What's New in Newborn Screening?

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Information on Newborn Screening

Newborn screening in Illinois is mandated and administered by the Illinois Department of Public Health. Screening is ideally performed between 24-48 hours of age. Screening can indicate the possibility of a disorder, which requires follow up with specialists and further testing to confirm a diagnosis. In Illinois, all newborns are screened for amino acid disorders, biotinidase deficiency, congenital adrenal hyperplasia, congenital hypothyroidism, cystic fibrosis, fatty acid oxidation disorders, galactosemia, organic acid disorders, severe combined immune deficiency, sickle cell disease, urea cycle disorders, critical congenital heart disease and hearing loss. Additions to the Illinois Department of Public Health Newborn Metabolic Screening Act expanded newborn screening to include certain lysosomal storage disorders. Pilot screening for five lysosomal storage disorders began on November 3, 2014. Due to provisions in the Affordable Health Care Act, health insurance must cover screening for all conditions listed in the Department of Health and Human Services' Recommended Uniform Screening Panel (RUSP).

Lysosomal Storage Disorders are metabolic disorders that can be inherited in either an autosomal recessive or X-linked recessive pattern. They are characterized by abnormal accumulation of substances inside the lysosome. There are different treatment options for these lysosomal storage disorders, and these should be discussed with a medical specialist. If screening results point to an abnormality, referral should be made to a metabolic disease specialist. This referral needs to be made immediately to prevent fatality or permanent damage. If a diagnosis is made, referral to a genetic counselor is recommended, so that parents can gain a better understanding of how that disorder is inherited.

Which Lysosomal Storage Disorders have been added to newborn screening in Illinois?

FABRY DISEASE

- *Newborn screening measures the activity of alpha-galactosidase.*
- *If not identified early during newborn screening, signs can include pain in extremities, decreased sweating and gastrointestinal issues.*
- *Symptoms typically present in childhood or adolescence, but they can occur any time from infancy through adulthood.*
- *Treatment includes pain management, medications to treat GI symptoms and enzyme replacement therapy.*
- *Treatment is not usually necessary in infancy or early childhood.*
- *Early detection may help to prevent serious, related health outcomes such as stroke, renal damage, cardiomyopathy and permanent tissue damage.*

GAUCHER DISEASE

- *Newborn screening measures the activity of ß-glucosidase.*
- *If not identified early during newborn screening, signs and symptoms can include bone disease, anemia, swelling of the liver and spleen, seizures and neurological symptoms.*
- *This disease has three forms, Type II being the most severe with a limited life expectancy of two years or less.*
- *Symptoms vary by form, and the onset of symptoms is variable.*
- *Symptomatic treatment is provided by a team of specialists, with enzyme replacement therapy available to treat Type I Gaucher disease.*
- *Early detection is important as it can prevent permanent damage to the spleen, liver, bone marrow and—in some cases—to the brain.*

KRABBE DISEASE

- *Newborn screening measures the activity of galactocerebrosidase.*
- *This disease has three forms: infantile, juvenile and adult onset.*
- *If not identified early during newborn screening, symptoms in infantile Krabbe disease begin early in infancy and can include seizures, deafness, blindness, muscle weakness and irritability.*
- *Treatment may include a stem cell transplant early in infancy prior to onset of symptoms.*
- *If not treated early on, this disease can be fatal before age two.*

MUCOPOLYSACCHARIDOSIS TYPE I (MPS I)

- *Newborn screening measures the activity of alpha-L-iduronidase.*
- *This is a multi-system disease with symptoms including intellectual deficits and developmental delays, stiff joints, enlarged liver and spleen, skeletal problems, heart and lung disease, hydrocephalus, hearing loss and corneal clouding.*
- *This disease has three forms: Hurler, Scheie and Hurler-Scheie Syndromes, with Hurler Syndrome being the most severe.*
- *Onset can occur within infancy or later in childhood, depending on the type.*
- *Treatment is coordinated by multiple specialists, and can include enzyme replacement therapy, bone marrow transplants for severe cases and developmental, occupational and physical therapy.*

MUCOPOLYSACCHARIDOSIS TYPE II (MPS II)

