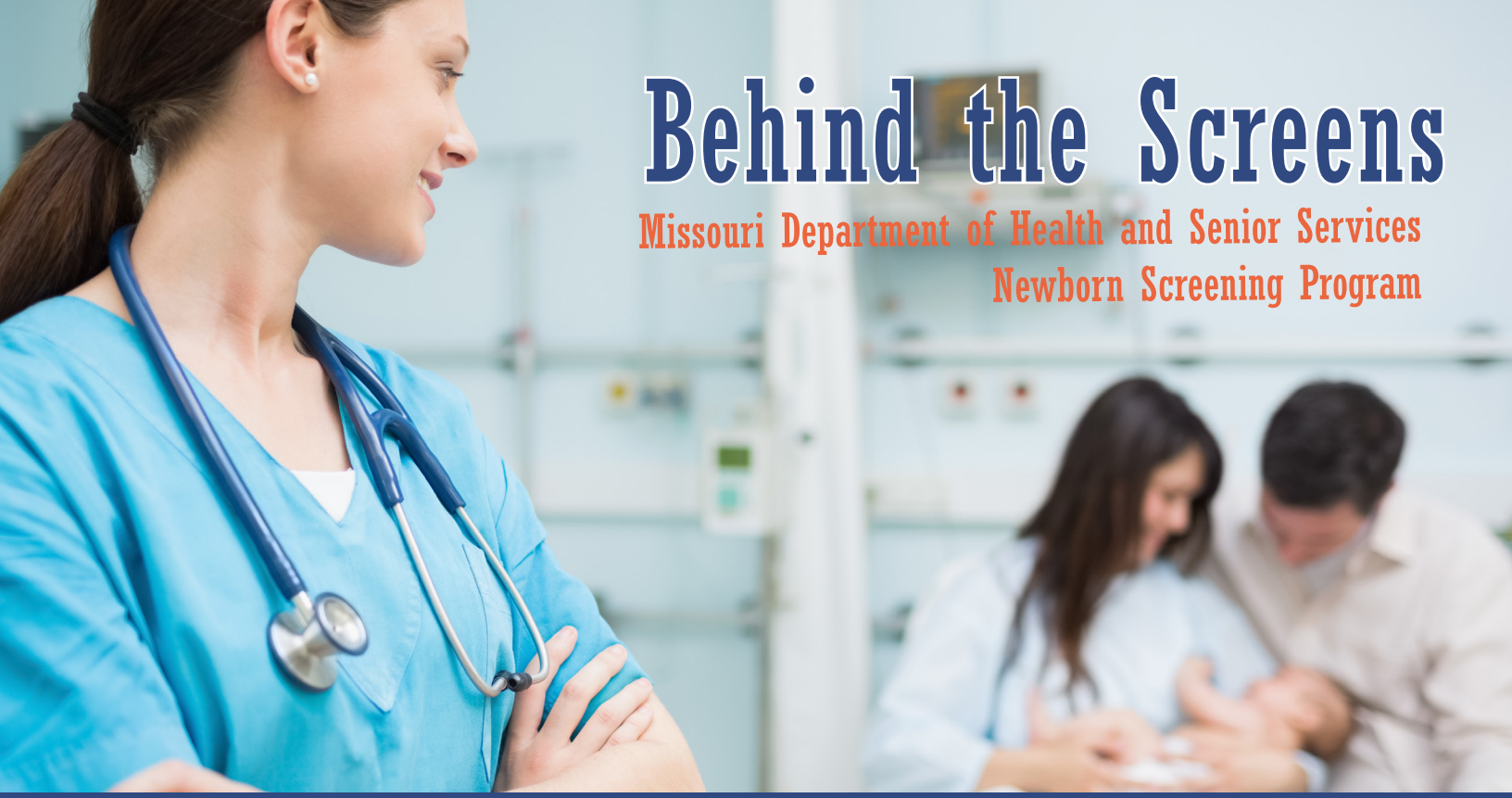

Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

Winter 2023 ★ Missouri Department of Health and Senior Services ★ Health.Mo.Gov ★ Volume 6 Number 1

