



Genetics Newborn Screening Program

Health Professional Fact Sheet

PHENYLKETONURIA (PKU)

Phenylketonuria (PKU), a metabolic disorder, is the result of a defective phenylalanine metabolic pathway. The absence or deficiency of phenylalanine hydroxylase prohibits the conversion of phenylalanine to tyrosine. This causes phenylalanine to accumulate at elevated levels in the blood. Patients with undiagnosed PKU have progressive developmental delay in the first year of life, severe mental retardation, seizures, autistic-like behavior, and a peculiar odor. Screening in Connecticut was instituted in 1964.

GENETIC BASIS

- Autosomal recessive condition
- Recurrence risk is 25%
- Prenatal diagnosis and carrier testing is available by molecular testing

CT PREVALENCE 1:10,409

CLINICAL FEATURES OF UNTREATED DISEASE

Severity

- Physical disabilities: microcephaly, seizures, cerebral palsy, hyperactivity, eczema, musty body odor
- Developmental disabilities: severe mental retardation; IQ < 50 in 95% of untreated individuals, autistic-like behavior
- Mortality: not fatal

Symptomatic diagnosis

A symptomatic diagnosis is difficult, as symptoms may not appear until after irreversible neurological deficits are present.

Variants

- Mild persistent hyperphenylalaninemia and variant PKU are milder forms of PKU. Patient's blood phenylalanine levels are less (2-20 mg/dl) than those with classic PKU (>20 mg/dl). This blood phenylalanine may remain elevated throughout life with mild or absent developmental disabilities.
- Women with elevated blood phenylalanine concentrations are at risk for having children with maternal PKU effects including mental retardation, microcephaly, intrauterine growth restriction and post-natal growth retardation, and congenital heart disease.

CLINICAL OUTCOME WITH SCREENING AND TREATMENT

With optimal dietary control, which needs to be maintained throughout life, normal growth and development and normal range of intelligence should result.

SCREENING FOR PKU

Laboratory tests

- Tests for levels of phenylalanine on filter paper blood specimen.
- Testing is by Tandem Mass Spectrometry.

Abnormal Test Results

- The primary care provider is notified by the DPH, Genetic NBS Tracking Unit nurse consultant.
- A referral is made by the DPH, Genetic NBS Tracking Unit nurse consultant to one of the Genetic Regional Treatment Centers.
- The primary care provider will be advised by the DPH, Genetic NBS Tracking Unit nurse consultant to contact the Genetic Regional Treatment Center to make a prompt referral and arrange for confirmation testing and evaluation.



Keeping Connecticut Healthy

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