Indiana State Department of Health
Maternal Child Health Division

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Genomics & Newborn Screening (GNBS) News

Hello Newborn Care Team!

September is newborn screening awareness month! Each year, nearly 4 million babies are routinely screened for certain genetic, metabolic and endocrine disorders prior to discharge from a birthing center or hospital using only a few drops of blood from the newborn's heel as part of this large public health program.

There are three parts to a newborn screen: heelstick, hearing and pulse oximetry. Newborn screening is the first step toward providing lifelong care, but often additional testing is needed to confirm a diagnosis. Genetic testing is an ideal follow-up for conditions that may provide families with additional information for best health outcomes.

Newborn screening began in the United States in 1963 when Dr. Guthrie discovered screening for phenylketonuria (PKU). Now, 50 years later, there are newborn screening tests for more than 30 treatable conditions, and newborn screening leads to treatment and early intervention for thousands of babies every year. Newborn screening is essential and is one of the greatest public health achievements in the United States.

Contact Us!

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Early Hearing Detection & Intervention (EHDI) Team

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Please email the GNBS team for further help or to ask questions!

Visit our website for educational materials and resources at www.NBS.IN.gov and www,birthdefects.IN.gov

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Indiana Newborn Screening Facts

In Indiana, newborns are screened for 49 genetic conditions. This includes sickle cell anemia, cystic fibrosis, endocrine disorders, metabolic disorders, hearing loss, critical congenital heart disease, severe combined immunodeficiency and spinal muscular atrophy. Three new conditions added to the Indiana screening panel in 2020. These conditions are Krabbe, Pompe and MPS-I(Hurler).

In 2018, Indiana had a total of 94,676 heelstick screens completed including initial and repeat screens. There were a total of 2,075 abnormal screens requiring repeat heelstick and/or confirmatory testing. A total of 1,510 babies were confirmed with a disease or trait from the newborn screening panel.

To learn more about GNBS, please visit our website at www.NBS.IN.gov.

Did you know?

Providers can view newborn screening (NBS) results on the Indiana Newborn Screening Tracking & Education Program (INSTEP) or by contacting the NBS laboratory. For more information or access, please email ISDHNBS@isdh.IN.gov.

Need information for a specific GNBS condition?

The American College of Medical Genetics and Genomics (ACMG) provides great resources, including provider ACT or "ACTion" sheets and algorithms for newborn screening conditions. These ACT sheets offer health care providers recommended short -term actions following an abnormal result that is received from the NBS lab, as well as basic information about the disorder. The ACMG ACT Sheets and algorithms are also available at your fingertips on your smartphone or tablet on the ACT Sheet Mobile App.

Visit www.acmg.net for further information or www.NBS.IN.gov for links to these resources.

For family-friendly information on newborn screening conditions, please visit our website or www.newbornscreening.info.

BDPR
Indiana Birth Defects and Problems Registry

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