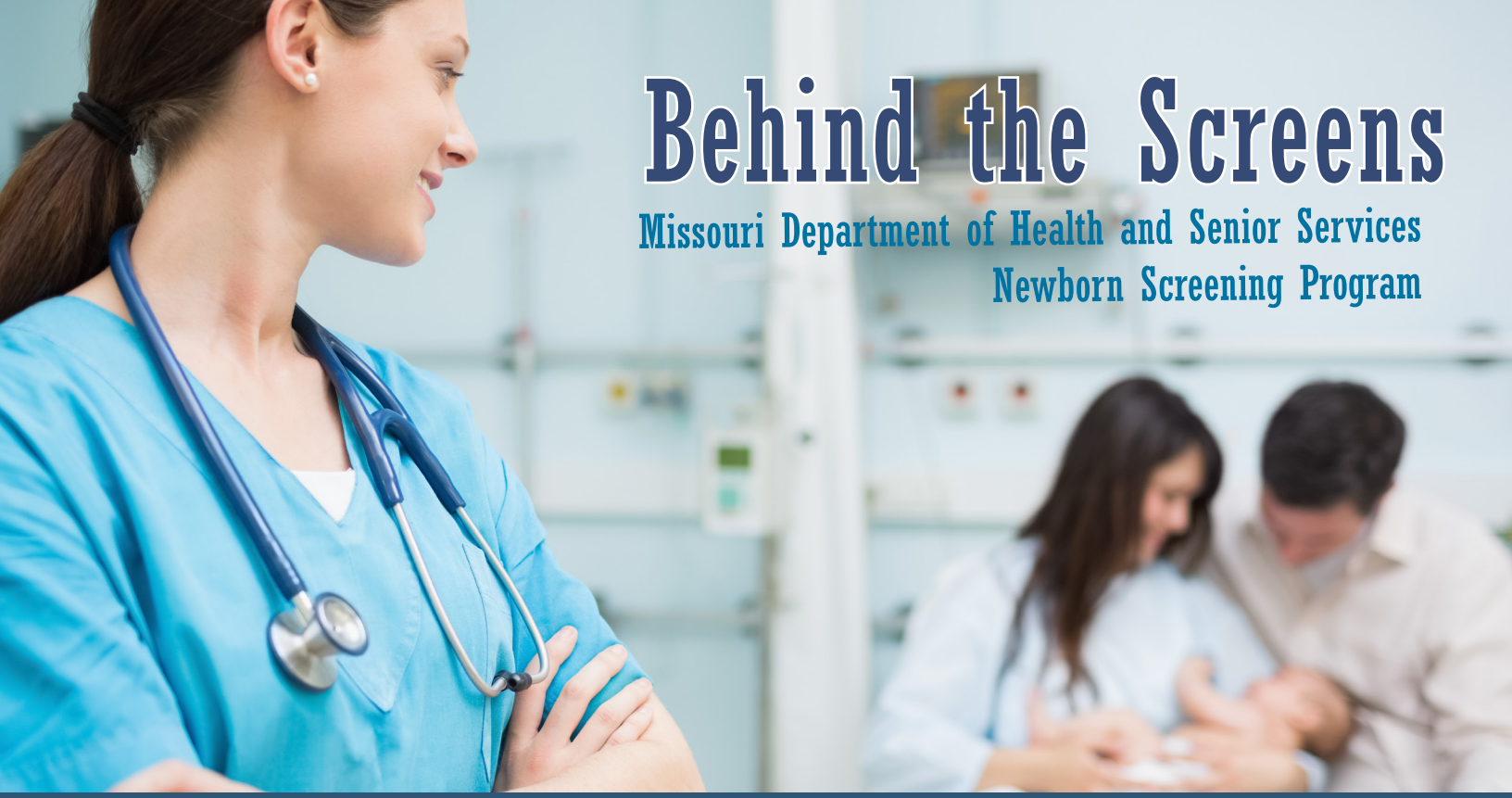

Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Pompe disease, also called glycogen storage disease type II, is a lysosomal storage disorder that affects many different parts of the body. Lysosomes are the recycling centers of the cells and contain an enzyme, acid alpha-glucosidase (GAA), to breakdown and reuse certain materials from the foods we eat. In people with Pompe disease, the GAA enzymes are missing or not working properly. A missing or non-functioning GAA enzyme means trouble breaking down the larger sugar molecule called glycogen into smaller parts. The build-up of glycogen damages muscles throughout the body, most notably the heart and skeletal muscle. This ultimately leads to general muscle weakness, breathing problems, and feeding difficulties.

