

Family action sheet for phenylketonuria (PKU)

If a newborn blood screening result shows that your baby could have a serious condition called phenylketonuria (PKU), this result does not mean that your baby has this condition, but more tests are needed.

All babies born in California have a routine blood screen shortly after birth. The goal of this newborn screening is to find those at risk for serious medical conditions. Babies can look healthy at birth and still have one of these conditions. Babies with these conditions benefit from early diagnosis and treatment.

What is phenylketonuria or PKU?

PKU is inherited (passed down) in families where both parents are PKU carriers (that is, they do not have the disorder themselves). In PKU, there is a defect in a gene. The gene's job is to produce a substance that breaks down (metabolizes) an amino acid called phenylalanine. Phenylalanine is in the protein we eat. Left untreated, excess phenylalanine builds up in the blood and brain and can cause intellectual disabilities, convulsions, and skin problems such as eczema.

Is there treatment for PKU?

Yes, there are treatments for PKU. They are very effective at preventing the severe effects of untreated PKU if they are started soon after birth. Babies with PKU drink a medical formula product prescribed by a doctor. Use of this formula continues through childhood and adulthood. Persons with PKU also follow a low-protein diet for life. Alternative therapies are available for adults. Many new treatments and even cures are in development.

Newborn screening takes blood from a newborn's heel to screen for medical conditions



Next steps

1. Your baby's health care provider will help arrange more testing as soon as possible

These tests will let you know if your baby has PKU.

2. Work closely with your baby's health care provider and follow recommendations

Your baby's provider may refer your baby to a doctor with more experience in PKU. It is important that you keep all appointments and start treatment if instructed.

Where can I get more information?

Your baby's health care provider or PKU doctor is the best person to teach you about PKU. Find more information at these websites:

- **Baby's First Test** (<https://www.babysfirsttest.org>). Type "PKU" into the "Find a Condition" Box. Follow the link. For Spanish, click on the top right black box marked "en Español"
- **MedlinePlus** (<https://medlineplus.gov/genetics/condition/phenylketonuria/>)
- **California Coalition for PKU and Allied Disorders** (ccpkuad.org)

