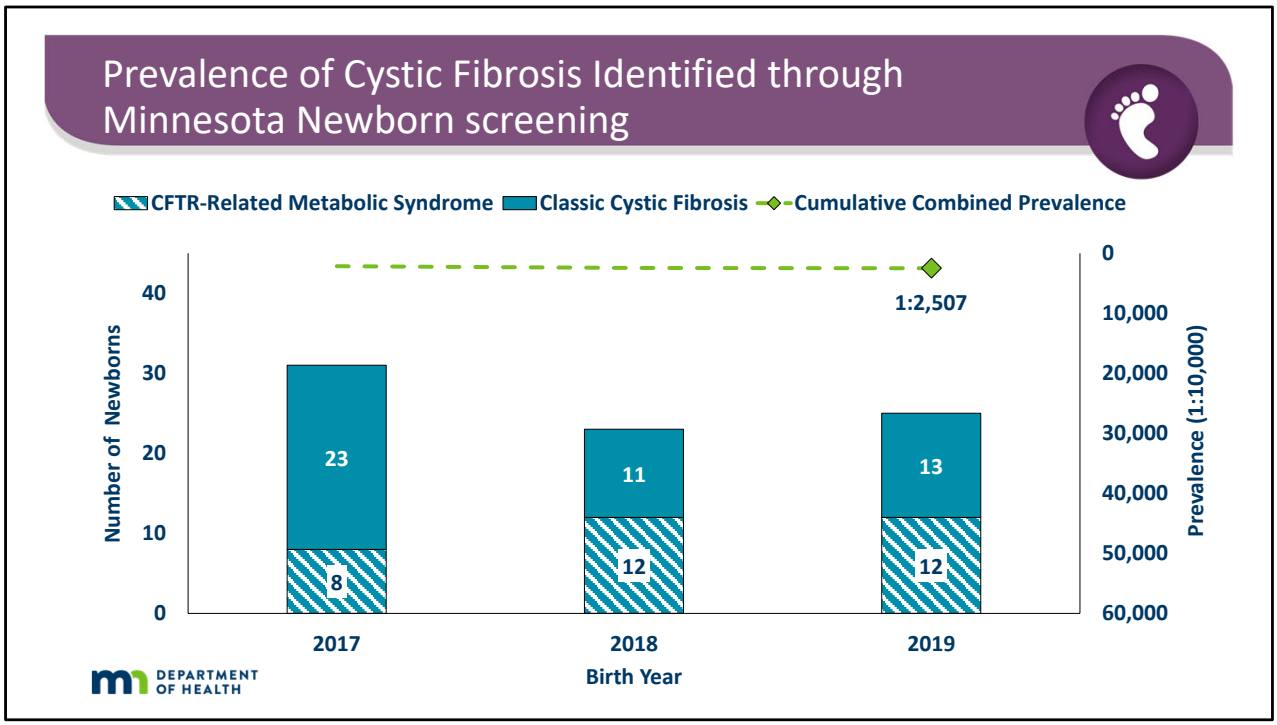
Around 200 infants are identified each year with a condition detected through newborn screening. Screening allows an average of one in every 330 Minnesota infants to receive an otherwise unrecognized diagnosis and benefit from available treatment. Newborn screening prioritizes not missing newborns with a condition on the screening panel, and this can result in false positive results. False positive results have multiple causes including carrier status, overlap between normal and abnormal physiology, environmental impacts on the specimen, and other conditions or treatments in the child or mother.



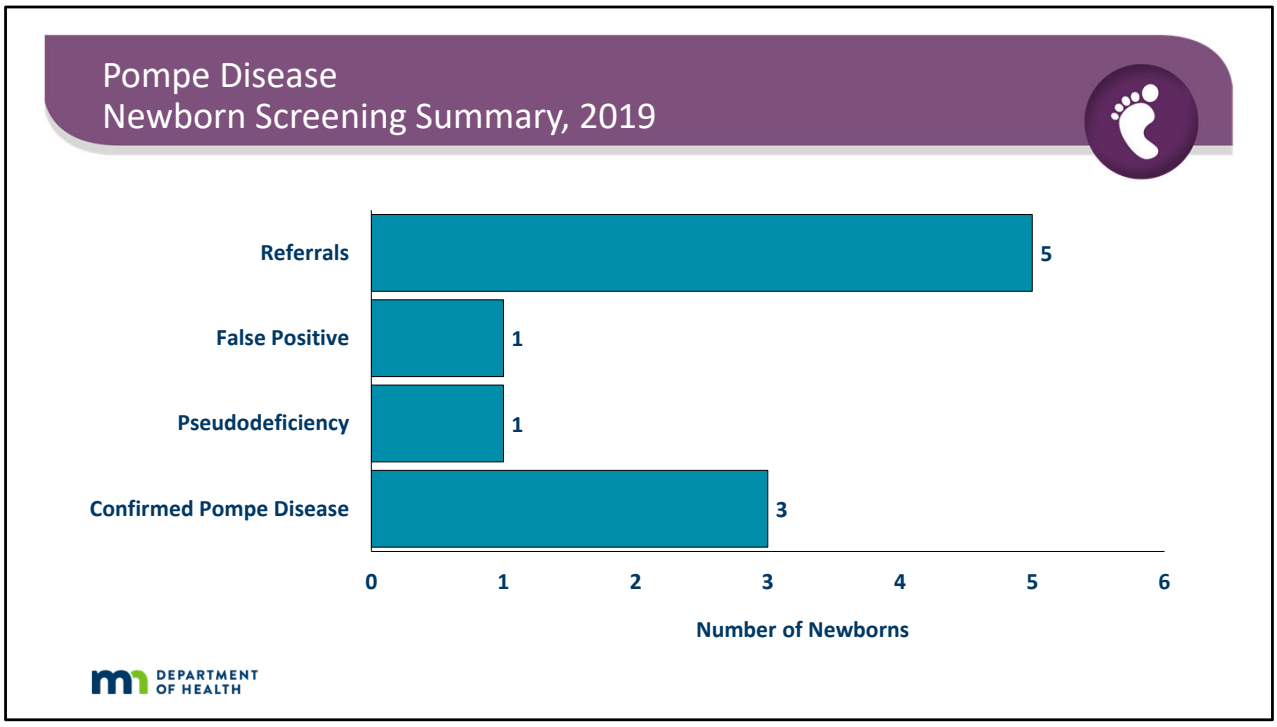
In 2019, 223 newborns had a screening result indicating a risk for cystic fibrosis (CF). Further evaluation found that 25 (11%) infants had either CFTR-related metabolic syndrome (CRMS) or classic CF and 183 (82%) were unaffected carriers.



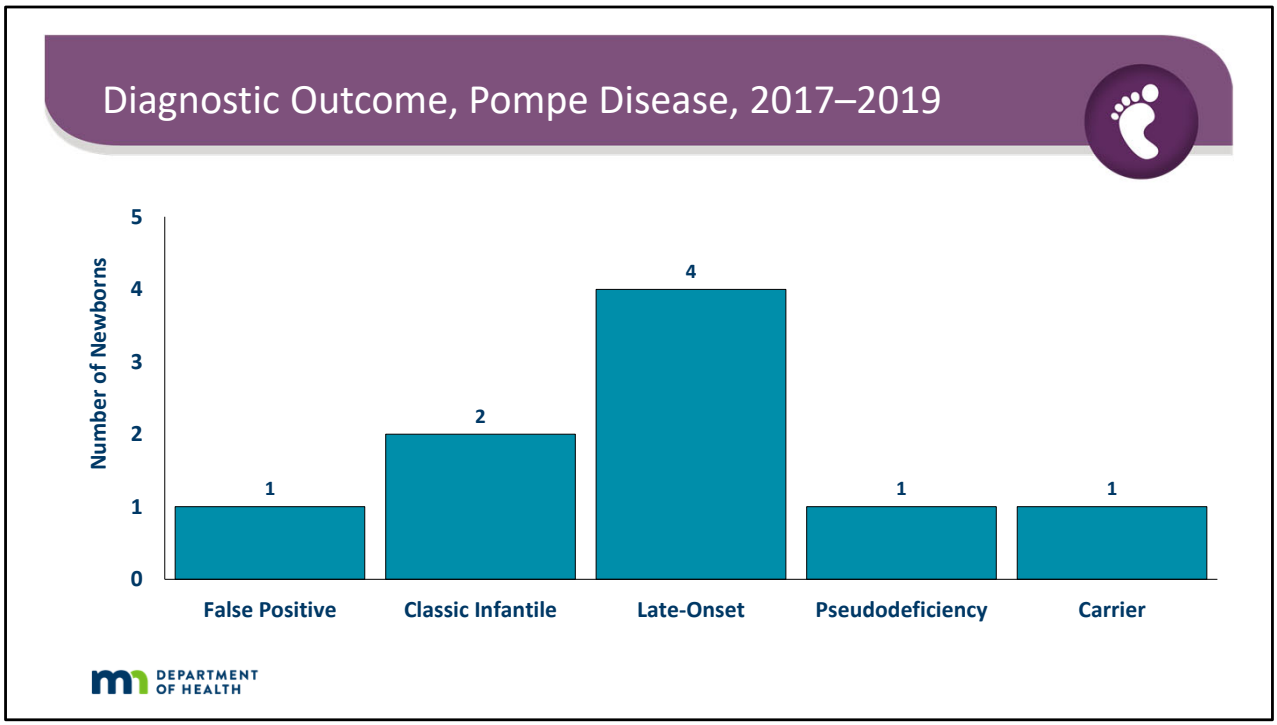
Between 2017 and 2019, 48 (7%) infants were identified with classic CF and 31 (4%) with CRMS. Most infants (615, 89%) with an abnormal CF result on their newborn screen were found to be unaffected carriers.



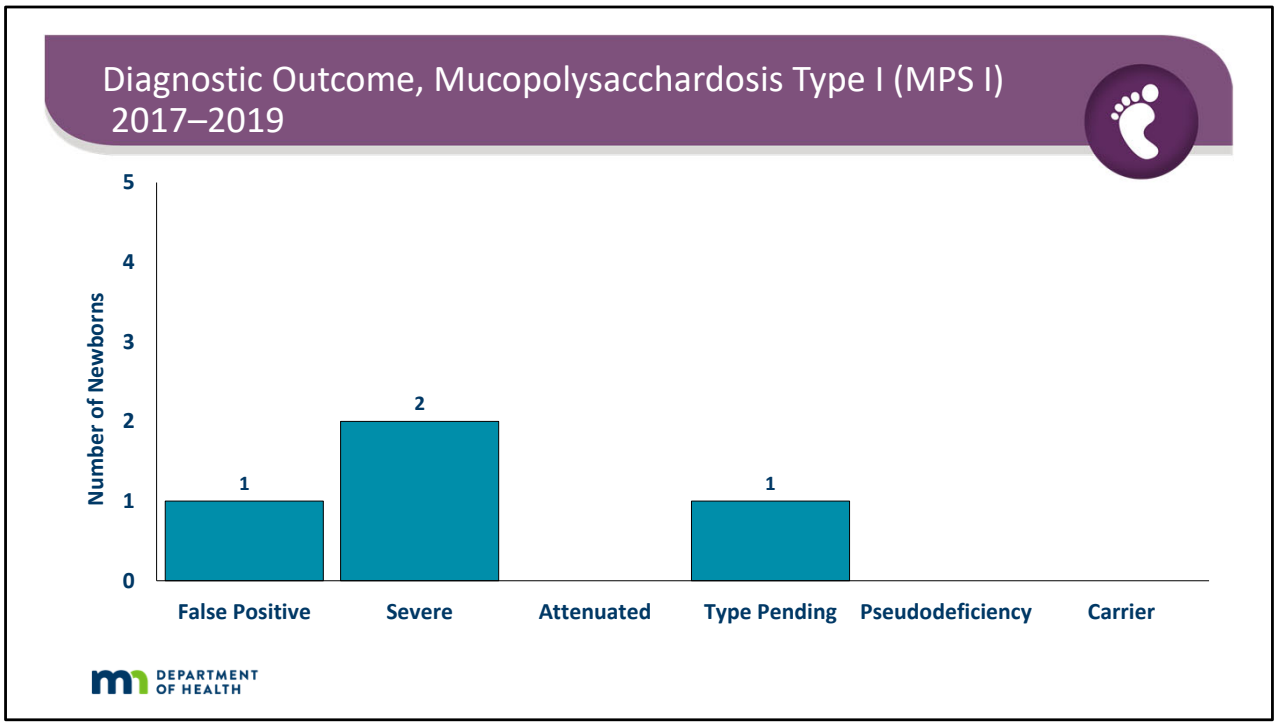
In Minnesota, an average of 16 infants are identified with classic CF and 11 with CRMS each year. This means about one in every 2,500 newborns screened is identified with either classic CF or CRMS.