Pompe disease is an autosomal recessive genetic condition. Two copies of the non-working gene must be inherited, one from each parent, in order to have Pompe disease. Parents of a child with an autosomal recessive condition typically do not show signs and symptoms of the condition because they carry one good functioning copy of the gene.

There are three forms of Pompe disease: classical infantile onset, non-classical infantile onset, and later-onset. The precise GAA variant(s) inherited determine the onset and severity of disease causing symptoms. Classical infantile onset Pompe disease is the most severe type typically appearing in the first two months of life. This type of Pompe disease is known to cause cardiomyopathy. Non-classical infantile onset usually occurs by age one. Symptoms include delayed motor skills, progressive muscle weakness which leads to respiratory failure without cardiomyopathy. Later-onset Pompe disease may develop in childhood, adolescence, or even adulthood. Symptoms are usually milder and progress more slowly than other forms of Pompe disease.

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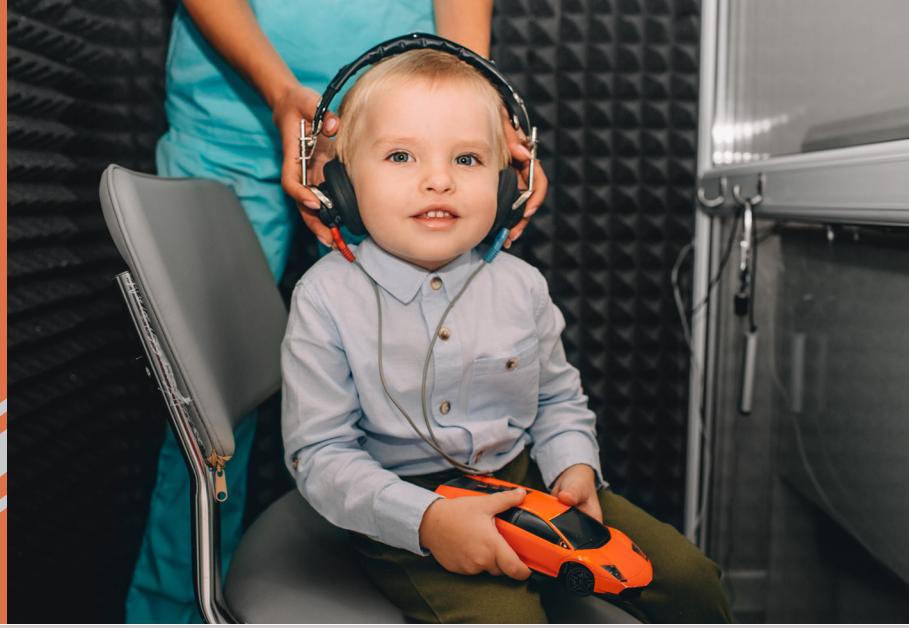
What's New?

Navigate Newborn Screening, a free online newborn screening educational module has been launched by Expecting Health. Navigate Newborn Screening provides free newborn screening information for families. It helps families build the knowledge and skills they need to find more information, communicate with their healthcare providers, and serve as leaders in the newborn screening system. To learn more, visit Navigate Newborn Screening.

Did You KNOW?



Did you know there is a Joint Committee on Infant Hearing (JCIH)? The JCIH is composed of representatives from audiology, otolaryngology, pediatrics, and teachers of children who are deaf or hard-of-hearing (DHH). Widely respected and endorsed among professionals concerned with children who are DHH, the JCIH is charged with making recommendations concerning newborn hearing screening and the early identification of children with, or at-risk for, hearing loss. State Early Hearing Detection and Intervention (EHDI) programs use JCIH recommendations as guidelines for best practices. (Missouri guidelines for hospital hearing screening programs are based on JCIH recommendations and can be found at https://health.mo.gov/living/families/genetics/newbornhearing/guidelines.php.)

