



The Hospital + Healthsystem
Association of Pennsylvania

Leading for Better Health

Facts About Newborn Screening in Pennsylvania

Legislative Authority

Act of September 9, 1965, (P.L.497, No.251), known as the [Newborn Child Testing Act](#).

Department of Health Regulations

[Screening and Follow-up for Diseases of the Newborn](#)

Newborn Screening Process Newborn screening is intended to ensure that all babies are screened for certain serious conditions at birth, and for those babies with abnormal screening results, assist in appropriate referral for diagnosis, treatment, or early intervention.

The Pennsylvania Department of Health has oversight responsibility for the newborn screening process. The process is as follows:

- A supply of filter paper is shipped to each hospital from the laboratory that the Pennsylvania Department of Health has contracted with to perform the newborn screenings
- When a baby is born, a heel stick is performed on the newborn and the blood taken from the heel stick is deposited on the filter paper
- The filter paper is then sent to the laboratory, which performs the designated newborn screenings
- Lab results are first sent to the hospital that submitted the filter paper, the primary care physician on record, and the Pennsylvania Department of Health
- Hospitals are required to inform the parents of the lab results

If the lab results from the newborn screening indicate an abnormal result for one of the screened diseases, the hospital is required to make a referral to a treatment center.

Mandated Screenings

Currently, mandatory newborn screenings must be completed and reported to the Pennsylvania Department of Health for the following seven diseases:

- Phenylketonuria
- Maple syrup urine disease
- Sickle-cell disease
- Galactosemia
- Congenital adrenal hyperplasia
- Primary congenital hypothyroidism
- Pompe disease

Supplemental Conditions for Mandatory Follow-Up

[Newborn Child Testing Act \(Act 36 of 2008\)](#) and [Screening Legislation Act 36](#)





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February 2016

Page 2

Diseases for which supplemental screening tests are done under section 3(a)(2) of the Act include:

- Organic Acid Disorders
- Fatty Acid Oxidation Disorders
- Amino Acid Disorders
- Cystic Fibrosis
- Biotinidase Deficiency
- Severe Combined Immunodeficiency
- Krabbe
- Fabry
- Niemann-Pick
- Gaucher
- Hurler Syndrome

As outlined in the law, if a hospital opts to perform the supplemental follow-up newborn screenings for the diseases listed under section 3(a)(2), then the hospital is required to report the results to the Department of Health.

Screening for Critical Congenital Health Defect (CCHD) by Pulse Oximetry

Babies born with congenital heart defects are at significant risk for death or disability if their condition is not diagnosed soon after birth. Seven defects are classified as CCHD and can be detected using pulse oximetry screening, which is a test to determine the amount of oxygen in the blood and pulse rate. Act 94 of 2014, requires that all birthing facilities perform CCHD screening using pulse oximetry on a newborn child in its care before discharge from a birthing facility. Babies with a CCHD should be seen by cardiologists and can receive specialized care and treatment that could prevent death or disability early in life.

Hearing Screening

Hearing screening is a short test to determine if newborns might have hearing loss. Hearing screening is easy and not painful. There are two ways to screen a baby's hearing. The first test measures a tiny sound from the ear that indicates that the baby is hearing properly. This test is called an Otoacoustic Emissions test. The second test measures the baby's nerve response to sound. This is called an Auditory Brainstem Response. All babies should be screened for hearing loss no later than one month of age. It is best if they are screened before leaving the hospital. For more information on hearing screening, please see the [Infant Hearing Education, Assessment, Reporting and Referral Act 89 of 2001](#).

Resources

Information is available about newborn screening and other Department of Health programs at the Department of Health [website](#).

A list of all the conditions on the mandatory and supplemental/follow-up only panel is available on the Department of Health website at [Conditions for Screening and Follow-Up](#).

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