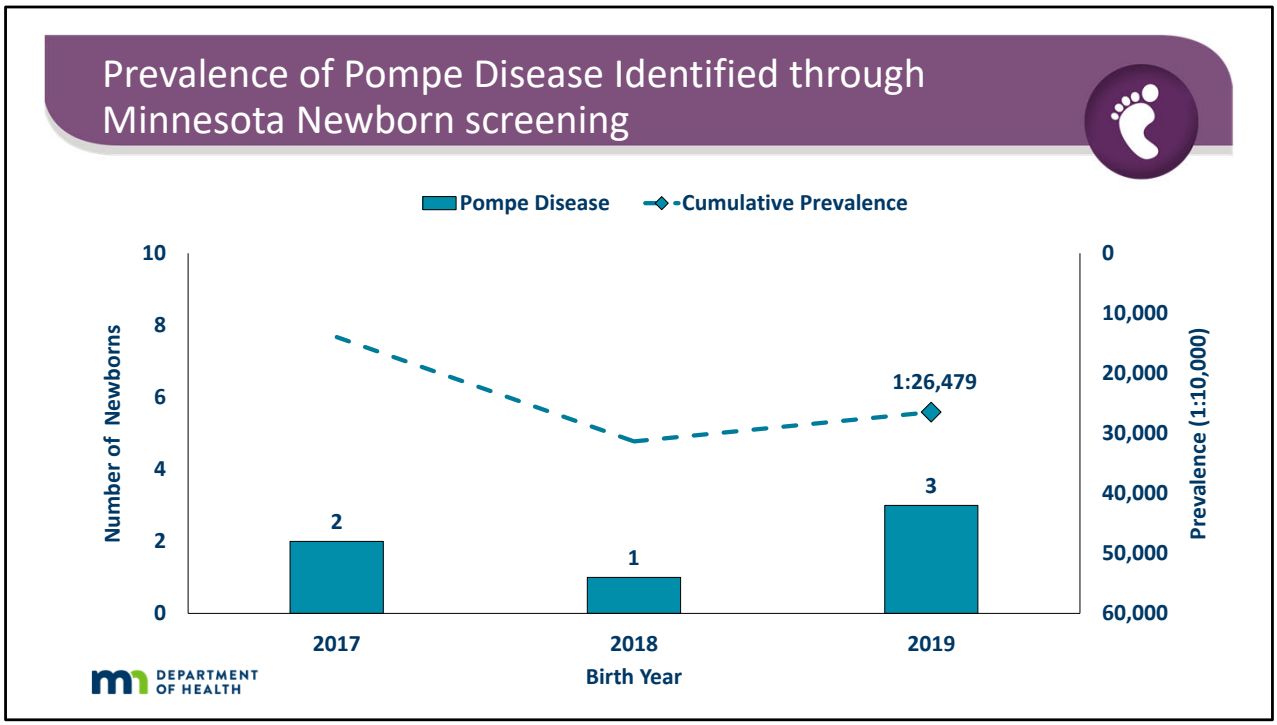
In 2019, five newborns had a screening result indicating a risk for Pompe disease. Of these, three (60%) were confirmed by further evaluation to have the disease.



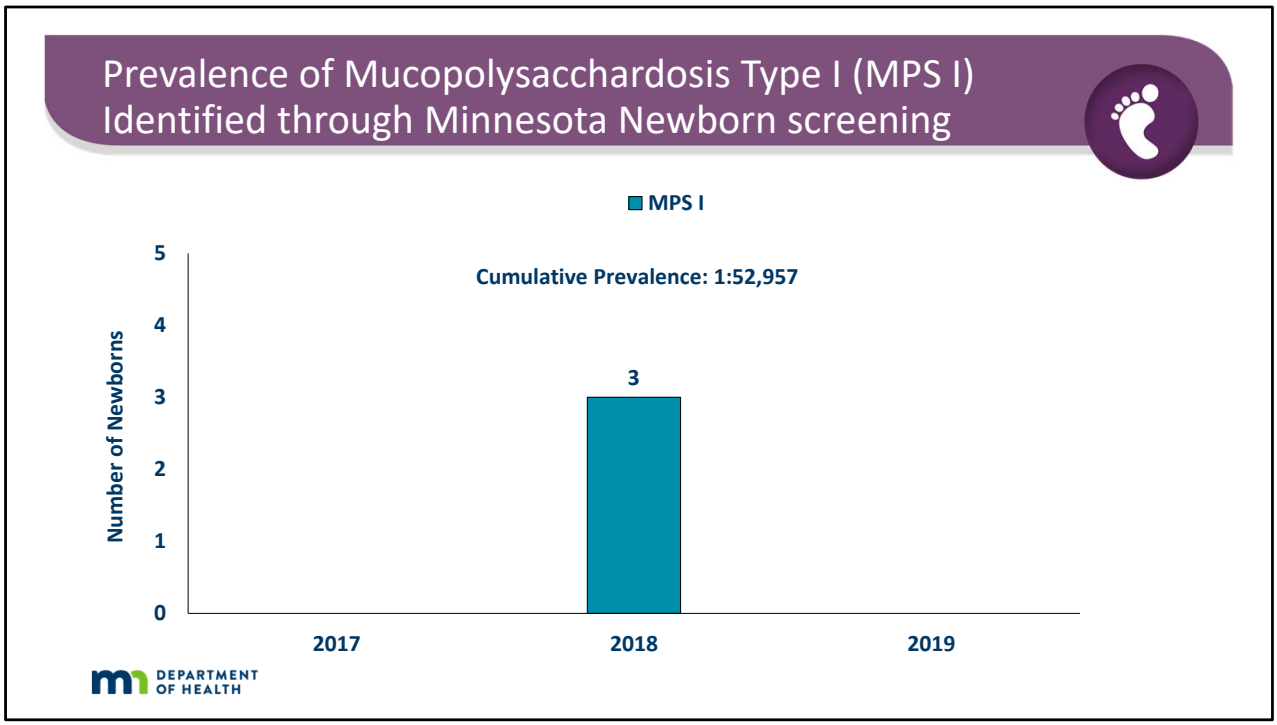
Between 2017 and 2019, six (67%) infants were confirmed to have Pompe disease. Two (22%) infants were identified with the classic infantile type while four (44%) infants have been classified as having late-onset Pompe disease. Newborn screening sometimes identifies infants with a pseudodeficiency (low enzyme activity but otherwise healthy) or unaffected carriers of Pompe disease. One of each of these healthy infants with abnormal screens was found between 2017 and 2019.



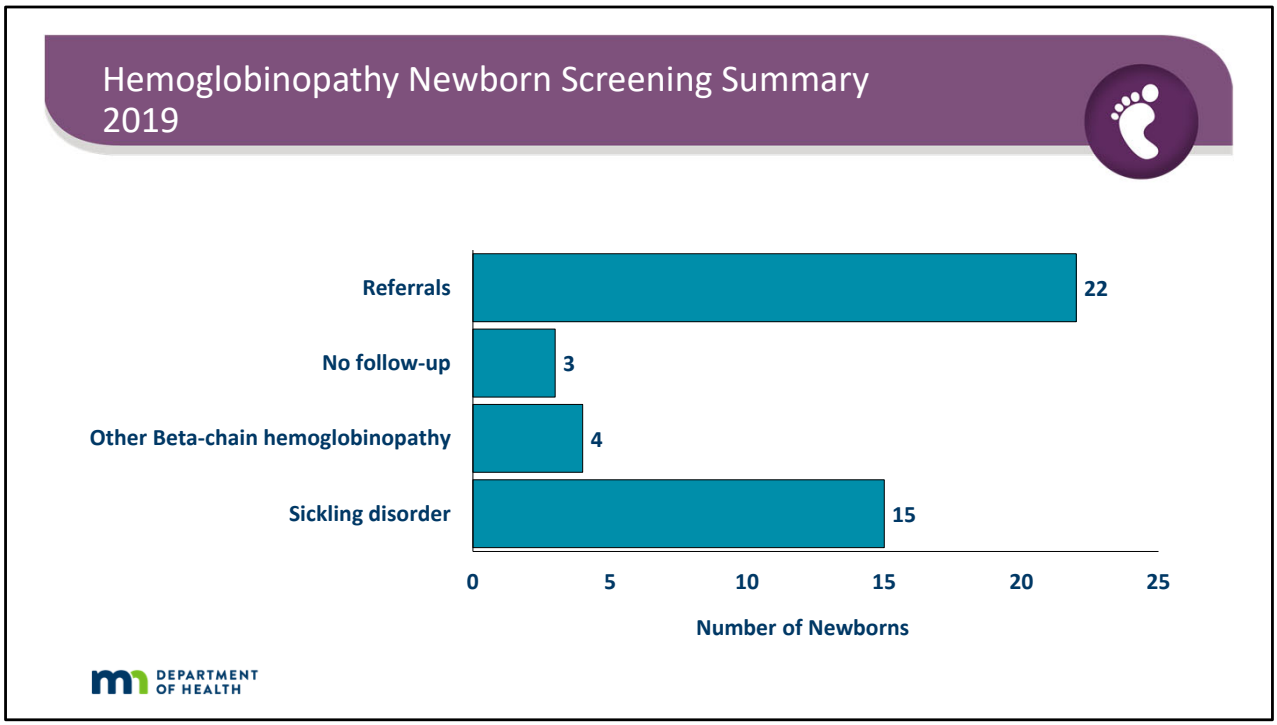
Between 2017 and 2019, three (75%) out of the four infants with an abnormal mucopolysaccharidosis type I (MPS I) screen were confirmed to have the disease. Of these, two (67%) were diagnosed with the severe type and one (33%) was diagnosed with MPS I, but the type was undetermined at the time of confirmation.



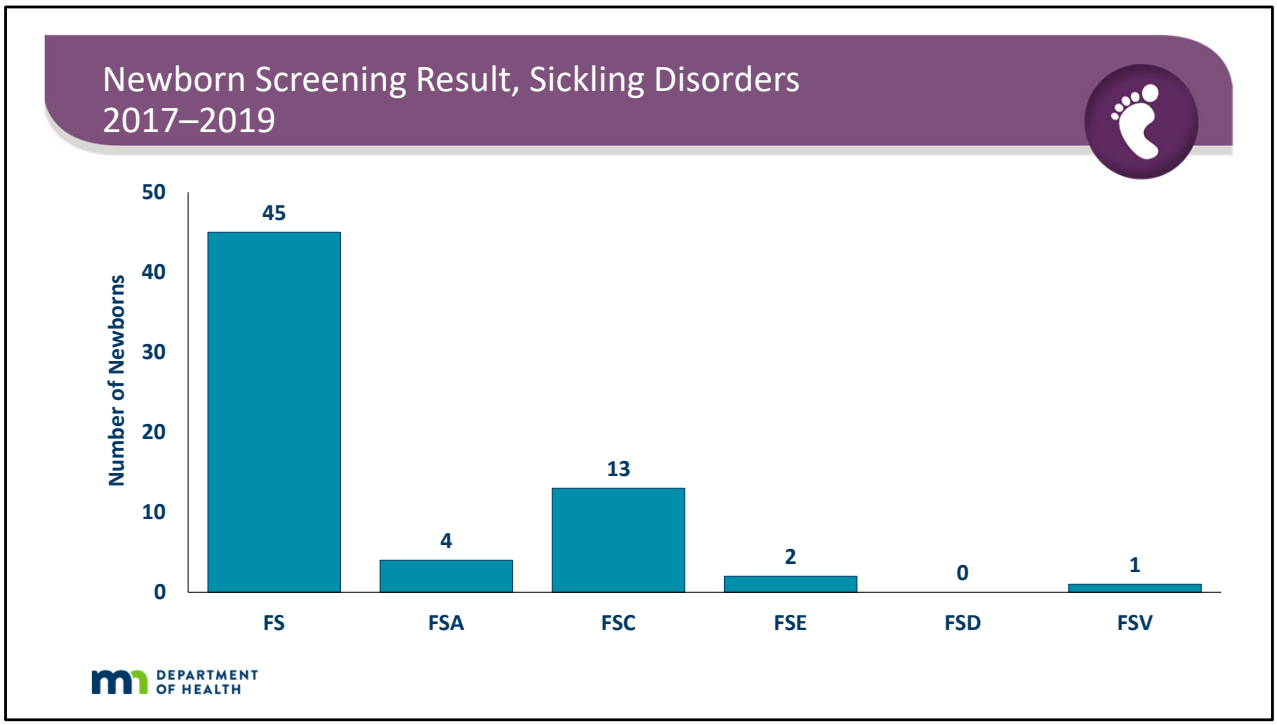
In Minnesota, an average of two infants are identified with Pompe disease each year. This means about one in every 26,000 newborns screened is identified with Pompe disease.



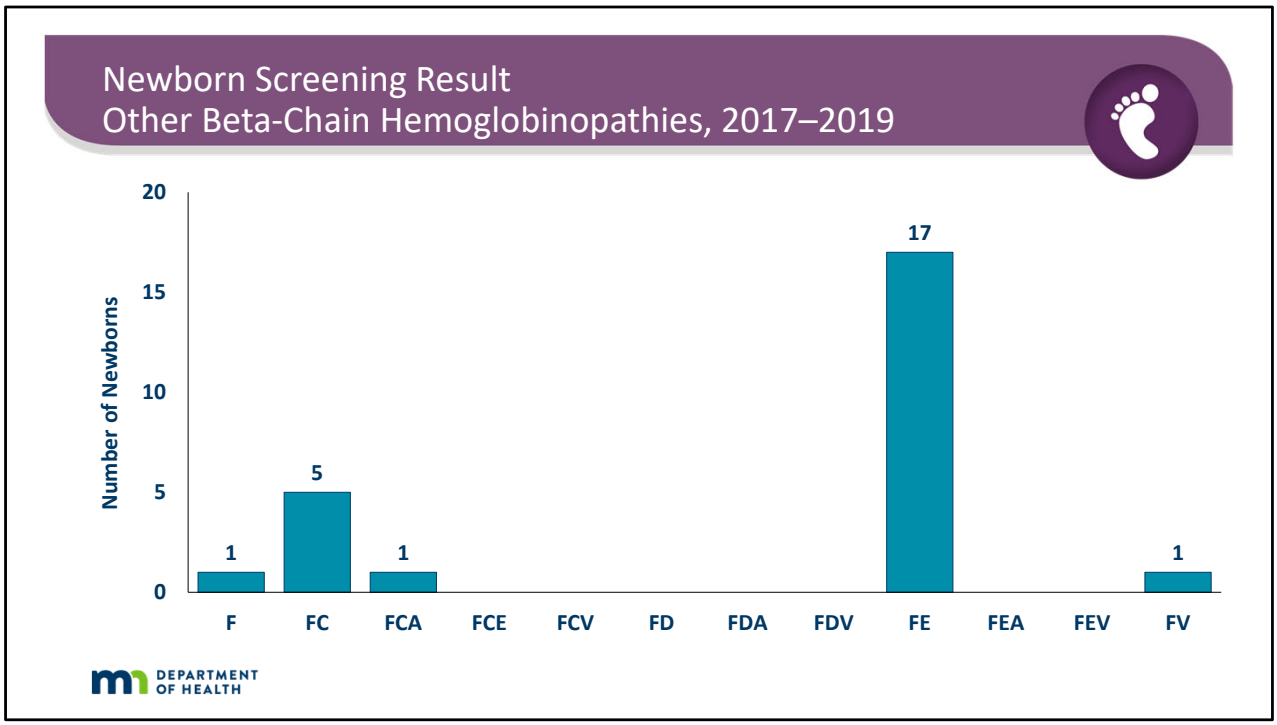
Between zero and three infants are identified by newborn screening with mucopolysaccharidosis type I annually. This means about one in every 53,000 newborns screened is identified MPS I.



