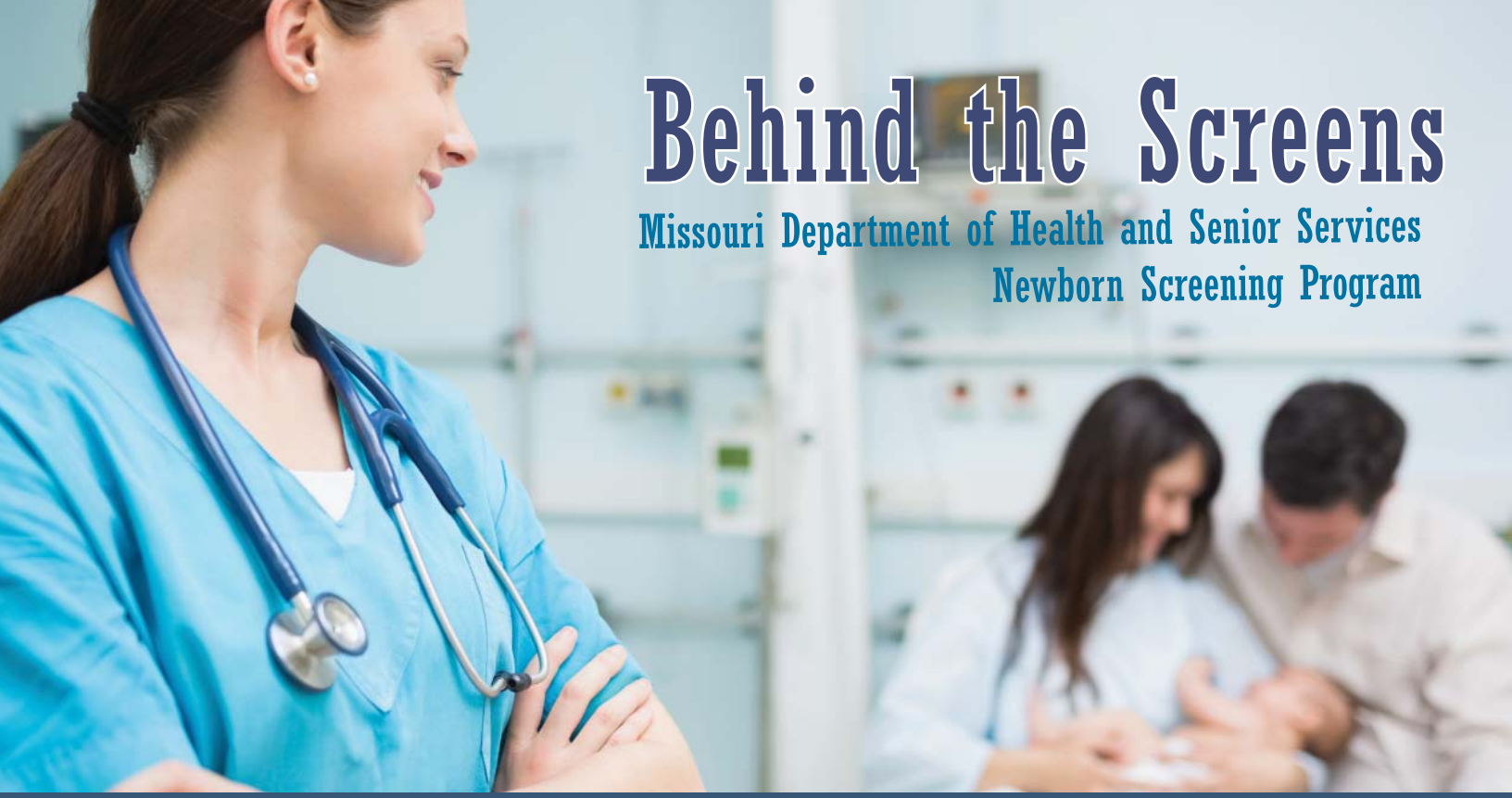

Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Fabry is an X-linked lysosomal storage disorder. Individuals affected by Fabry are not able to break down specific fat molecules at the cellular level. This results in a buildup of fat molecules most often affecting the cells lining the blood vessels in the skin and cells in the kidneys, heart, brain, and nervous system.

