

**Phenylketonuria (PKU)**

*Parent Fact Sheet*

A newborn screening test is a **screen** and not diagnostic testing. An “abnormal” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is **very important** to determine if your baby has the disorder indicated.

**Disorder Indicated:** Classic Phenylketonuria (PKU) is a condition in which the body cannot break down phenylalanine, an amino acids found in proteins. If left untreated, PKU can cause developmental delays, brain damage or even death. However, if the condition is detected early and treatment is begun, individuals with PKU can lead healthy lives.

Incidence	1 in every 10,000 to 15,000 newborns.
Signs and Symptoms  <i>Please note: these findings may not be present in young infants or in milder forms of the disease</i>	When a child has classic Phenylketonuria, you may see symptoms including: <ul style="list-style-type: none"> <li>• Irritability</li> <li>• Seizures (epilepsy)</li> <li>• Dry, scaly skin (eczema)</li> <li>• “Musty” or “mouse-like” body odor</li> <li>• Pale hair and skin</li> <li>• Developmental delays</li> </ul>
Next Steps <i>may</i> include:	<b>Follow up with your child’s pediatric provider</b> Clinical assessment Laboratory Testing
Treatment <i>(if indicated)</i>	Restricted diet (follow up with your child’s pediatric provider)
Additional Resources	VDH Newborn Screening <a href="http://vdhlivewell.com/newbornscreening">http://vdhlivewell.com/newbornscreening</a> Baby’s First Test <a href="http://www.babysfirsttest.org">www.babysfirsttest.org</a> Genetics Home Reference <a href="https://ghr.nlm.nih.gov/">https://ghr.nlm.nih.gov/</a> Children’s PKU Network <a href="http://www.pkunetwork.org/Childrens_PKU_Network/Home.html">www.pkunetwork.org/Childrens_PKU_Network/Home.html</a>

*Educational content adapted from [www.babysfirsttest.org](http://www.babysfirsttest.org)*