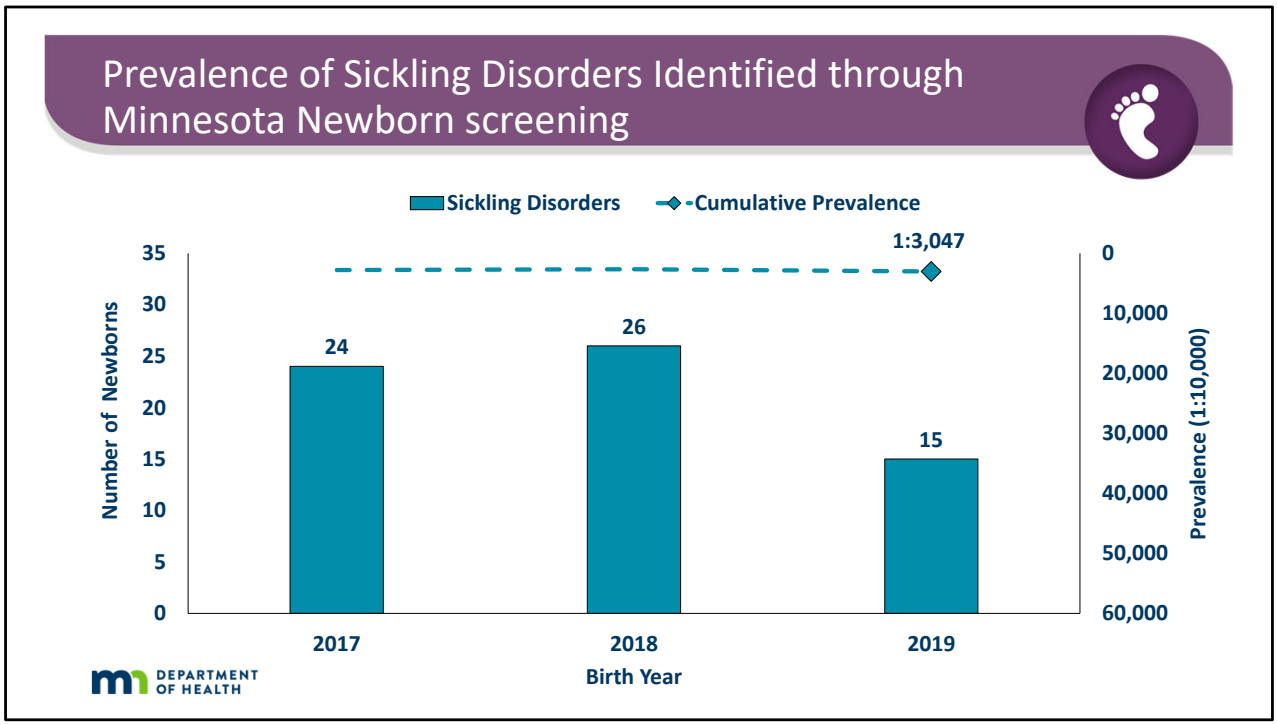
In 2019, 22 newborns had a screening result indicating a risk for a hemoglobinopathy. Of these, 15 (68%) were confirmed by further evaluation to have a sickling disorder and four (18%) were confirmed to have another beta-chain hemoglobinopathy. The remaining three infants were lost to follow-up.



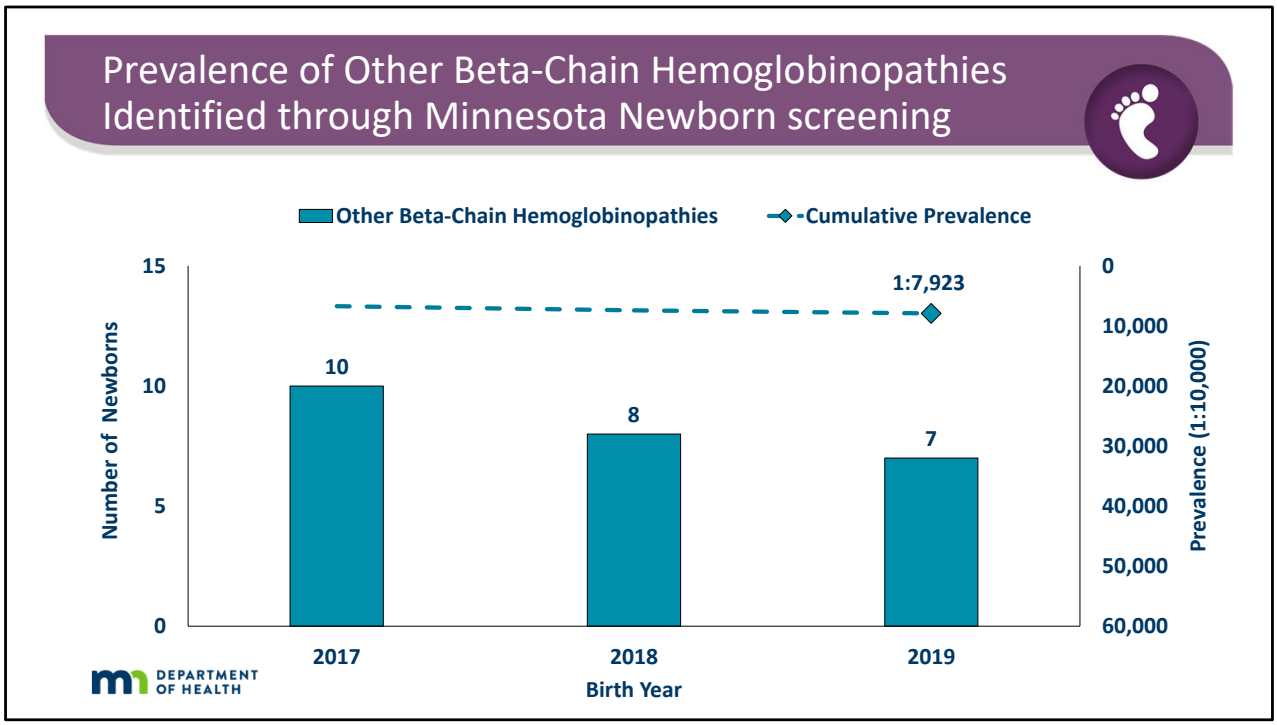
Between 2017 and 2019, most newborns (45, 69%) identified with a sickling disorder had an FS result on their newborn screen. Infants with an FS result are missing normal hemoglobin (hemoglobin A), a result typically associated with sickle cell disease. The next most common result was FSC (13, 20%); infants with this result are typically diagnosed with sickle C disease.



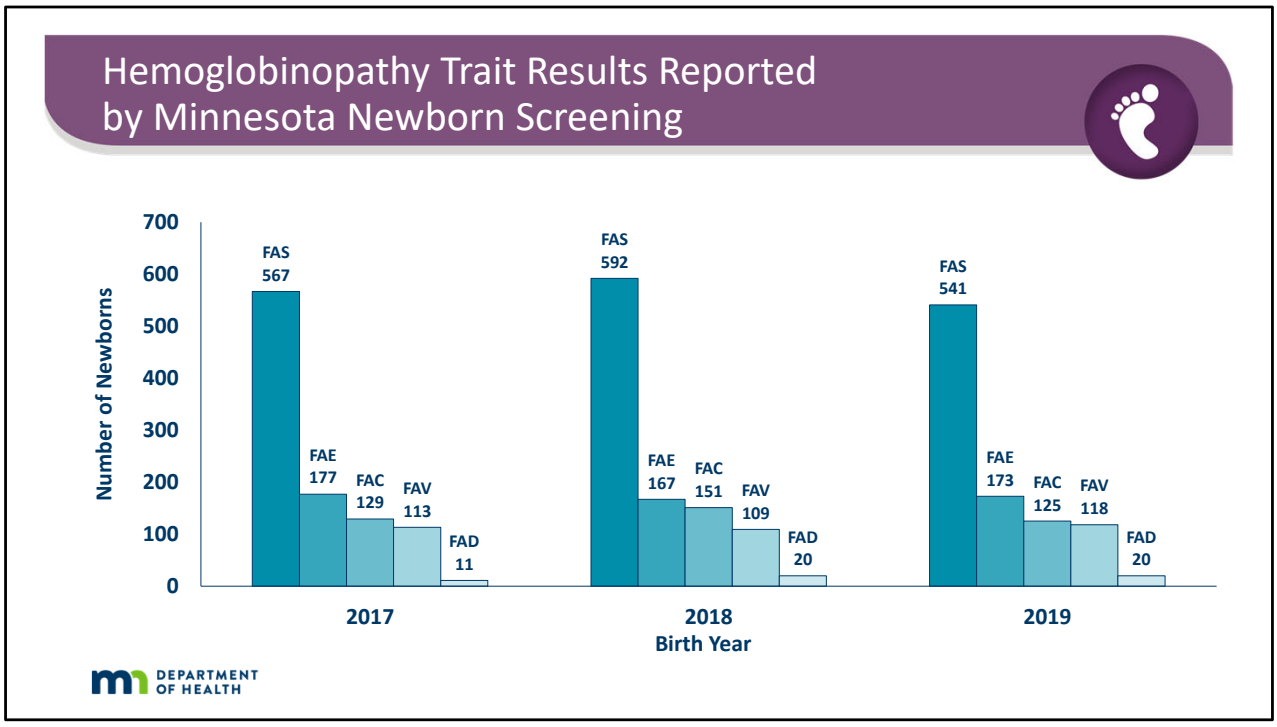
Between 2017 and 2019, most infants (17, 68%) identified with another beta-chain hemoglobinopathy had an FE result on their newborn screen. Infants with an FE result have hemoglobin E substituted for normal hemoglobin (hemoglobin A), a result typically associated with hemoglobin E disease.



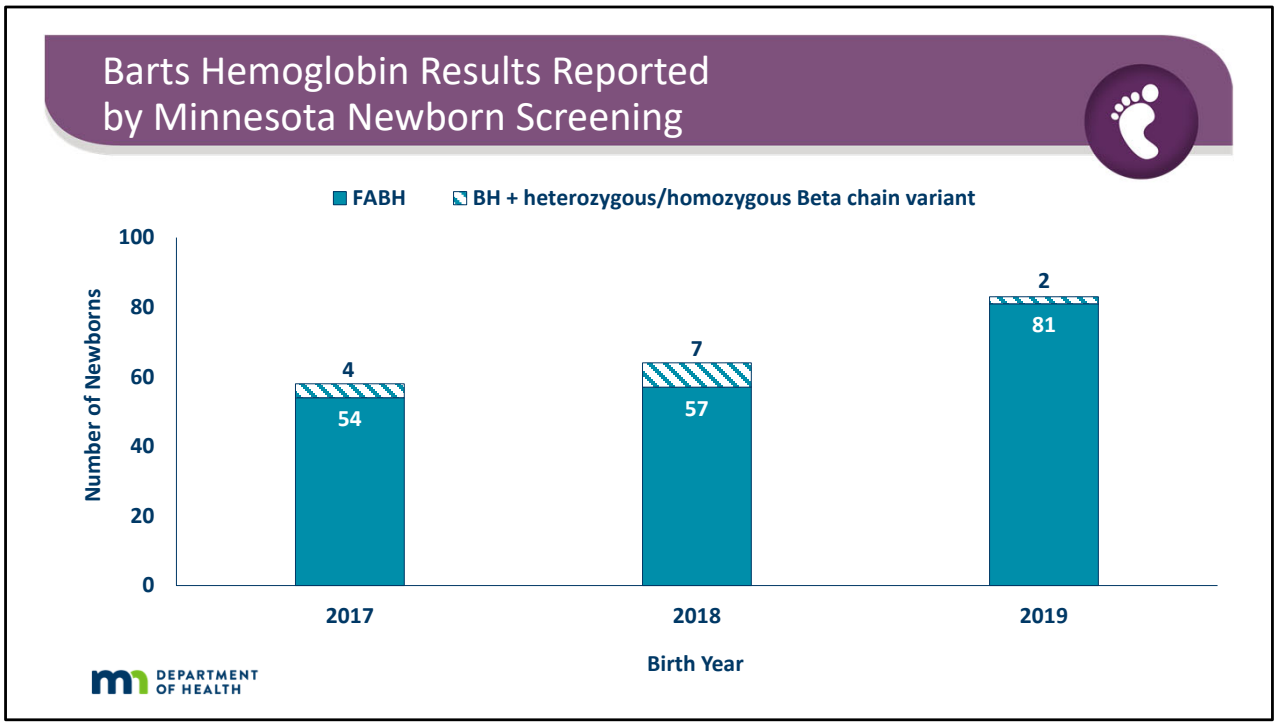
In Minnesota, an average of 22 infants are identified with a sickling disorder each year; this is about one in every 3,000 newborns screened.



In Minnesota, an average of eight infants are identified with another beta-chain hemoglobinopathy each year; this is about one in every 8,000 newborns screened.