Since its establishment in 1969, the JCIH has issued position statements with guidelines for EHDI. Until recently, state EHDI programs looked to the 2007 JCIH position statement for guidance. In October 2019, JCIH released its 2019 position statement: "Principles and Guidelines for Early Hearing Detection and Intervention Programs." The new statement builds upon the 2007 guidelines and emphasizes all infants should be screened using a physiologic measure at no later than one month of age, those infants who did not pass the initial hearing screening and the subsequent rescreening should have an audiologic evaluation by three months of age, and all infants diagnosed with a permanent hearing loss should receive intervention by six months of age. However, there is more. In its executive summary, the authors of the 2019 position statement highlighted the following newborn hearing screening standards as excerpted below:

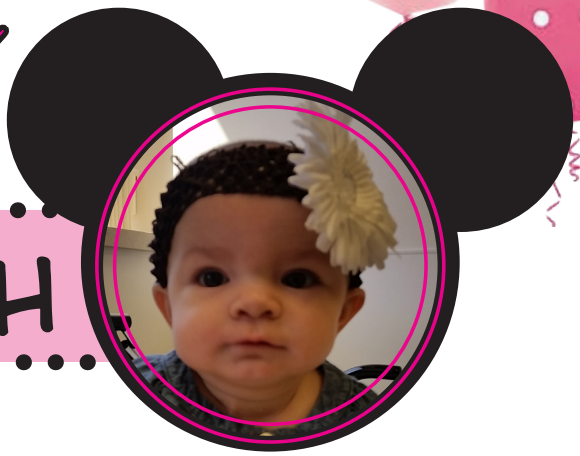
- Endorsement of the necessity for audiology oversight of hearing screening programs.
- Recognition of the critical need for the ability to calibrate screening equipment using a uniform and validated standard across all screening devices.
- Recognition of the need for manufacturers of screening equipment to provide data on the proportion of children who are DHH who pass the screening but are subsequently found to have a variety of degrees and types of hearing loss.
- An endorsement, for well-born infants only, who are screened by automated auditory brainstem response (AABR) and do not pass, that rescreening and passing by otoacoustic emissions (OAE) testing is acceptable, given the very low incidence of auditory neuropathy in this population.
- An endorsement of rescreening in the medical home in some circumstances. If the rescreening is performed in the provider's office, the provider is responsible for reporting results to the state EHDI program.

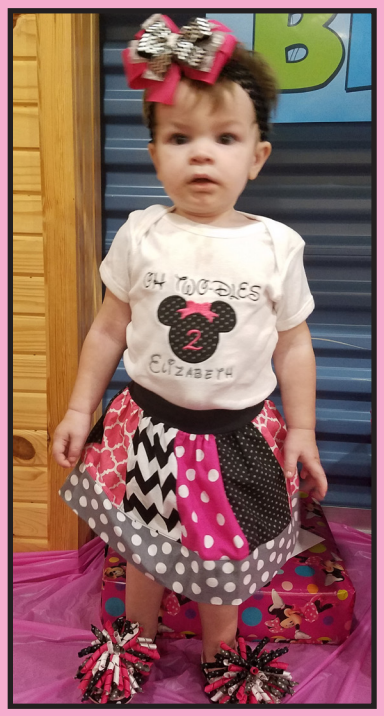
The 2019 position statement also addresses pediatric diagnostic audiology, medical evaluation, early intervention, medical home and ongoing surveillance, rights of infants/toddlers and families, EHDI information technology infrastructure, benchmarks and quality indicators, future directions, and research needs.

The 2019 JCIH position statement can be found in its entirety in the Journal of Early Hearing Detection and Intervention at https://digitalcommons.usu.edu/jehdi/vol4/iss2/1/.

PATIENT Spotlight

ELIZABETH





"Elizabeth was born with a rare genetic disorder called Mucopolysaccharidosis 1 or Hurler Syndrome. We did not know about it when she was born on August 28. We received a call from our pediatrician a couple of weeks after she was born saying we were needing to do a retest. By the time she was just over a month old, we had our diagnosis and were already starting her enzyme replacement therapy. On January 7 she had her bone marrow transplant. Hurlers is a terminal illness. The bone marrow transplant does not cure Hurlers, it only helps treat some of the things that Hurlers causes. Without the transplant, life expectancy is usually around 10 years old. Most Hurler families are not as lucky as we were to have caught it during newborn screening. Most don't find out they have it until they start having problems, usually first noticed with the heart, respiratory issues, or kyphosis usually around 9 to 13 months old. Because of newborn screening, we had a diagnosis and started enzyme therapy replacement within 42 days. She had her transplant at 4 months and 10 days old. This is so important because they typically won't do the transplant after two years old. The early transplant also helps to prevent progression of some of the things Hurlers causes.