The incidence of classical Fabry is approximately 1 in 40,000 to 60,000 males and an unknown number of females. Less severe, late-onset forms of Fabry are more common. This disorder is found in all ethnic, racial and demographic groups. Males with Fabry typically have more severe symptoms than females.

Children with classical Fabry typically begin to show signs of the disorder between the ages of four to six years. Early signs of classical Fabry include episodes of neuropathic pain, ophthalmological problems, gastrointestinal problems, auditory problems, altered temperature sensitivity, angiokeratomas, kidney failure of unknown cause, and heart murmur.

While there is no cure for Fabry, treatments are available to help reduce signs and symptoms and prevent future complications. Treatments may include special dietary restrictions, enzyme replacement therapy, pain control, ACE inhibitors, and aspirin. Children who are treated early for Fabry can have healthy growth and development, however, some children may still experience some signs and symptoms such as pain or hearing loss. Without early treatment, individuals with Fabry are at risk for heart attacks, stroke, or kidney disease.

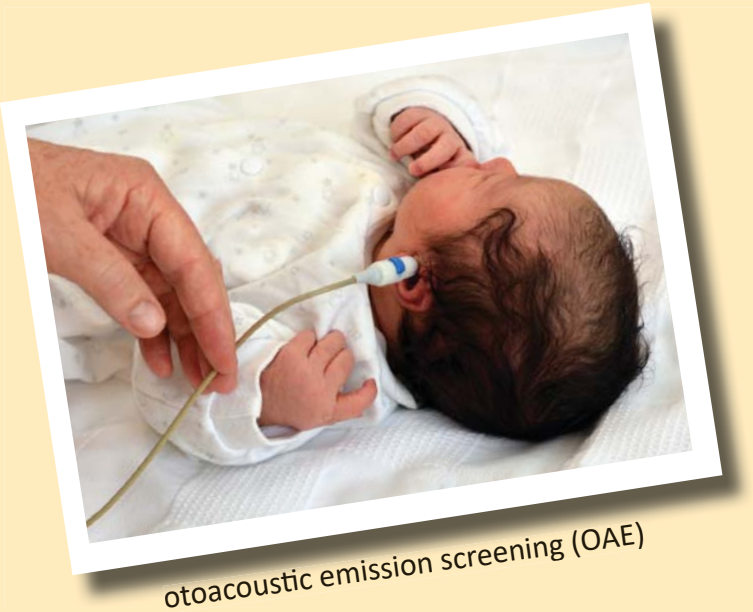
Fabry was added to Missouri's newborn screening panel on January 2, 2013. As a result of newborn screening and genetic counseling, at-risk families can be identified and evaluated for Fabry. Early detection through newborn screening allows for careful monitoring and the initiation of treatment as soon as clinically appropriate.

What's New?

The Department of Health and Senior Services (DHSS) literature warehouse has multiple publications to assist you in providing excellent education to the families in your care. All DHSS publications are free to all Missourians including health care providers! Materials produced by the Bureau of Genetics and Healthy Childhood may be ordered by using the warehouse online order form at https://health.mo.gov/living/families/genetics/newbornhealth/orderform.php. Visit these links to see two of our newest items:

Newborn Screening Infographic https://health.mo.gov/living/families/genetics/pdf/literature/436.pdf

Cytomegalovirus (CMV) Infection http://health.mo.gov/living/families/genetics/pdf/literature/228.pdf



otoacoustic emission screening (OAE)

automated auditory brainstem response (AABR)

Did You KNOW?

Did you know that there are two different ways to screen a baby's hearing: otoacoustic emission screening (OAE) and automated auditory brainstem response (AABR)? While the methods are different from each other, both do an excellent job of finding babies who may have serious hearing loss.