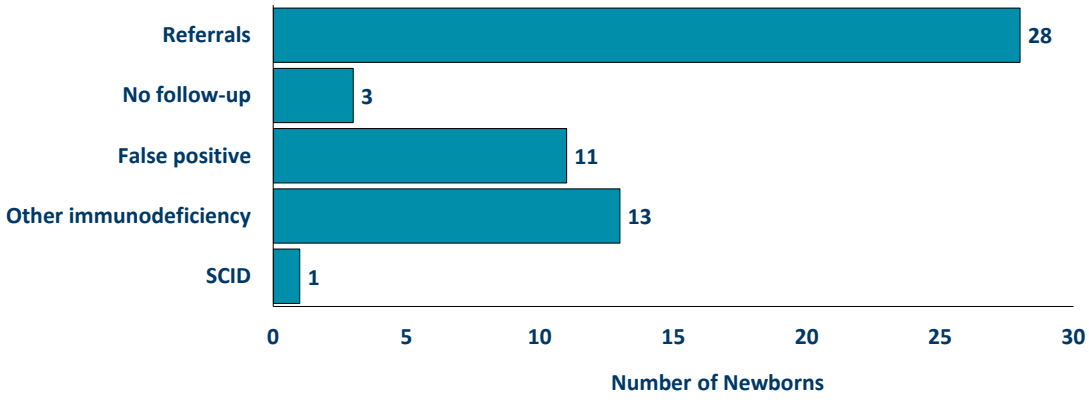


The most common hemoglobin trait found on newborn screening is sickle cell trait (FAS), with an average of 567 newborns identified annually. Infants with a hemoglobin trait are healthy and do not have hemoglobin disease since they have normal hemoglobin (hemoglobin A) present.



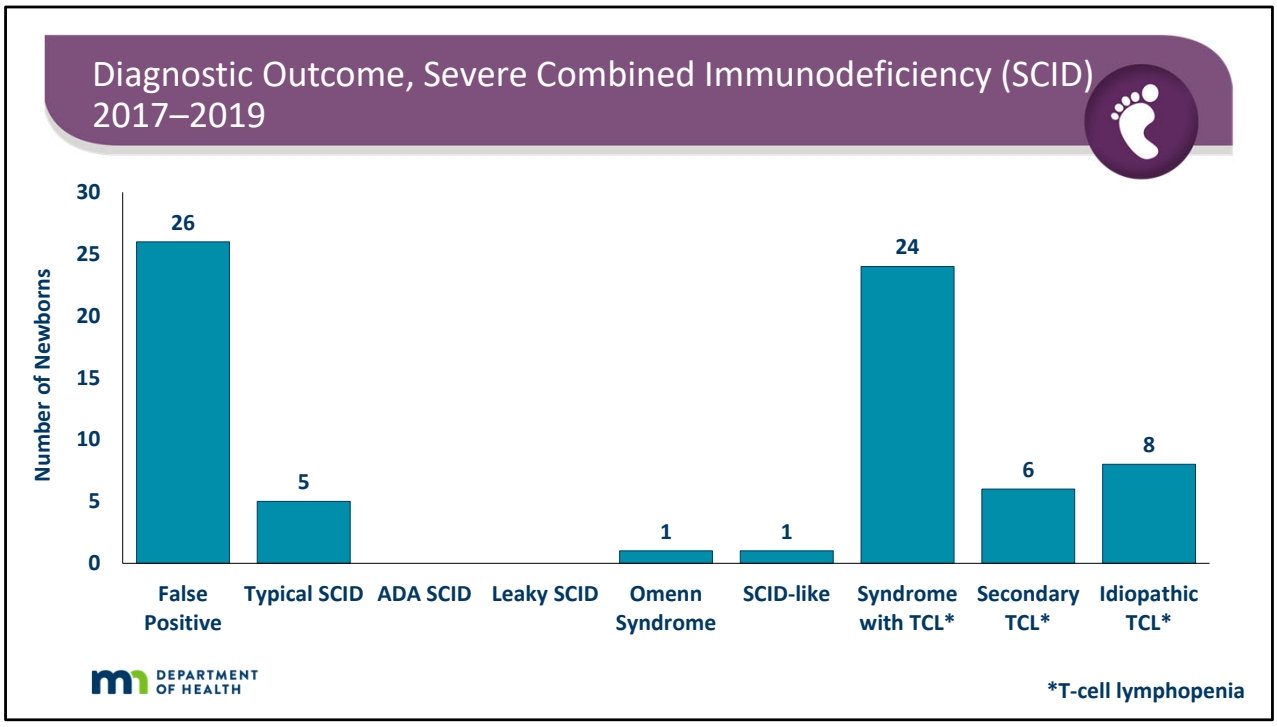
Between 2017 and 2019, a total of 192 (an average of 64 annually) newborns were identified with Barts hemoglobin, and an additional 13 (an average of 4 annually) newborns were identified with Barts hemoglobin and another beta-chain variant (such as hemoglobin S). Barts hemoglobin in the newborn period is associated with types of alpha thalassemia.

Severe Combined Immunodeficiency (SCID) Newborn Screening Summary, 2019

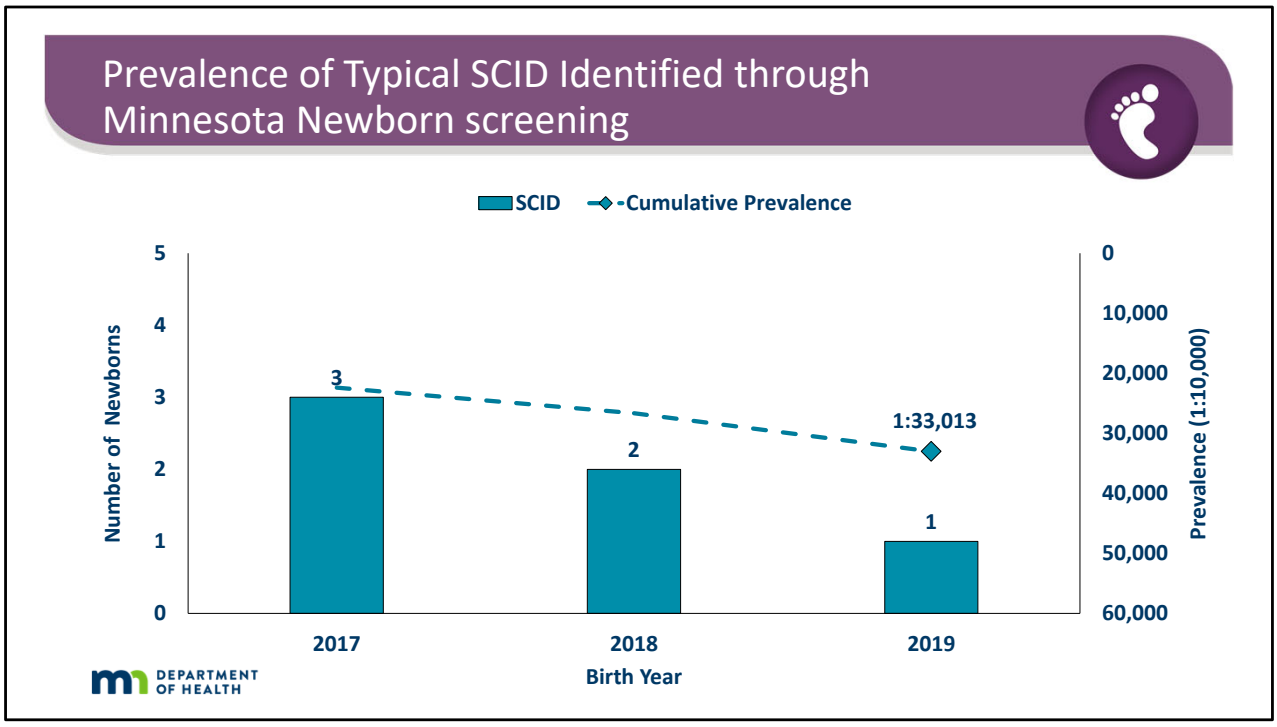


DEPARTMENT OF HEALTH

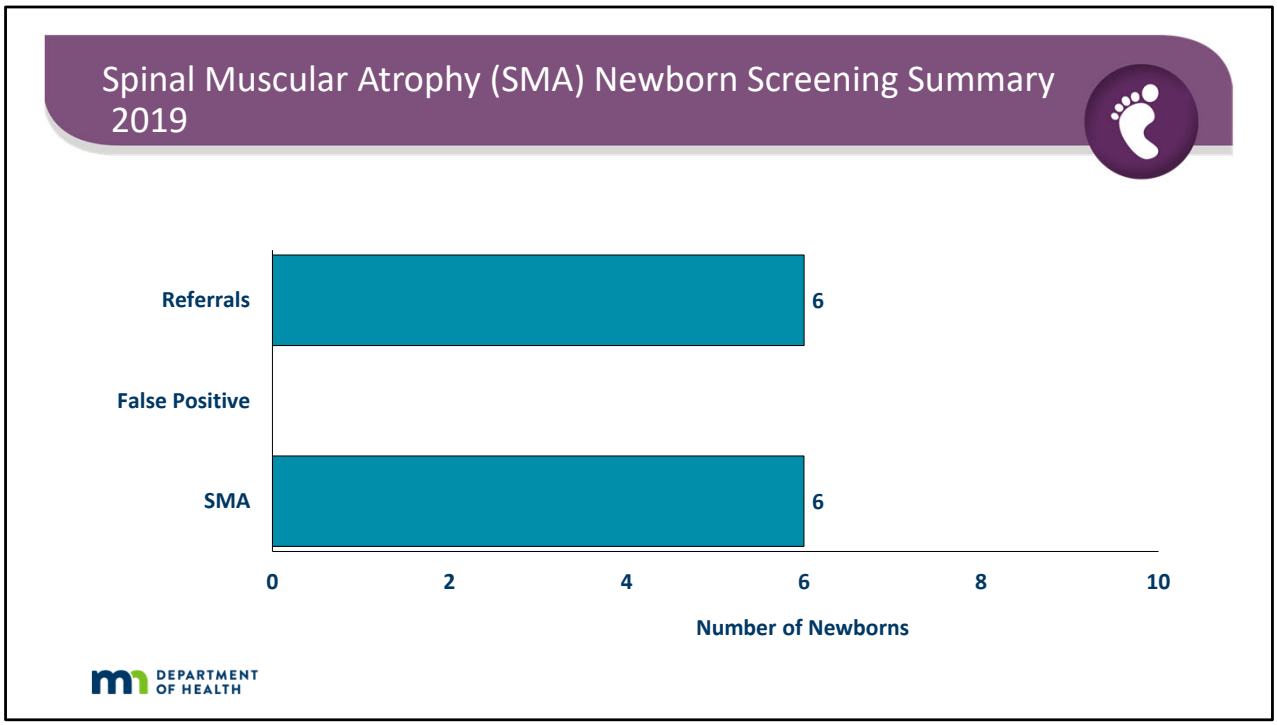
In 2019, 28 newborns had a screening result indicating a risk for severe combined immunodeficiency (SCID) disease. Of these, 1 (4%) was confirmed to have typical SCID and 13 (46%) were diagnosed with another cause for immunodeficiency (such as 22q11.2 deletion syndrome/DiGeorge syndrome).



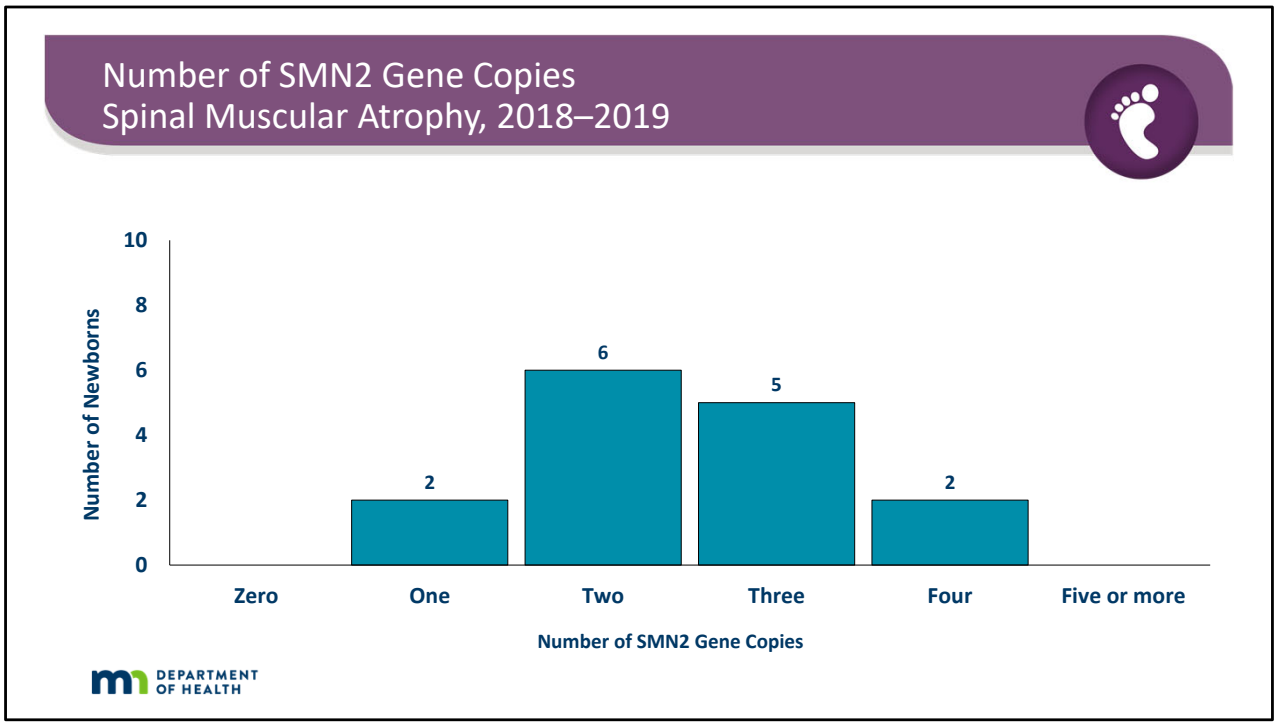
Between 2017 and 2019, the most common diagnosis for infants with an abnormal severe combined immunodeficiency (SCID) result on their newborn screen was a syndrome with T-cell lymphopenia (24, 53%). Only five (11%) were diagnosed with typical SCID. Additionally, the results of over two-thirds (26, 37%) of infants with an abnormal SCID newborn screen are determined to be false positive, commonly due to prematurity.



