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# Phenylalanine

General:

Phenylalanine hydroxylase deficiency, synthesis defects of the cosubstrate tetrahydrobiopterin and phenylalanine hydroxylase maturation disorders result in hyperphenylalaninemia. Phenylketonuria (PKU) is observed, if no phenylalanine hydroxylase activity is detectable in liver.

**Symptoms**: eczematoid skin modifications, psychomotor retardation, neurological symptoms (aggressive behavior, muscle hypertonia, convulsions), microcephaly and disturbance in melanin synthesis. As preventive measure, hyperphenylalaninemias must be detected by biochemical screening programs already in the newborn!

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|---|---|
| Indication: | Suspicion of phenylketonuria |
| Material: | 1 ml serum |
| TAT: | 5-7 days* |
| Method: | HPLC |
| Units: | µmol/l |
| Ref.- range: | see report |
| Note: | see also **Newborn screening** |

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



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