



Genetics Newborn Screening Program

Family Fact Sheet

PHENYLKETONURIA (PKU)

Protein is an important part of our diet. Protein can enhance body growth and strength when enzymes break the protein down into its proper parts. Babies who have the disorder called phenylketonuria (FEE-nil-KEE-tone-u-ree-ah) cannot break down the amino acid phenylalanine (FEE-nil-AL-an-eeen). Babies with phenylketonuria (PKU) have high levels of phenylalanine in their body. If PKU is not treated, the high levels of phenylalanine can cause mental retardation, cerebral palsy, skin problems, and other related physical problems.

Connecticut began screening for PKU in 1964. In Connecticut, about 1 in every 10,409 newborn babies has PKU.

How does a baby get PKU?

PKU is an inherited condition. Both the mother and the father have one altered gene that will not allow the body to break down phenylalanine. A baby has to receive this gene from both the mother and the father to have this condition.

How is a baby tested for PKU?

Prior to discharge from the facility of birth, a small sample of blood is taken from the baby's heel. The sample is then sent to the State laboratory to screen for PKU. Abnormal screening results are reported to the Department of Public Health (DPH), Genetic Newborn Screening Tracking Unit nurse consultant. If the results are positive, the DPH nurse consultant will notify the baby's primary care physician of the need for a prompt referral to one of the Genetic Regional Treatment Centers at Yale or the UCONN Health Center for confirmation testing and follow-up treatment if necessary.

What is the treatment for PKU?

The Regional Treatment Center specialists will prescribe treatment for PKU if necessary, which includes a special diet restricted in phenylalanine and a specialized metabolic formula. Early diagnosis of PKU leads to early treatment, which is important for the health of your baby.

The information provided is offered for general informational and educational purposes only. It is not offered as and does not constitute medical advice. In no way are any of the materials presented meant to be a substitute for professional medical care or attention by a qualified practitioner, nor should they be construed as such. Contact your physician if there are any concerns or questions.



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