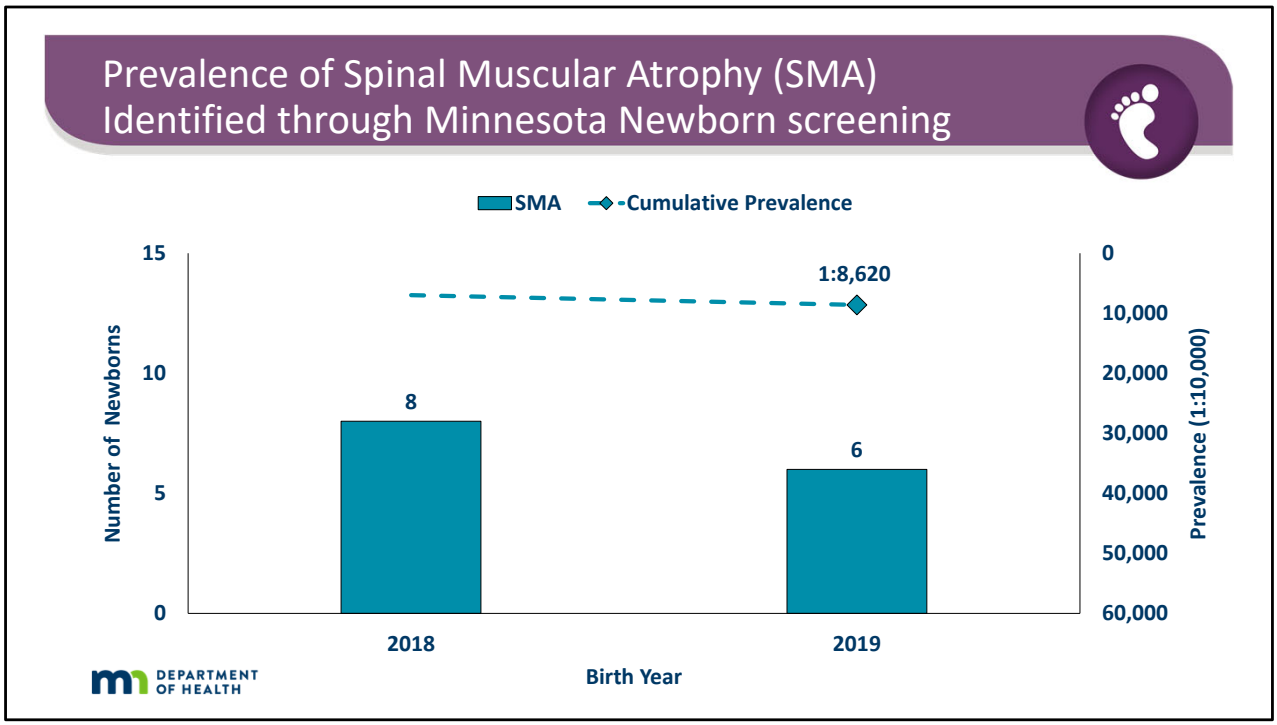
In Minnesota, an average of two infants are identified with typical SCID each year; this is about one in every 33,000 newborns screened.



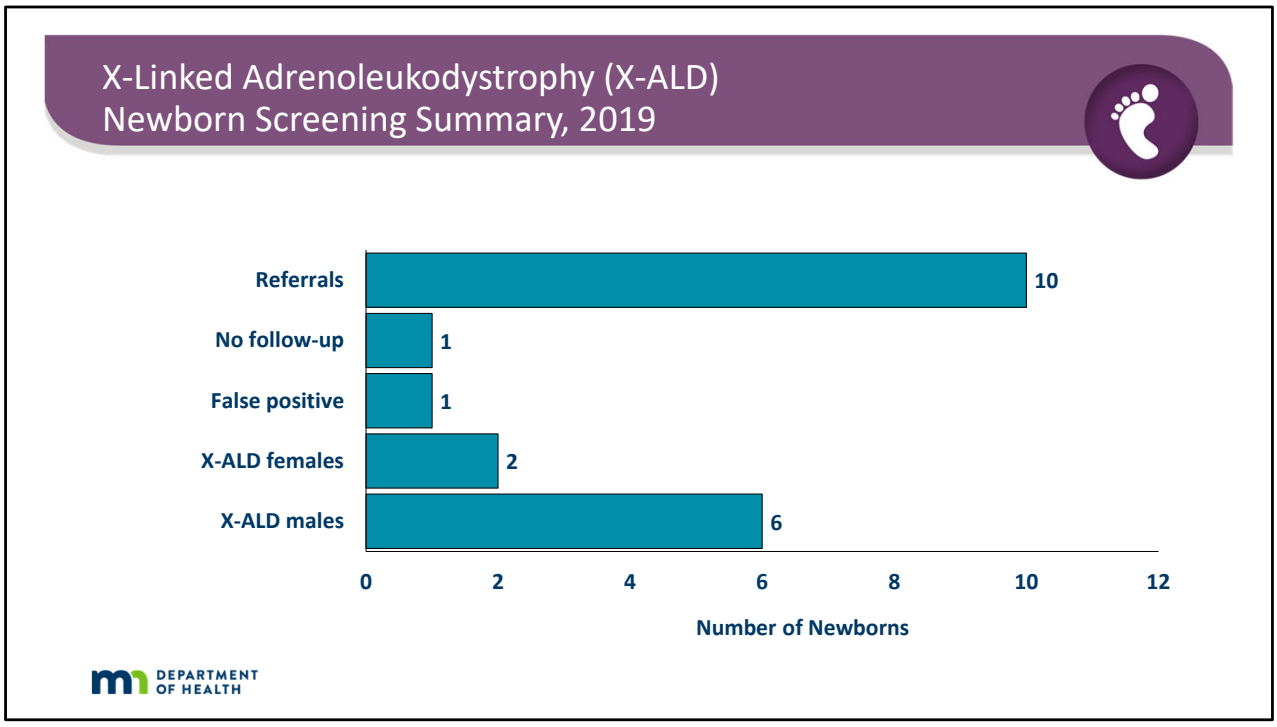
In 2019, six newborns had a screening result indicating a risk for spinal muscular atrophy (SMA). All (100%) of these newborns were confirmed to have SMA.



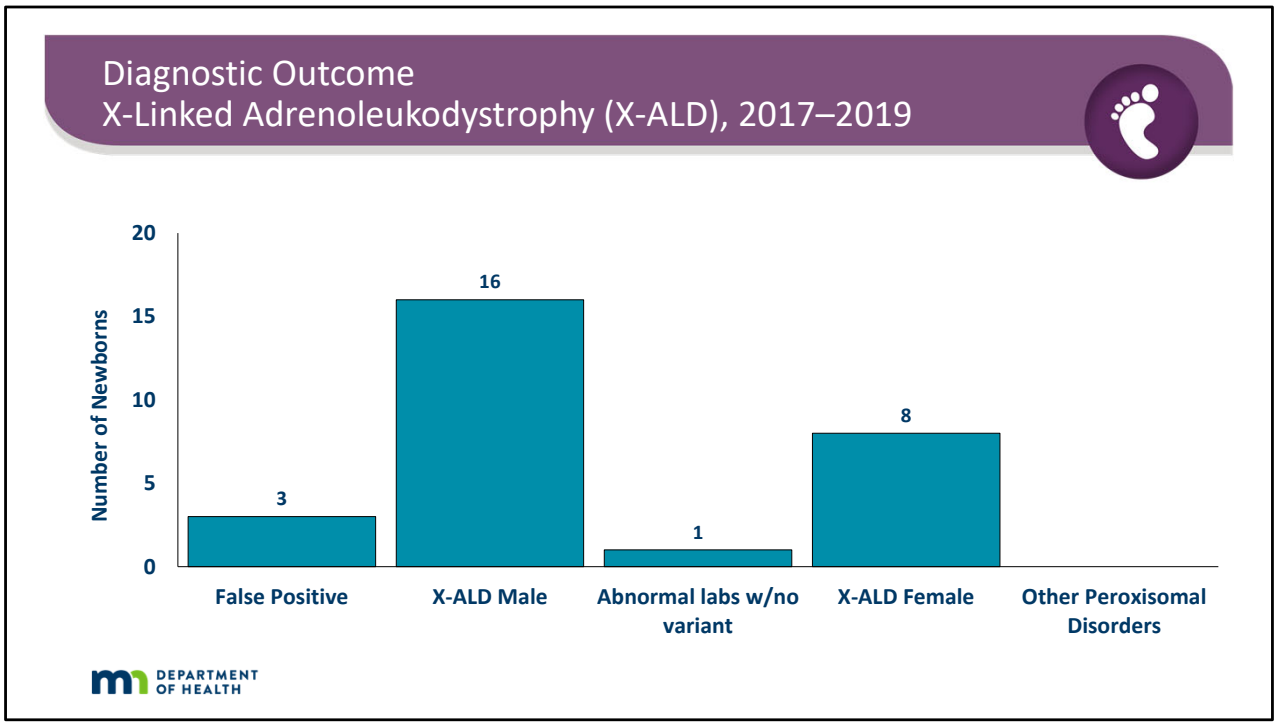
For individuals with spinal muscular atrophy (SMA), the number of SMN2 gene copies impacts the severity of the disease. The more SMN2 gene copies there are, the less severe the symptoms. Infants with the most severe type of SMA typically have three or fewer copies of the SMN2 gene. Between 2018 and 2019, most (73%) of the infants with confirmed SMA were found to have two or three copies of the SMN2 gene.



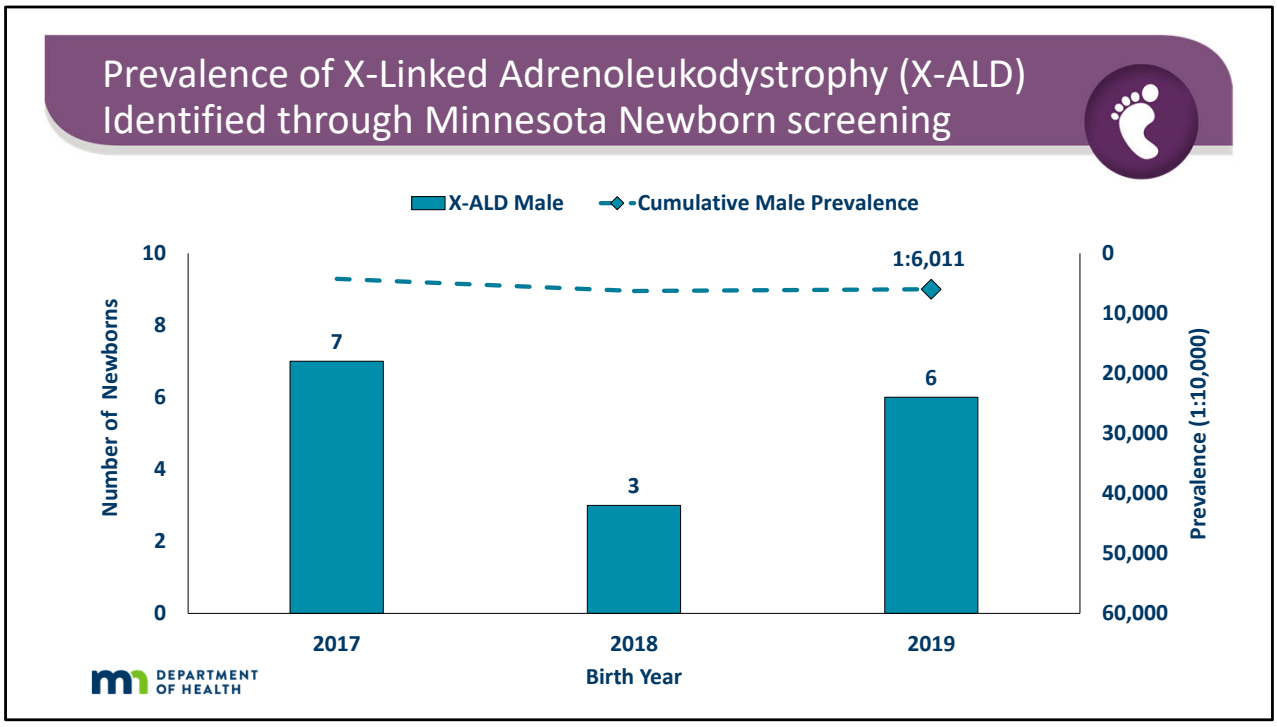
In Minnesota, an average of five infants are identified with spinal muscular atrophy (SMA) each year. This means about one in every 8,600 newborns screened is identified with SMA.



