Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

October 2020 ★ Missouri Dept. of Health and Senior Services ★ www.health.mo.gov ★ Volume 4 Number 3

Featured Disorder

Congenital adrenal hyperplasia (CAH) is a collection of inherited conditions that affect the adrenal glands. In a person with CAH, the adrenal glands are very large and are unable to produce certain hormones the body needs to function. Left untreated, CAH causes an imbalance in these hormones in one of three ways. The first way is, the adrenal glands make too little cortisol. Cortisol affects energy levels, blood sugar levels, blood pressure, and the body's response to stress, illness, and injury. The second way is, the adrenal glands produce too little aldosterone. Aldosterone is needed to help the body maintain the proper balance of sodium and water to maintain blood pressure. The third way is, the adrenal glands produce too much androgens, or male hormones. Adequate levels of these hormones are needed for normal growth and development in both males and females. Early detection and treatment can help children with CAH to have normal and healthy development.

CAH is an autosomal recessive disorder caused by mutations in one of several genes. For an infant to have CAH, both copies received, one from each parent, must have an error that affects an adrenal-gland enzyme.

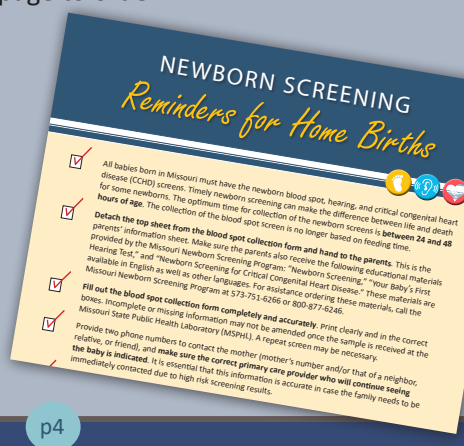
The signs of CAH vary from person to person and the form of CAH defines when symptom manifestation is likely to occur. There are two main forms of CAH: classic and non-classic CAH. Classic CAH is divided further into two categories: salt-wasting and simple virilizing or non-salt wasting. Classic is the more severe form. Non-classic is the less severe form and less likely to manifest in the newborn period.

Salt-wasting CAH accounts for about 75% of classic CAH cases. This is the most severe form. The adrenal glands make too little aldosterone, causing the body to be unable to retain enough sodium. Too much sodium is lost in urine, thus the name, salt-wasting.

Continued on page 2

What's New?

We have been eagerly working on updating some of the educational materials available for patient and staff education. The *Home Birth Reminders* and the *Top Ten Reminders* have been updated. A mailing with these documents will be sent out in the upcoming months! Be sure to check out all the resources available free to you by visiting https://health.mo.gov/living/families/genetics/newbornscreening/publications.php, select Your Publications - Order Form at the bottom of the page to order.


NEWBORN SCREENING
Reminders for Home Births

All babies born in Missouri must have the newborn blood spot, hearing, and critical congenital heart disease (CCHD) screens. Timely newborn screening can make the difference between life and death for some newborns. The optimum time for collection of the newborn screens is **between 24 and 48 hours of age**. The collection of the blood spot screen is no longer based on feeding time.

Detach the top sheet from the blood spot collection form and hand to the parents. This is the parents' information sheet. Make sure the parents also receive the following educational materials provided by the Missouri Newborn Screening Program: "Newborn Screening," "Your Baby's First Hearing Test," and "Newborn Screening for Critical Congenital Heart Disease." These materials are available in English as well as other languages. For assistance ordering these materials, call the Missouri Newborn Screening Program at 573-751-6266 or 800-877-6246.

Fill out the blood spot collection form completely and accurately. Print clearly and in the correct boxes. Incomplete or missing information may not be amended once the sample is received at the Missouri State Public Health Laboratory (MSPHL). A repeat screen may be necessary.

Provide two phone numbers to contact the mother (mother's number and/or that of a neighbor, relative, or friend), and **make sure the correct primary care provider who will continue seeing the baby is indicated**. It is essential that this information is accurate in case the family needs to be immediately contacted due to high risk screening results.

Featured Disorder continued from page 1

Symptoms of salt-wasting CAH appear within days or weeks of birth, and if undiagnosed, can result in death. Symptoms can include: failure to thrive, lethargy, irritability, vomiting, diarrhea, tachycardia, hypoglycemia, metabolic acidosis, and hypotension. Other signs of salt-wasting CAH are due to low cortisol levels and high androgens. These include ambiguous genitalia in females or enlarged genitalia in males.

Newborns with a classic CAH form should start treatment very soon after birth to reduce the effects of CAH. Classic CAH is treated with steroids that replace the low hormones. The main form of treatment for classic forms of CAH is hydrocortisone pills. Patients with salt-wasting CAH require additional treatment with fludrocortisone, which replaces aldosterone.

Simple virilizing, non-salt wasting, CAH accounts for about 25% of classic CAH cases. This form is less severe than salt-wasting CAH, but more severe than non-classic CAH, due to a lesser deficiency of aldosterone. There are also no severe or life-threatening sodium-deficiency symptoms in newborns with non-salt wasting CAH. However, like salt-wasting CAH, non-salt wasting CAH involves too little cortisol and too much androgen. Signs of non-salt wasting CAH begin before birth. These signs differ between males and females, which usually become apparent during childhood due to early puberty. Early signs of non-salt wasting CAH include enlarged clitoris, resembling a small penis; labia that are fused together, resembling a scrotum; or small testicles.

