PKU Phenylketonuria genetic test

General:

Phenylketonuria is an autosomal recessive metabolism disease. The frequency of phenylketonuria is between 1: 5,000 and 1:10,000. The gene encoding phenylalanine hydroxylase is located on chromosome 12q22 to 12q24. Due to complete enzyme deficiency in the liver, phenylalanine accumulates in blood and is eliminated via kidneys. Untreated children show severe mental deficiencies, epileptic attacks, hypertonia of the muscles, growth retardation of the brain, pigment disturbances of the skin, eczemalike skin symptoms, general over-excitability and characteristic urine and sweat smell.

OMIM	261600
Gene:	PAH, Locus 12q22 to 12q24
Indication:	Positive newborn screening, hyperphenylalaninemia
Material:	5 ml EDTA blood
TAT:	7-10 days*
Method:	PCR, sequencing of all 13 exons
Ref.- range:	see report

For complete list of laboratory test offered at Freiburg Medical Laboratory, please visit http://www.fml-dubai.com/parameter-listings/

Updated 25/05/2024