- *Newborn screening measures the activity of iduronate-2-sulfatase.*
- *MPS II is also known as Hunter Syndrome.*
- *This disease has two forms: Type A (severe form) and Type B.*
- *Type A is usually diagnosed in early childhood, while Type B may have a later onset.*
- *This is a progressive, multi-system disease with a range of symptoms and signs depending on the type: from joint stiffness to skeletal deformities, from retinal dysfunction to retinal degeneration and from hearing impairment to hearing loss.*
- *Signs and symptoms can also include GI symptoms, skin lesions, cardiovascular issues, coarse facial features, recurrent ear infections, enlargement of the spleen and liver and cognitive impairment.*
- *Treatment is coordinated by a team of specialists, and can include enzyme replacement therapy as well as developmental, occupational and physical therapy.*
- *The test for MPS II is still under development, and screening for this disorder will be implemented at a later date.*

NIEMANN-PICK DISEASE

- *Newborn screening measures the amount of the enzyme acid sphingomyelinase (ASM).*
- *This disease has four forms, with Type A and Type B detected through newborn screening.*
- *Type A is the most severe form, with an early onset (three-six months of age) and fatality between 18 months and four years of age.*
- *Type A symptoms can include: cherry-red macula, an enlarged liver and spleen, brain damage, progressive weakness and loss of early motor skills.*
- *There is currently no effective treatment for Type A.*
- *Type B has an onset of late childhood/adolescence with some survival rates extending into adulthood.*
- *Type B symptoms can include: recurrent respiratory infections, progressive pulmonary disease and an enlarged liver and spleen.*
- *Treatment may require several specialists offering symptom amelioration.*
- *In the future, bone marrow transplant and enzyme replacement therapy may be recommended to treat Type B.*

POMPE DISEASE

- *Newborn screening measures alpha-glucosidase.*
- *This disease has two types: the infantile type has a typical onset in early infancy, and the juvenile/adult type ranges in onset from early childhood to late adulthood.*
- *If not identified early during newborn screening, infantile type symptoms include feeding and respiratory difficulties, poor weight gain, muscle weakness, recurrent pulmonary infections, hypotonia and hypertrophic cardiomyopathy.*
- *Juvenile/adult type symptoms can include muscular dystrophy and respiratory weakness.*
- *Treatment includes enzyme replacement therapy and physical therapy.*
- *If treatment is delayed in infantile type, death can occur within the first year of life.*

Severe combined immune deficiency (SCID) is a group of rare primary immune deficiencies that are inherited. Newborns with SCID have an absence—or deficiency—of T lymphocytes. There are effective treatments available with early intervention. Medical providers should not administer live vaccines to infants with suspected or diagnosed SCID. Genetic counseling is recommended as all forms are due to specific gene mutations.

- *Newborn screening measures the amount of T-cell receptor excision circles (TREC).*
- *If not identified early during newborn screening, signs and symptoms can include recurrent ear infections, thrush, bronchitis, pneumonia and diarrhea that are difficult to treat, poor nutrition and failure to thrive.*
- *Treatment is coordinated by an immunologist, and should include a bone marrow transplant within the first three months of life. Other treatment modalities include prophylactic antibiotic, antifungal and antiviral medications, immunoglobulin supplementation and enzyme replacement therapy.*
- *Without early diagnosis and treatment, SCID is typically fatal within the first year of life.*

Critical congenital heart disease (CCHD) is a group of heart defects that typically require surgical or catheter intervention within the first year of life. Screening is targeted to detect seven congenital heart defections. Congenital heart defects are the most common birth defect, responsible for about a quarter of infantile deaths. If a heart defect is diagnosed, a genetic counseling referral should be recommended, since 15% of heart defects are related to a genetic cause or syndrome. Genetic causes of heart defects can include Trisomy 21 (Down syndrome), Noonan syndrome, Turner syndrome, Trisomy 18, Trisomy 13, DiGeorge syndrome and others.

- *Pulse oximetry screening for CCHD measures the amount of oxygen in the blood, and is ideally performed between 24–48 hours of age.*
- *If not identified early during newborn screening, symptoms can include cyanosis, difficulties breathing, poor weight gain and tiring easily when feeding.*
- *Treatment is coordinated by a cardiologist and includes medications, catheter intervention and surgery.*
- *Delay in diagnosis and treatment can cause cardiogenic shock, neurologic injury, developmental delay, disability, or death.*

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