Phenylketonuria (PKU)

Phenylketonuria is an inherited autosomal recessive disorder of phenylalanine metabolism. PKU is usually detected within the first few days of life by newborn screening. It is characterized by the absence or deficiency of an enzyme, phenylalanine hydroxylase, that allows phenylalanine to accumulate in the blood and is toxic to brain tissue. Early detection and treatment is essential to prevent associated neurological symptoms and mental retardation.

Prevalence:	1:20,000
Analytes Measured:	Phenylalanine
Reporting Ranges:	Elevated phenylalanine
Feeding Effect:	None
Timing Effect:	$<$ 24 hours of age: Repeat screen within 7 days $\geq$ 24 hours of age: Results are valid
Confirmation:	Repeat newborn screen.
Treatment:	Referral is made to a Metabolic Specialist. Dietary restrictions of phenylalanine with regular monitoring of serum phenylalanine levels.
Comments:	Without treatment, most infants develop mental retardation that is usually severe. These infants may also develop additional neurological symptoms such as seizures, hyperactivity and aggressive behavior. With good dietary control and monitoring, these potential effects of PKU can be minimized.

Maine Newborn Screening Program
(207) 287-5357 or 1-800-698-3624