

FREQUENTLY ASKED QUESTIONS

What is newborn screening?

Newborn screening is a set of screenings a baby receives after birth. Newborns receive a dried blood spot (DBS) screen, a critical congenital heart defect screen (CCHD), and a hearing screen. These screens should be performed when babies are 24 to 48 hours old. If the baby was born in a hospital, the newborn screening tests will be completed prior to being discharged. If the baby was born at home or in a birthing facility, the midwife or doctor attending the birth will provide information as to what steps need to be taken to have the newborn screening tests completed.

What are the ten mandated disorders screened for in Pennsylvania via the dried blood spot screen?

Newborns must be screened for the following disorders:

Phenylketonuria (PKU);
Maple syrup urine disease (MSUD);
Sickle Cell Anemia;
Congenital hypothyroidism (CH);
Congenital adrenal hyperplasia (CAH);
Classic Galactosemia (GAL);
Glycogen Storage Disease Type II (Pompe Disease) (GAA);
Mucopolysaccharidosis Type 1 (MPS-1);
X-linked Adrenoleukodystrophy (X-ALD);
Spinal Muscular Atrophy (SMA) – **Effective 3/1/2019**

Can a newborn be screened for more than the ten disorders tested from the DBS?

Yes, the Pennsylvania newborn screening panel includes a mandatory screening panel and a mandatory follow-up panel. The mandatory follow-up panel includes 27 additional disorders that can be screened from the DBS. For a complete list of all newborn screening blood spot tests, please go to: www.doh.pa.gov/newbornscreening. It is important that expectant parents talk to the baby's doctor before birth to determine which conditions are screened for at the birthing facility. Different birthing facilities screen for different conditions. Parents can request their newborn be screened for a condition that is on the list but the birthing facility indicates it is not a standard condition that they screen for.

How is the dried blood spot screen performed?

A few drops of blood are taken from the newborn's heel, placed on a filter paper and sent to the newborn screening laboratory.

Is there a fee for newborn screening?

Pennsylvania does not charge for newborns to be screened for the ten mandated disorders tested on the dried blood spot screen. The cost for CCHD screening, hearing screening, and additional DBS screenings may be covered by insurance.

How can I find out the results of the newborn screen?

The results of the hearing screen and the CCHD screen are available immediately. The results of the DBS screen are usually available seven to 10 days after the baby is born. The baby's doctor should be able to provide the results. If the baby's doctor is unable to provide the results, the baby's doctor can request the results be sent from the Division of Newborn Screening and Genetics.

Why would the screening need to be repeated?

- **DBS** – If the screen was completed before the baby was 24 hours of age, if the blood was unable to be tested, or if a result required additional screening. Parents should take the baby for a repeat screen right away, unless told otherwise.
- **Hearing** – If the screen was unable to be performed or if the newborn failed the in-patient screen.

What happens if a result is abnormal?

- **DBS** - The baby's doctor will contact the parents and talk about the results. The newborn may need to see a specialist for more testing.
- **Hearing** – A repeat out-patient screening should be performed. It is recommended the out-patient hearing screening be completed by one month of age. Follow the health care provider's instructions.
- **CCHD** - The newborn should have further cardiac testing.

Are there cures for the DBS screened disorders?

There are no cures for these disorders, but there are treatments. Treatments vary by disorder and may include medication, special diets or a special formula. If the disorder is identified early, newborns may lead a healthy life with the help of these treatments.

Where can I get more information?

For more information visit www.doh.pa.gov/newbornscreening.