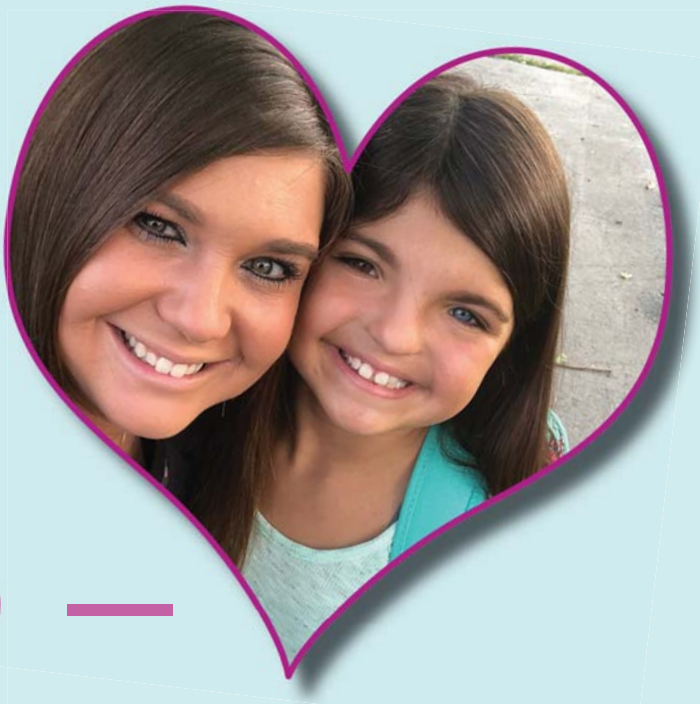
An otoacoustic emission is an event that happens in everybody's ear. It is a by-product of the normal hearing process in which a tiny sound is physically generated by an ear that is working to hear. It can be compared to a squeak of a closing door. The closing of the door is the important action that is taking place. The squeak is just something that happens during the work. The squeak, or otoacoustic emission, is only present in ears that have normal hearing. It can be measured as movement of the eardrum by a tiny probe placed in the baby's ear canal.

AABR measures hearing in a completely different way. Electrodes are placed on the baby's skin. A probe is placed in the ear canal. In this case, the probe is only there to deliver a very specific sound to the baby's ear. Since hearing, like all senses, ends up as electrical energy that the brain can interpret, the baby's brain's response to the sound can be measured from the electrodes. Ensuring that the baby's brain reacts in a normal pattern to soft sounds also ensures good hearing!

Tech Tips

- If you need to change a hearing screening result previously entered into MoEVR, send the revised result to DHSS on the Hearing Screening Only Result Form (https://health.mo.gov/living/families/genetics/newbornhearing/guidelines.php). Changes made in MoEVR do not transfer to the DHSS Newborn Hearing Screening Program.
- Screen babies for hearing loss after they are 24 hours old to ensure that debris has cleared from the ear canal.