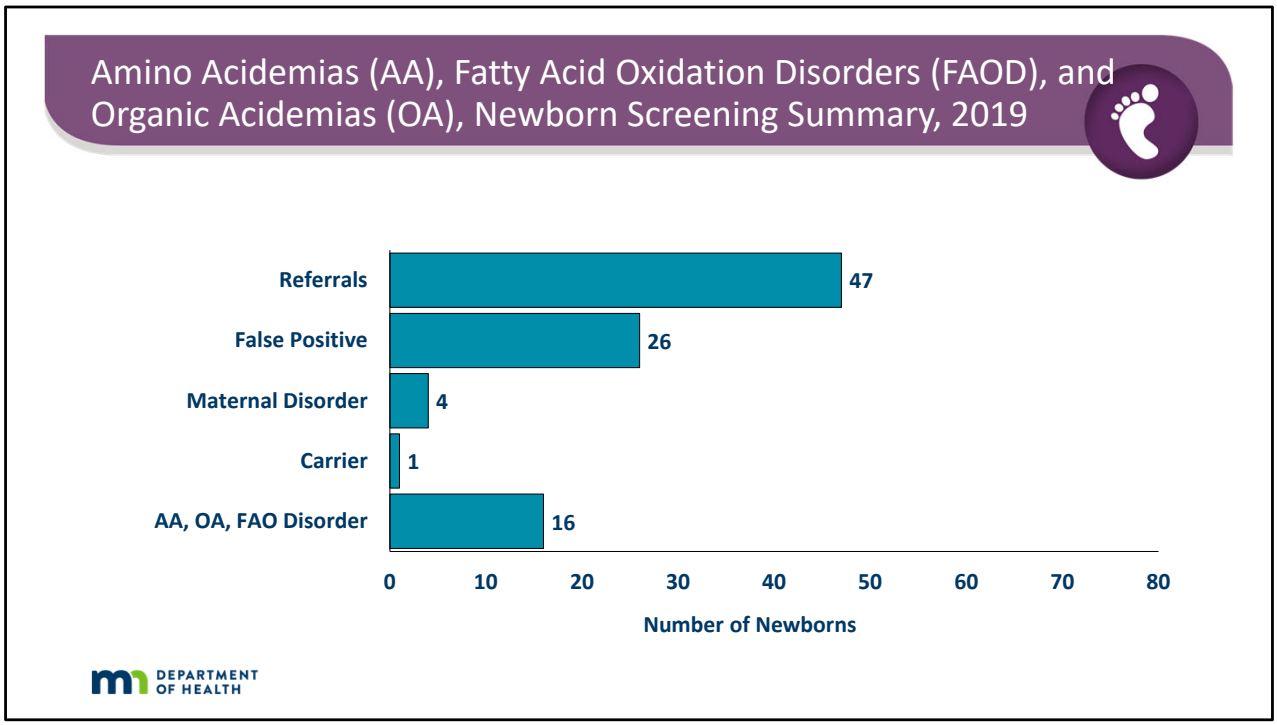
In 2019, 10 newborns had a screening result indicating a risk for X-linked adrenoleukodystrophy (X-ALD). Of these, six (60%) were confirmed males with X-ALD and two (20%) were confirmed females with X-ALD.



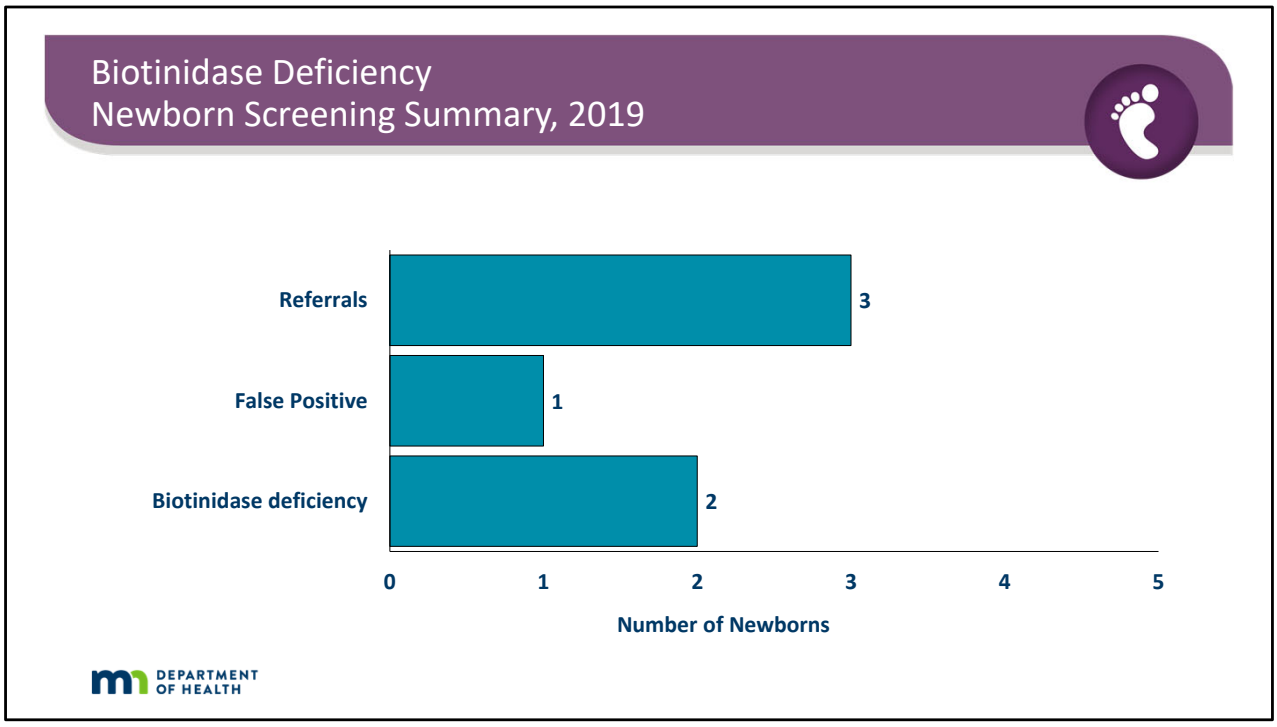
Between 2017 and 2019, 24 (86%) of 28 babies with an abnormal X-linked adrenoleukodystrophy (X-ALD) result on their newborn screen confirmed. Of these, 16 (67%) were males and eight (33%) were females.



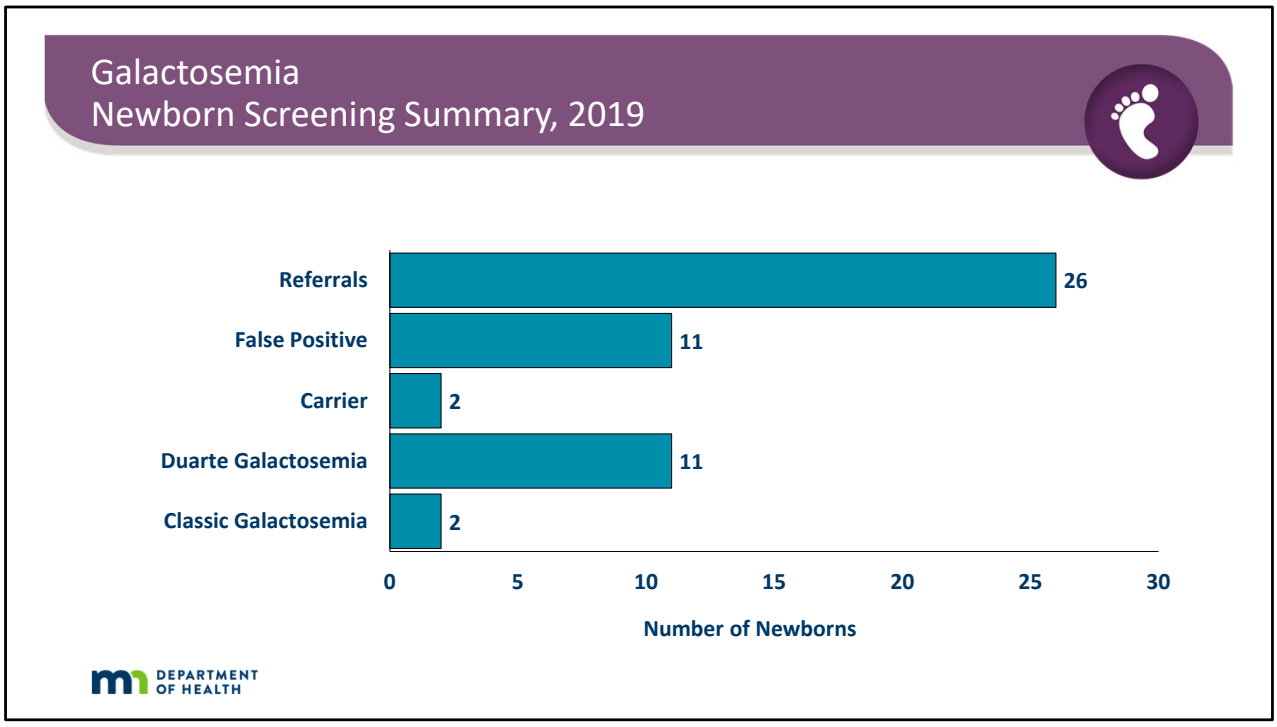
In Minnesota, an average of five male infants are identified with X-linked adrenoleukodystrophy (X-ALD) each year. This means about one in every 6,000 male newborns screened is identified with X-ALD.



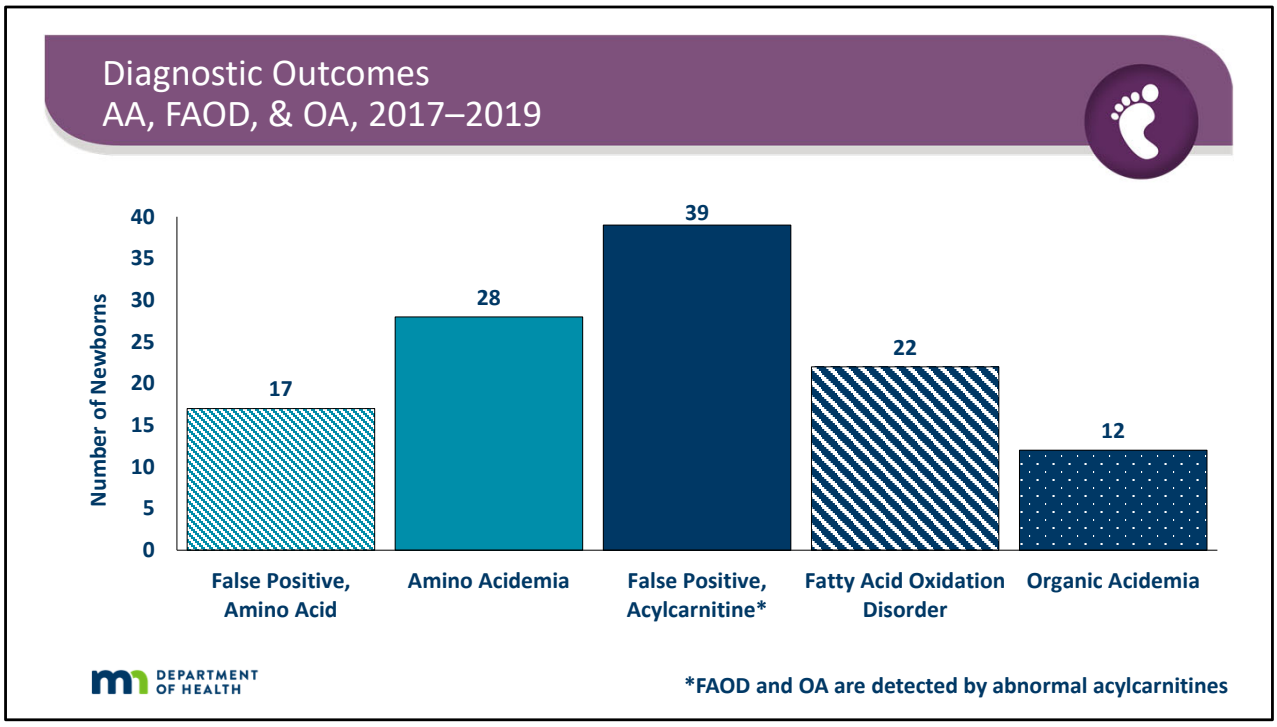
In 2019, 47 newborns had a screening result indicating a risk for an amino acidemia, fatty acid oxidation disorder, or organic acidemia. Of the 47 positive results, 16 (34%) infants were confirmed by further evaluation to have a condition from one of those three disorder groups, while over half were determined to be false positive.



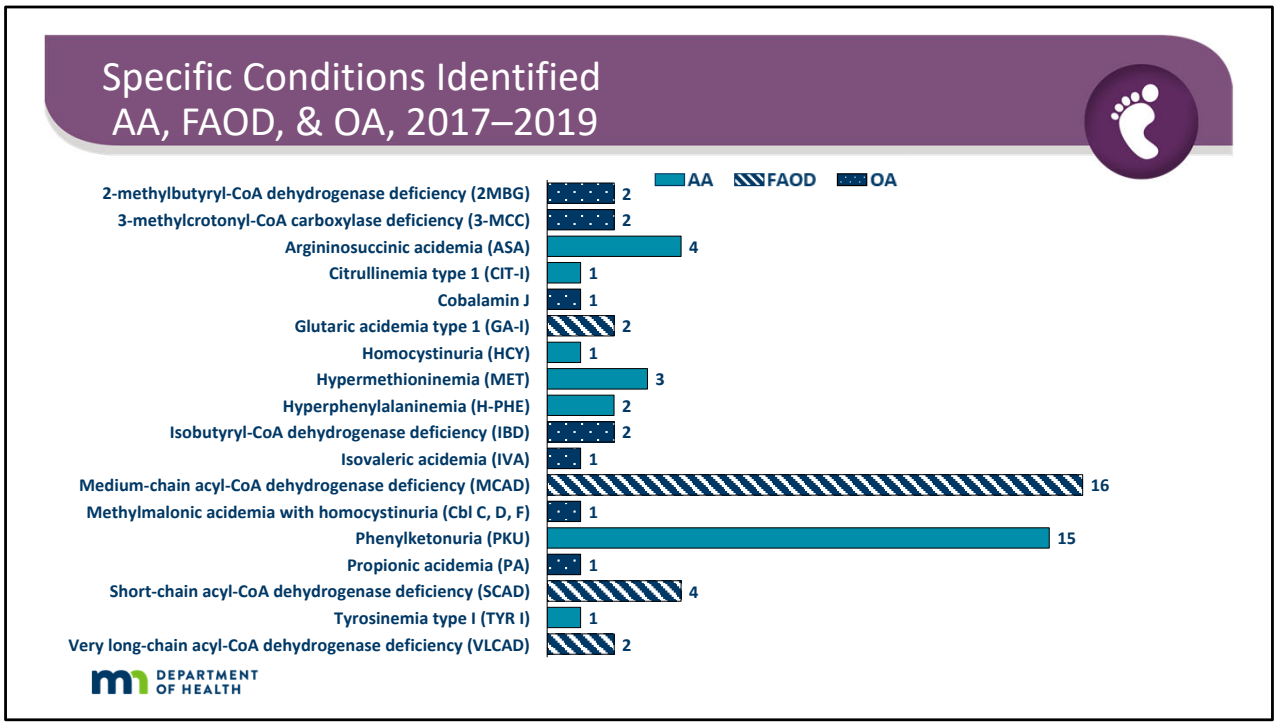
In 2019, three newborns had a screening result indicating a risk for biotinidase deficiency. Of these, two (67%) were confirmed by further evaluation to have this disorder.