Featured Disorder

Biotinidase deficiency (BIOT) is an inherited condition in which the body is not able to reuse or recycle the vitamin biotin. The body needs free biotin to help break down certain fats, proteins, and carbohydrates. Because of this, the body is unable to process vital nutrients. Early detection through newborn screening and immediate treatment gives babies the best chance for healthy development. BIOT occurs in about 1 of every 60,000 births. There are two main types, which differ in severity and signs; mild "partial biotinidase deficiency" and severe "profound biotinidase deficiency". Signs and symptoms can present within the first few months after birth. Those include seizures, weak muscle tone, skin and fungal infections, and trouble breathing. Many symptoms can be aggravated by other infections or illnesses. Treatment includes lifelong biotin supplements. Although biotin is a natural vitamin found in many foods, children with BIOT do not get enough and their bodies are not able to use it properly. Treatment cannot reverse damage already done, which is why early detection and treatment are key.

Tech Tips

Hearing rescreening should be done on both ears in the same session, regardless of the initial screening results. This is to ensure that fluctuation or progression in hearing levels are not missed.

For more information, please visit The Journal of Early Hearing Detection and Intervention.

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Patient Spotlight
Colin & Brennan

In 2014 my husband and I welcomed Brennan into the world knowing we would need to monitor his vision due to the possibility that he may inherit my eye disease, retinitis pigmentosa. However, when he failed his newborn hearing screening we were completely caught off guard. The hospital referred us to an audiologist who diagnosed Brennan with unilateral sensorineural hearing loss. Although we knew he would be fine, we also felt sad and confused. His first hearing aid had a tiny screw on the hearing aid door that had to be locked with a small screw driver in order to activate the hearing aid. I was not always able to lock the door. I felt insecure; not because I was a blind parent but because I could not lock my son's hearing aid so he could use it. Fortunately, MOHear and First Steps came into our lives. When our MOHear audiologist called me, I explained our problems and she assured me she would help. She said that there were several hearing aid options and that we had the right to request a different hearing aid model and a different audiologist. I had been so upset that it had not occurred to me that there were different and better options for our family.

Before our son Colin was born in 2015, we met with a genetic counselor who said there was little chance of vision or hearing loss due to no family history beyond Brennan and me. Once again, we were surprised. Colin failed the newborn hearing screening and was diagnosed with bilateral sensorineural hearing loss.

Today, Brennan wears a hearing aid and Colin loves his cochlear implant! The boys enjoy playing baseball, performing, participating in scouts and taking vocal lessons. They make us proud to be their parents! If it had not been for the newborn hearing screenings, Brennan and Colin's hearing losses would have gone unnoticed until they were older.

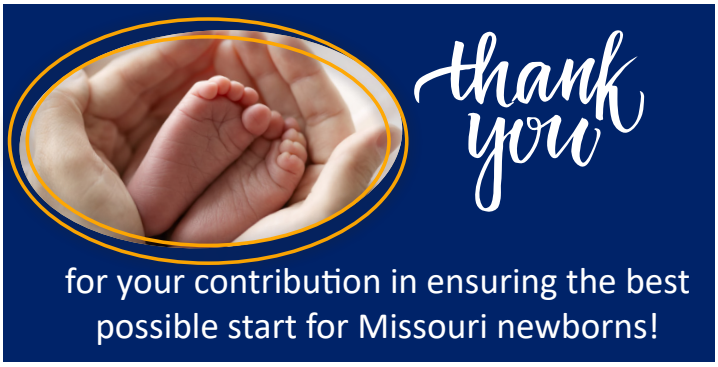
Megan,
Brennan and Colin's mom

Did You KNOW?

The Missouri Department of Health and Senior Services Newborn Screening (NBS) Program launched a full population pilot in December 2021 to screen all newborns for X-linked adrenoleukodystrophy (X-ALD). While screening results for X-ALD did not appear on the routine NBS laboratory reports during the pilot/implementation period, all newborns were tested for this disorder, and any displaying high-risk abnormal results were referred to a specialist for confirmatory testing. Effective December 1, 2022, the Missouri State Public Health Laboratory went live with the routine reporting of X-ALD on the standard NBS laboratory reports.

For more information on topics in this edition, please visit:

- babysfirsttest.org
- health.mo.gov/lab/newborn
- digitalcommons.usu.edu/cgi/viewcontent.cgi?article=1104&context=jehdi



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Missouri State Newborn Screening Laboratory
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Health.Mo.Gov/newbornscreening

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Hearing- and speech-impaired citizens can dial 711.