While the majority of CAH cases are classic, less than 1% are affected by non-classic CAH. Non-classic CAH is much less severe than classic forms of CAH. Signs of non-classic CAH can begin in childhood, adolescence, or adulthood. Both males and females with non-classic CAH may show signs of early puberty.

People with non-classic CAH may not need treatment if they do not have symptoms. Individuals with symptoms are given low doses of the same cortisol replacing medication taken by people with classic CAH. If symptoms resolve in adulthood, treatment may be discontinued. Females with salt-wasting CAH, who are born with ambiguous external genitalia, may need surgery. For example, surgery is necessary if changes to the genitals have affected urine flow. Females with non-classic CAH have normally developed genitals.

To learn more about living with CAH visit https://www.caresfoundation.org/what-is-cah/.

Tech Tips

- It is important for all newborn screenings to identify the doctor who will care for the newborn after discharge. Provide the physician's first and last name, and/or a clinic name, and office phone number. If this information is missing, it could delay timely follow-up needed for any abnormal results and prevent timely treatment of some life threatening disorders.

- Remember to complete the newborn screening form in its entirety for every screen collected for each baby. It is important to have all the requested information filled out on the form as completely and accurately as possible. This includes the collection date and time as well as the birthdate and time of birth. If this information is not accurately completed, this could prevent some of the testing from being resulted and a repeat screening will be required.

- Newborns that are younger than 24 hours and do not pass the hearing screening should be screened the next day. Newborns born by C-section are apt to have more fluid in the ear for longer periods. It is appropriate to conduct a third-day hearing screening when a baby born via C-section has not passed on two previous days. The last hearing screening result obtained prior to discharge is the hearing screening result that should be entered into MoEVR as the initial newborn hearing screening result. The MoEVR rescreening page is for hearing screening results that are obtained one to two weeks after birth.

Patient Spotlight

Kenzie

"Kenzie was born right on her due date in July of 2019. The next afternoon we were transferred to the NICU due to a variety of reasons, but after a few days, Kenzie appeared to have a good foothold. On the following Friday, our lives changed in a way we never could have predicted. Our NICU physician solemnly sat us down to share with us the news that Kenzie's newborn screen showed she was testing positive for Spinal Muscular Atrophy (SMA). SMA is a rare genetic neuromuscular condition of which we had no known family history. By the following Monday, we were already sitting in a specialist's office discovering that over the weekend they had confirmed Kenzie's diagnosis through additional testing.

People with SMA are missing the genetic coding for a crucial protein that is present to fuel motor neurons, and thereby muscles, throughout the body's core. In short, this meant that without treatment Kenzie would begin to decline around 2-3 months old and likely die sometime before 2 years of age. As you can imagine that day will be forever imprinted on our minds and hearts; not only because of her diagnosis before ever showing clear signs but also because we were simultaneously informed that acting early and pre-symptomatically to treat SMA can make all the difference. As a result of her early diagnosis, Kenzie was successfully treated at four weeks of age with a revolutionary and newly approved gene therapy, Zolgensma. While we are still waiting to understand what her capabilities will be, at 9 months she can sit up on her own. This is something she never would have done without early treatment. She is happy and thriving! Maybe one day she will walk, maybe not, but simply knowing she will live is life-changing.

None of this would have been possible without early diagnosis through newborn screening and fast action by our medical teams (you know who you are!). If we hadn't been informed of Kenzie's condition right away her quality of life moving forward would be much different. Including rare diseases on the newborn screen, especially those with treatment available, is absolutely critical and life-changing. THANK YOU MISSOURI for being among the first states to start including SMA in newborn screening and thereby saving our sweet girl's life."

Kenzie's Mom & Dad

Did You KNOW?

Did you know newborn screening in Missouri is continuing as usual? The Missouri Newborn Screening Program would like to extend our extreme gratitude for all health care workers responding to the COVID-19 crisis. We understand that the crisis has called for unprecedented adjustments to patient care. All newborn screening laws, rules, and recommendations remain unchanged. Please ensure that all babies receive blood spot, hearing, and critical congenital heart disease screening. Newborn screening is most complete and reliable when done after the baby is 24 hours of age. If baby must be discharged before 24 hours of age, please collect blood spot screening prior to leaving the hospital regardless of age. Assist parents in scheduling a repeat screen as soon as they may safely visit a collection site.

If current hospital COVID-19 policies prevent hearing screening, please help parents schedule a hearing screening as soon as safely possible.

The Missouri Newborn Hearing Screening Program will continue its follow-up process of contacting families and medical professionals via mail, fax, or phone. We will strive to meet the Joint Committee on Infant Hearing 1-3-6 recommendations to complete the hearing screen by 1 month of age, complete the audiological diagnostic evaluation by 3 months of age, and complete enrollment into early intervention by 6 months of age. However, we understand there may be delays in the follow-up process during this uncertain time.

During the COVID-19 crisis, it is crucial that CCHD screening is completed in the same manner as usual and acted upon as indicated.

Early diagnosis and intervention are critical for babies with conditions identified through newborn screening. Thank you for assisting parents in understanding the importance of completing recommended follow-up as soon as they are able to do so. For more information, visit https://health.mo.gov/living/families/genetics/newbornscreening/index.php.

MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

Bureau of Genetics and Healthy Childhood
Newborn Blood Spot, Hearing, and CCHD Programs
573.751.6266 or 800.877.6246

Missouri State Newborn Screening Laboratory
573.751.2662
www.health.mo.gov/newbornscreening

@HealthyLivingMo @HealthyLivingMo