

Notifying parents about the need for follow-up testing for metabolic disorders

Why this Q&A Sheet is Important:



This Q&A document identifies questions commonly asked by parents when they are notified about the need for follow-up testing for their infant. It was developed in response to recent research suggesting that notification to the parent about the need for follow-up testing for metabolic disorders is a critical point in time, which can have long-term consequences including psychological stress in parents and perhaps even lack of adherence to medical recommendations. Research shows that taking the time to clearly

communicate in person the need for additional testing may reduce negative long-term implications for parents. Therefore the focus of this document is to assist in the notification process to parents of newborns who have out-of-range newborn screening results.

If your patient has abnormal newborn screening results for a metabolic disorder, personnel from the state newborn screening program will contact you about the results, possibly identify the disorder in question, and explain the potential risk to your patient.

Depending on these results, you will be advised to contact the parents and inform them of the need to:

- (a) refer their infant to a metabolic center;
- (b) obtain immediate follow-up testing for their infant; or
- (c) obtain follow-up testing within the following week.

Please note: This document refers specifically to **metabolic disorders**, such as: *Phenylketonuria (PKU), Biotinidase deficiency, Galactosemia, Maple Syrup Urine Disease (MSUD), Homocystinuria, Medium-chain acyl Co-A Dehydrogenase deficiency (MCADD), Tyrosinemia I & II, HMG-CoA Lyase deficiency, Argininosuccinic Acidemia, Isovaleric Acidemia, HHH Syndrome, Glutaric Acidemia I & II, Citrullinemia, Argininemia, CPT deficiency, Propionic Acidemia, Methylmalonic Acidemia, β -Methylcrotonyl-CoA Carboxylase deficiency, LCHADD, VLADD, SCADD, β -Ketothiolase Deficiency.*

If your patient needs follow-up testing for a **non-metabolic disorder**, such as *congenital hypothyroidism, congenital adrenal hyperplasia, hemoglobinopathy (such as sickle-cell disease), or congenital toxoplasmosis*, then this document is not intended for you.

Commonly Asked Questions:

Why does my child need follow-up testing?

Follow-up testing is common and does not necessarily mean that your infant has a metabolic disorder. There are many reasons why your child may need follow-up testing. For example, blood for the initial screening test may have been collected too early or there wasn't enough blood in the sample. It is estimated that only 1 out of every 12 babies who receive follow-up testing actually have a metabolic disorder. Abnormal screening results are often due to premature birth and/or low birth weight but can also indicate a disorder.

When was my infant initially screened? I don't remember this being done.

Soon after your infant was delivered a nurse took a few small drops of blood from your infant's heel. This sample was used to run a number of newborn screening tests. These tests identify babies with metabolic disorders so that, if they are identified, they can be treated before any negative health consequences occur. Many parents don't remember newborn screening occurring.

I heard that false-positive test results are common with newborn screening. What is a "false-positive"?

A "false-positive" is when initial screening shows a positive result but follow-up testing indicates that the child does not have a disorder. "False-positives" occur in newborn screening because screening is designed to detect even slight variations in the newborn screening results, many of which will be "false-positive". However, metabolic disorders can be life-threatening and so it is important to make the test sensitive to these slight variations in order to be certain that every infant who has a disorder will be detected. The problem is that some parents may be unnecessarily alarmed until additional testing is able to rule out a metabolic disorder.

What is my child receiving follow-up testing for?

Your child is having follow-up testing for a metabolic disorder.

(You may choose to provide information on the range of disorders, such as fatty acid oxidation disorders, or the specific condition for which your patient is having follow-up testing)

Commonly Asked Questions:

What is a metabolic disorder?

Metabolism is the process by which the body breaks down, stores and transforms food into energy for use by the body. A metabolic disorder is an inherited condition that can lead to abnormal levels of chemicals in the body due to its inability to undergo normal metabolic processing. If metabolic disorders are not detected and treated early they can lead to significant health problems.

When will I get a final diagnosis?

You will be called within (a few days/weeks) with the results. If you do not hear from us by (specific time) please feel free to call my office at ###-####.

What happens if my child is diagnosed with a metabolic disorder?

If your child is diagnosed with a metabolic disorder you will be referred to a metabolic center where he/she will be provided with medical treatment, such as frequent feedings, a special diet, and/or medication to prevent serious health consequences. You should follow all instructions provided by the doctor(s) at the metabolic center. You can also contact me (your pediatrician) at any time as I will be in touch with the metabolic specialist who will be looking after your child. It is important to remember that treatment is available for all the metabolic disorders currently screened for in New England.

¹ Waisbren, S.E. Albers, S. Amato, S. et al. (2003). Effect of Expanded Newborn Screening for Biochemical Genetic Disorders on Child Outcomes and Parental Stress. *JAMA*, 290, 2564-72.

² Zytkevich, T.H., Fitzgerald, E.F., Marsden, D., et al. (2001). Tandem mass spectrometric analysis for amino, organic, and fatty acid disorders in newborn dried blood spots: a two-year summary from the New England Newborn Screening Program. *Clin Chem*. 47, 1945-1955.

For More Information

- For more information about your patient's newborn screening results, you can contact your public health department's newborn screening program and request a copy of your patient's screening results.
- For more information on metabolic disorders, visit the "**Physicians & Scientists**" section of the New England Consortium of Metabolic Programs website at: www.newenglandconsortium.org. This site contains:
 - descriptions of metabolic disorders
 - newborn screening protocols
 - commonly asked questions on specific disorders
 - acute illness protocols (ACT sheets).
- Contact a metabolic center if you require information or advice at any time.