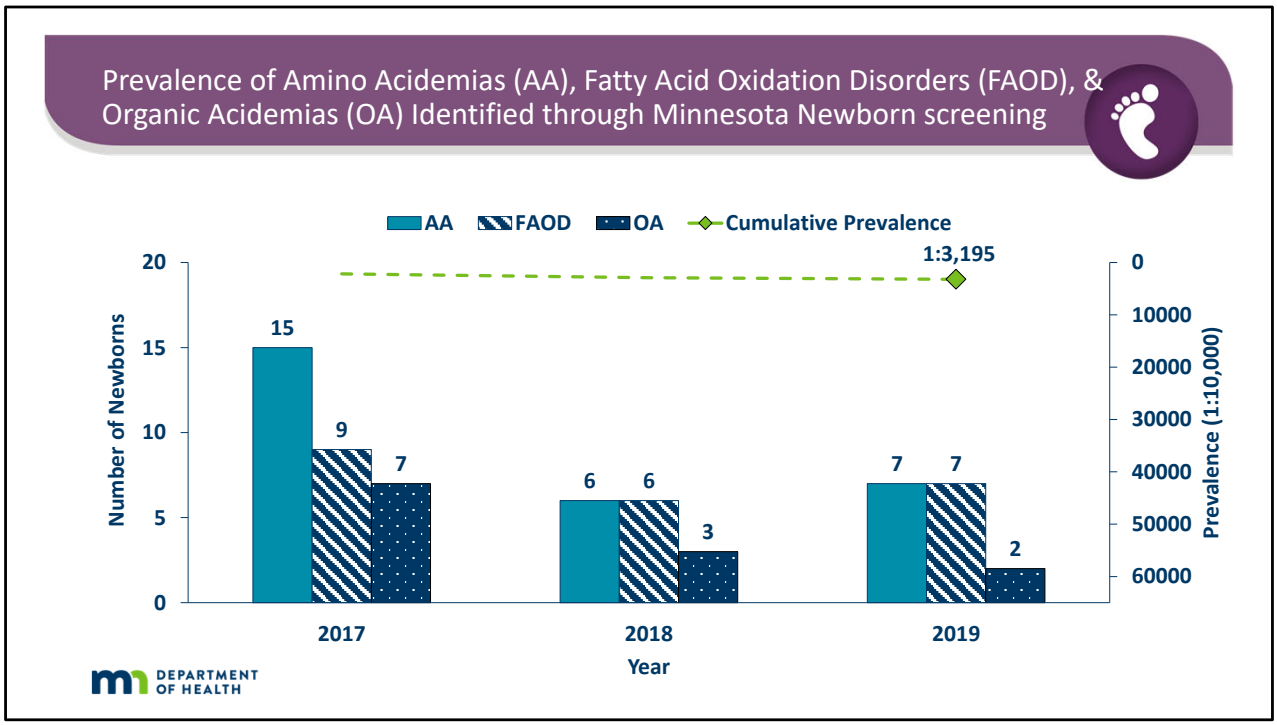
In 2019, 26 newborns had a screening result indicating a risk for galactosemia. Of these, 13 (50%) were confirmed to have galactosemia; a majority (11; 85%) with the mild Duarte type and two (15%) with classic galactosemia.



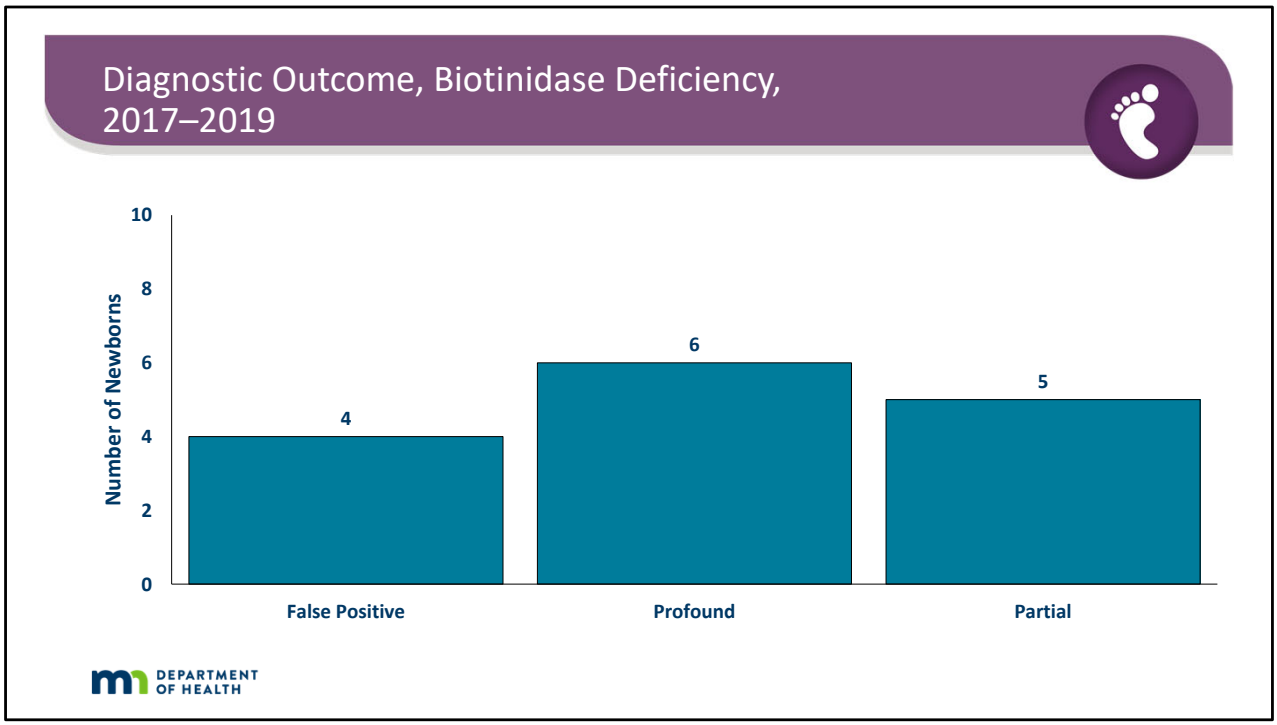
Between 2017 and 2019, 28 (62%) infants with abnormal results for an amino acidemia were found to be affected. Only 34 (47%) infants with an abnormal acylcarnitine result had a confirmed disorder. Combined, over half of the infants with abnormal amino acid or acylcarnitine results were found to have a metabolic disease.



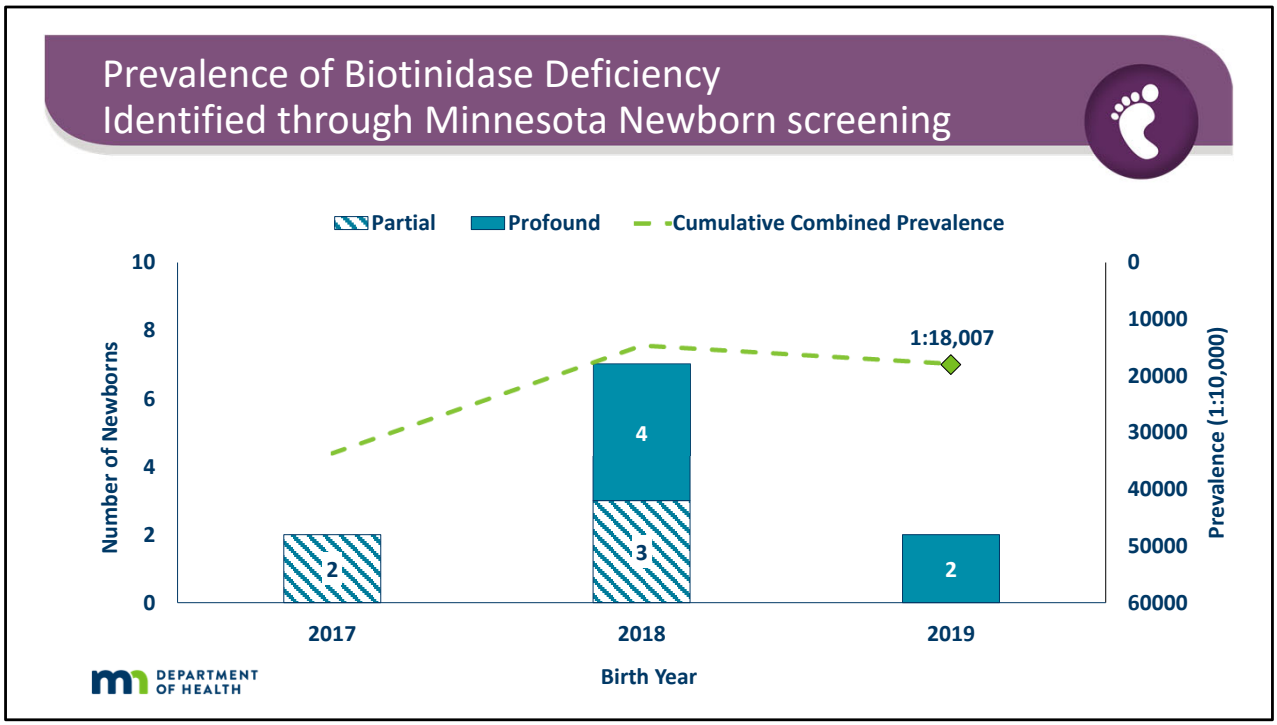
Between 2017 and 2019, the most common diagnoses for infants with an abnormal amino acid or acylcarnitine screening result were medium-chain acyl-CoA dehydrogenase (MCAD) deficiency (16, 26%) and phenylketonuria (PKU; 15, 25%). Sixteen rare conditions make up nearly half (49%) of the other diagnoses.



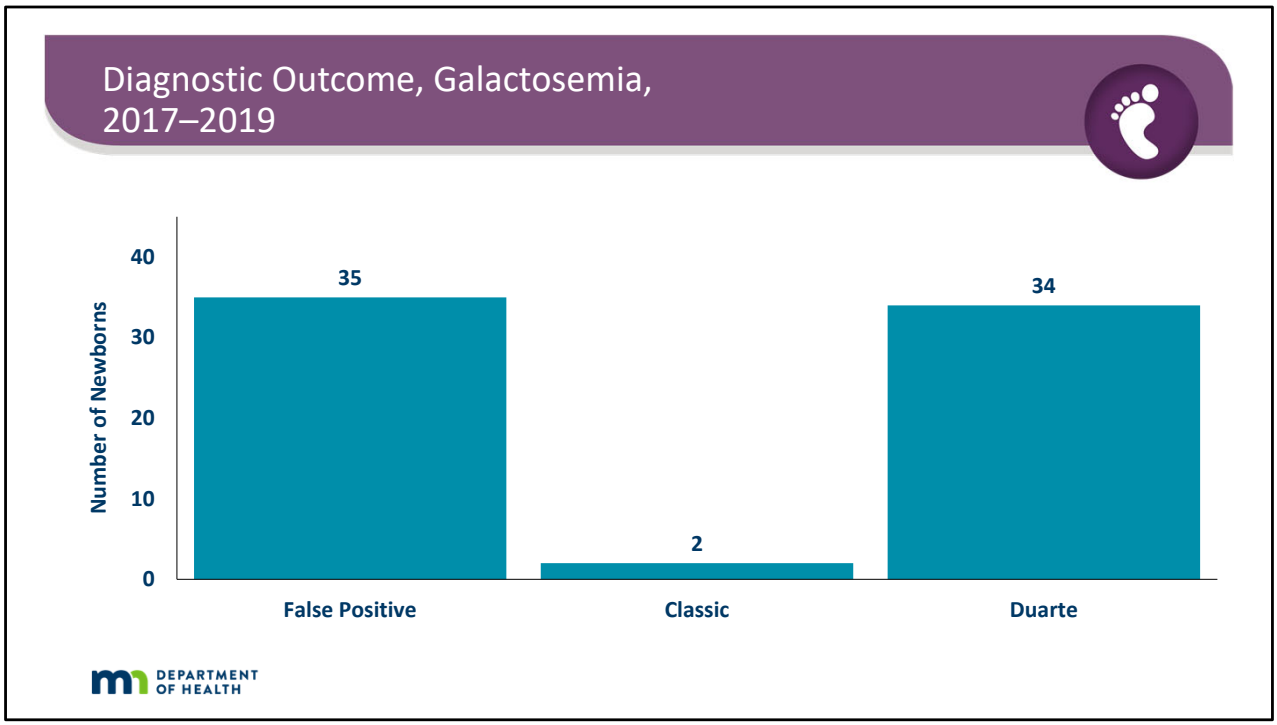
In Minnesota, an average of 21 infants are identified with an amino acid disorder, fatty acid oxidation disorder, or organic acid disorder each year. This means about one in every 3,200 newborns screened is identified with one of these conditions.