Today Elizabeth is a happy, energetic four year old. She is in her second year of preschool and a joy to everyone she is around. To see her, most have no idea of the things she has gone thru in her short life so far nor the things to come. We don't have near the complications from Hurlers that many kids have and I fully credit newborn screening for that."

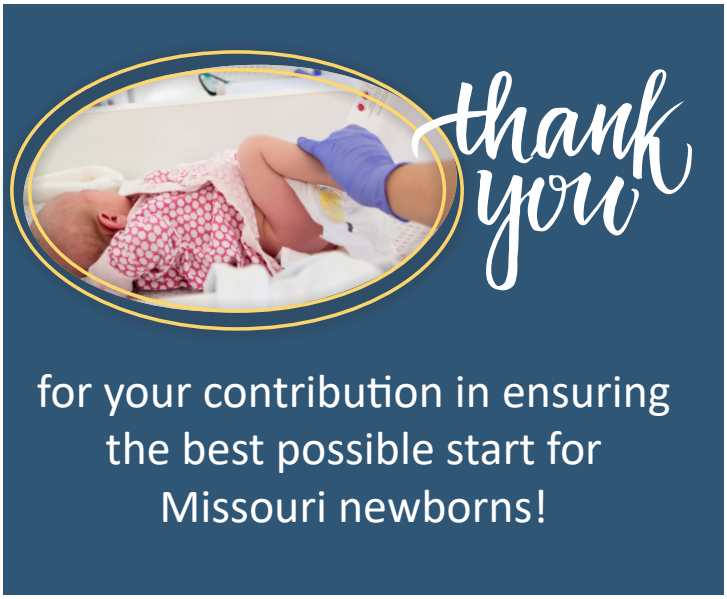
Tina, MOM

> "We don't have near the complications from Hurlers that many kids have and I fully credit newborn screening for that."

Featured Disorder continued from page 1

Multiple specialists make up the care team that will work together to create an effective treatment plan. These specialists include pediatricians, neurologists, orthopedists, cardiologists, and dieticians. Treatment for Pompe disease varies due to type and symptoms present. Targeted enzyme replacement therapy (ERT) is one type of treatment that can help slow disease progression. Other treatment options include supportive therapies such as respiratory support; physical, occupational, and speech therapy; orthopedic devices including braces; and the use of canes or walkers. Dietary modifications may also be recommended. Some affected individuals may need a high-calorie diet consisting of soft foods due to muscle weakness affecting the ability to chew and swallow.

Newborn screening for Pompe is imperative because without treatment many children do not reach their second birthday. When Pompe is detected early and treatment is started promptly, many are able to live longer lives with improved growth, development, and organ function. Missouri began screening for Pompe disease in 2013. Visit https://rarediseases.org/rare-diseases/pompe-disease/ to learn more.



Tech Tips

- Hearing rescreening results may be entered in the Missouri Electronic Vital Records (MoEVR) system's "Rescreen Hearing" page. Please do not enter initial hearing screening results in the "Rescreen Hearing" page. The "Rescreen Hearing" page may be left blank if your hospital does not perform outpatient hearing rescreening. Direct questions about the MoEVR "Rescreen Hearing" page to Linda at (573) 751-6266 or (800) 877-6246.
- Remember to fill in gestation age and birth weight on the screening form for accurate reporting. If both are blank, no results will be given for several tests. This will require a repeat newborn screen. It is important to recollect the newborn screen as soon as possible so there is no delay in follow-up for babies that may have an abnormal result.
- All specimens received at the newborn screening laboratory are examined for specimen acceptability. Poor quality specimens may not have enough blood to perform all the testing, may have been collected improperly, or may have been delayed in the mail. For satisfactory specimens, allow a sufficient amount of blood to soak through, completely filling the preprinted circle on the filter paper. Fill all required circles and do not layer successive drops of blood or apply blood more than once in the same collection circle. Examples of satisfactory and unsatisfactory blood spot specimens can be found by visiting https://health.mo.gov/lab/newborn/pdf/poor_quality_specimens.pdf.

MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

Missouri DHSS Department of Health and Senior Services

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