PATIENT SPOTLIGHT



Meet — Kynlee —

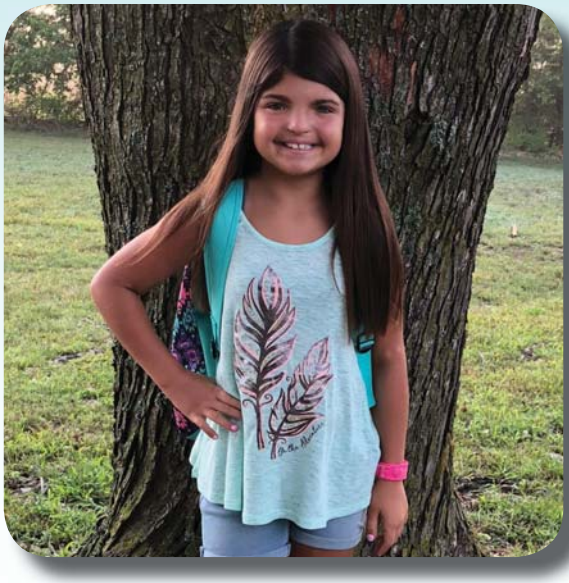
"Early on the morning of November 6, 2009, I headed to the hospital for my scheduled induction. I had a normal pregnancy, with the exception of kidney stones, and I couldn't be more excited to meet my baby girl! Little did I know how much my life was about to change! When they laid my baby girl on the scale across from me, I looked at her in awe, but my "Mommy gut" told me that something wasn't right. Shortly after, the pediatrician came to my room to examine Kynlee. It was then that my anxiety heightened, as she confirmed my "Mommy gut" feelings were right. She explained that Kynlee had characteristics of Waardenburg Syndrome, a rare genetic disorder that affects approximately 1 in every 40,000 people, with the most concerns being hearing loss/deafness. Kynlee's white forelock and wide spaced eyes were 2 of the visible characteristics that the pediatrician quickly picked up on. She stressed the importance of her newborn hearing screening and explained if she didn't pass, follow up testing would be very important. When Kynlee didn't pass her newborn hearing screening, my worries continued to increase. The hospital set up an appointment for a hearing rescreen 10 days after we were discharged. I remember that being the longest 10 days of my life. I researched everything I could about Waardenburg Syndrome, hearing loss in children, and I cried a lot. When it was time for Kynlee's hearing rescreen appointment, I had accepted the fact that there was a good chance my daughter had hearing loss. During her rescreen, she passed the testing on her right ear, but still failed on her left. At that time, Kynlee was set up for diagnostic testing to help me better understand what was going on with her hearing. Once the diagnostic testing was done, it was determined that Kynlee had normal hearing in her right ear, and profound loss in her left. While this wasn't the diagnosis any parent would want for their child, I was relieved that she had at least some hearing. We were set up with an amazing Audiologist, who would become a good friend of ours in the years to come. She helped get Kynlee her first hearing aid well before she was 6 months old, which set her up for the best possible outcome. I was also put in touch with First Steps, who helped to make sure Kynlee stayed on track with her speech and taught our family some sign language to help with Kynlee's communication. At 3 ½ years, it was found that Kynlee's hearing loss had progressed to her right ear and she would need a hearing aid on that side as well. I had hoped this was something she wouldn't have to deal with but I found comfort in knowing that we caught it quickly and could intervene.

"Who knew something as simple as a newborn hearing screening could start the process to help change the outcome of my child's life?"

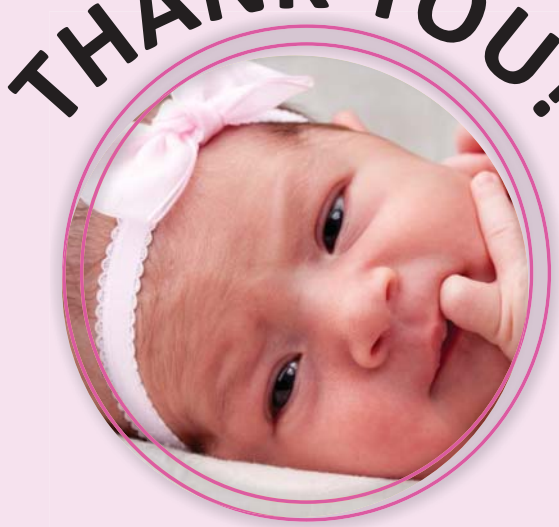
Continued on page 4

Kynlee continued from page 3

Kynlee is now 9 years old, a straight A student, and has normal speech development all thanks to what started with a test many of us take for granted! Who knew something as simple as a newborn hearing screening could start the process to help change the outcome of my child's life? Kynlee's hearing loss being found during her newborn hearing screening also sparked an interest in me for a career change. This is one of the main reasons I am now the full-time hearing screener for the same hospital that helped diagnose Kynlee. It is such a blessing to get to work in an area that I have both professional and personal experience with. I love babies and have a passion for infant hearing. This links both together. I'm able to be there for parents who might find themselves in the same situation I was and offer comfort and advice to help them through this challenging time in their lives."



"I am forever grateful to the Newborn Hearing Screening Program!"



Thank you for your contribution to ensuring the best possible start for Missouri newborns!

MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

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