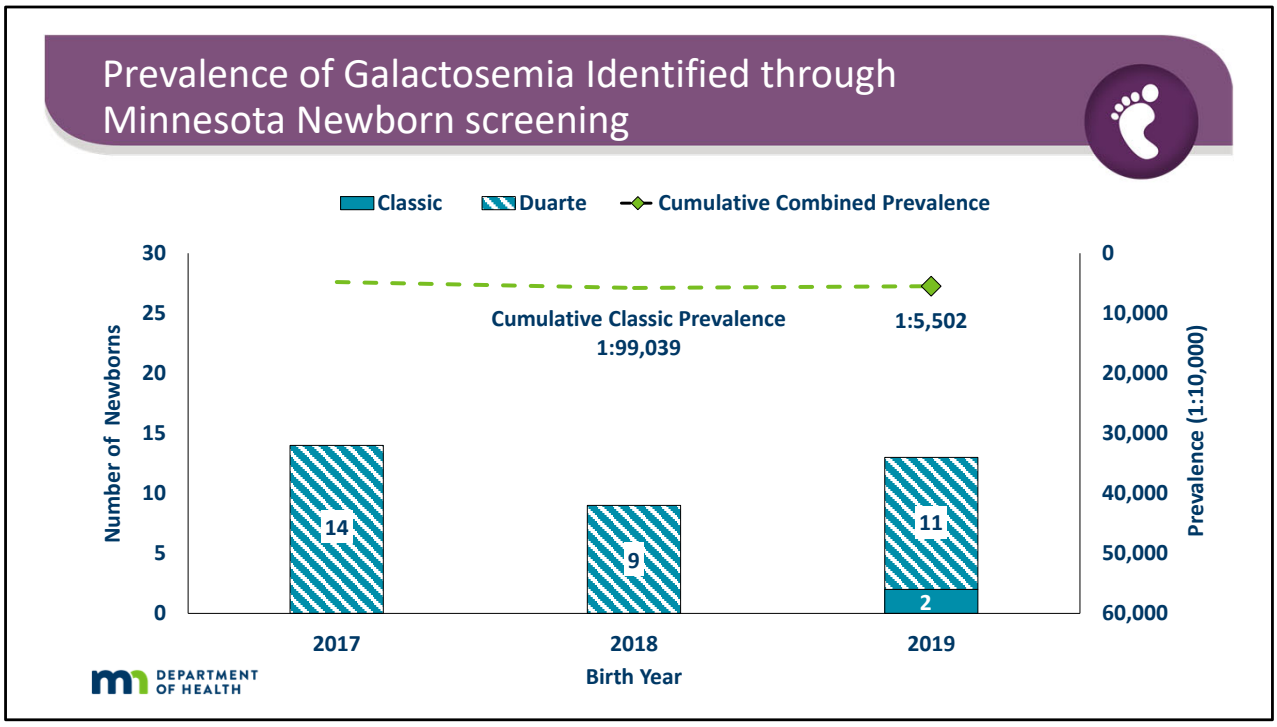
Between 2017 and 2019, six (40%) infants with an abnormal biotinidase deficiency result on their newborn screen had the profound form and another five (33%) had the partial (milder) form. There were four (27%) infants whose results were determined to be false positive; likely due to the impact of heat and humidity on the screening specimen.



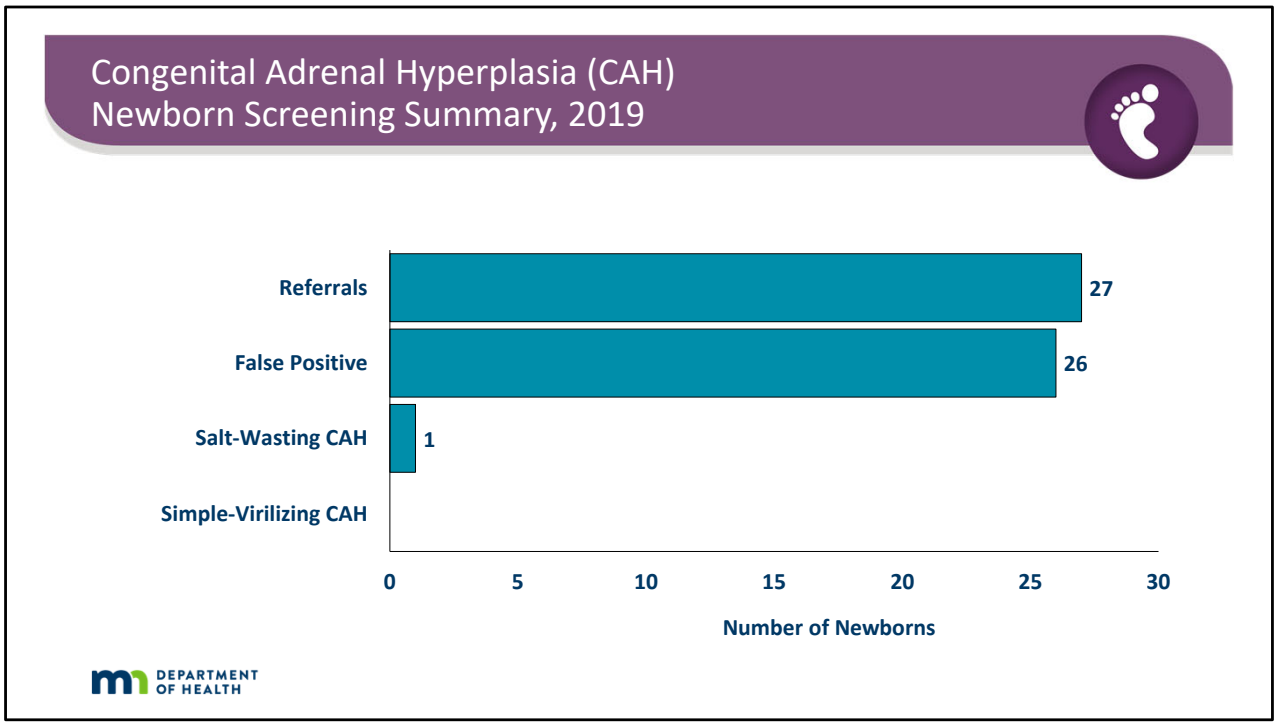
In Minnesota, an average of four infants are identified with biotinidase deficiency each year. This means about one in every 18,000 newborns screened is identified with this condition.



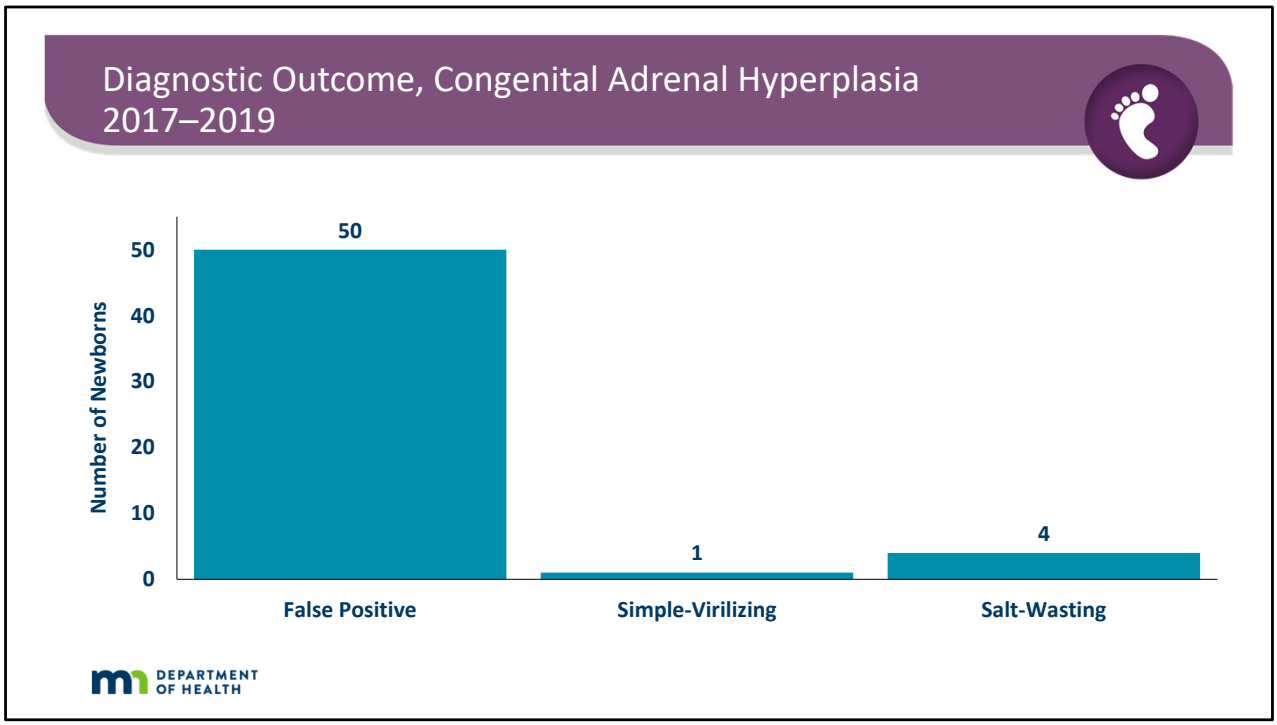
Between 2017 and 2019, the most common diagnosis for infants with an abnormal galactosemia result on their newborn screen was the milder type, Duarte galactosemia (34, 48%). Only two (3%) were diagnosed with classic galactosemia. There were thirty-five (49%) infants whose results were determined to be false positive; likely due to the impact of heat and humidity on the screening specimen. In addition, false positives have been shown to be more common in certain ethnic groups, including the Hmong.



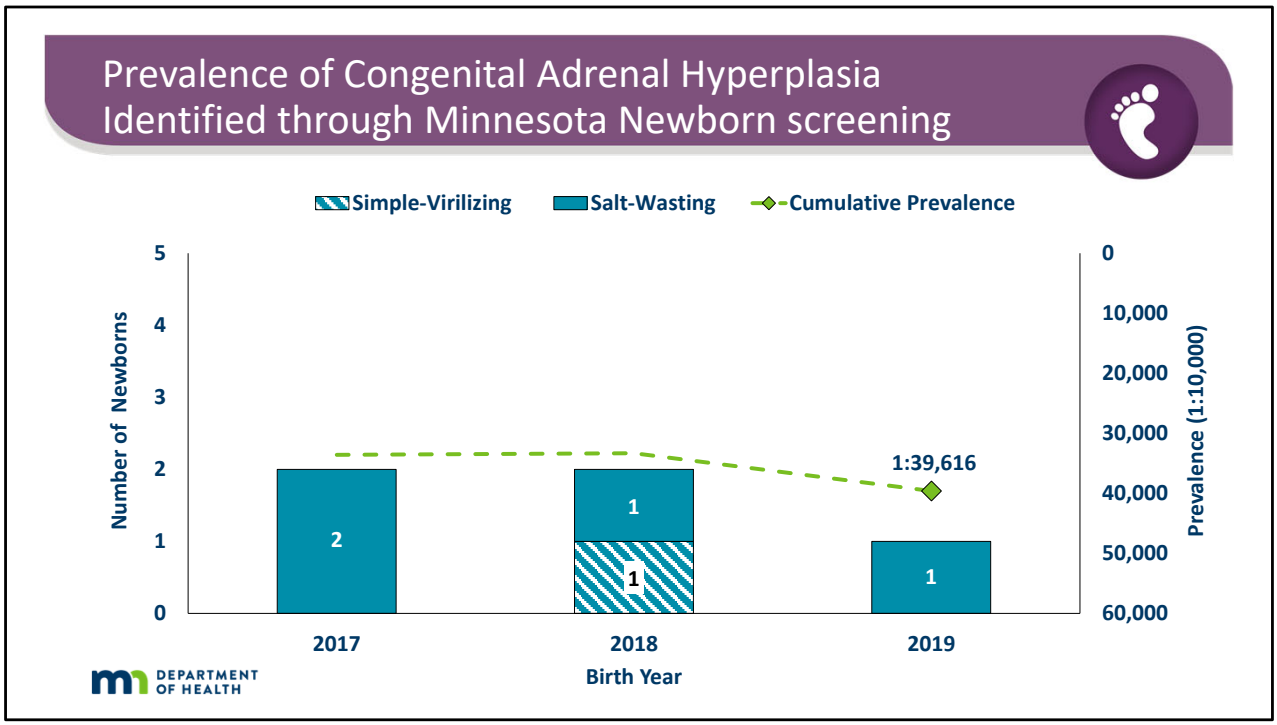
In Minnesota, an average of one infant is identified with classic galactosemia and 11 with Duarte galactosemia (milder type) each year. This means about one in every 5,500 newborns screened is identified with either type of galactosemia, about one in every 99,000 newborns is identified with classic galactosemia.



In 2019, 27 newborns had a screening result indicating a risk for congenital adrenal hyperplasia (CAH). Of these, one (4%) was confirmed by further evaluation to have CAH. There were 26 (96%) infants whose results were determined to be false positive; likely due to early specimen collection, prematurity, or perinatal stress.



Between 2017 and 2019, four (7%) infants with an abnormal congenital adrenal hyperplasia result on their newborn screen had the salt-wasting form and another one (2%) had the simple-virilizing form. Most (50, 91%) infants' abnormal screen results were found to be false positive; likely due to prematurity, collection prior to 24 hours, and perinatal stress.



In Minnesota, an average of two infants is identified with congenital adrenal hyperplasia (CAH) each year. This means about one in every 39,600 newborns screened is identified with CAH.