Thalassemia

see **Hemoglobin electrophoresis**

General:

Thalassemia is an inherited hemoglobinopathy with increased HbA2 and HbF leading to a distinct hypochromic anemia in most cases. Thalasse-mias are classified according to clinical symptoms and to distinct hemoglobin defects.

Clinical classification:

Thalassemia minor: Heterozygous carriers, slight β-thalassemia, mostly increased HbA2; temporary or minimal subicterus, moderate hepatosplenomegaly, typical blood differential: microcytosis, hypochromasia, poikilocytosis and target cells. Signs of increased hemolysis: reticulocytes, LDH and indirect bilirubin increased, haptoglobin decreased. Prognosis: good

Thalassemia major: Homozygous offspring, i.e. both parents are heterozygous carriers, severe β-thalassemia with overproduction of HbF (up to > 95%), (whereas the homozygous α-form mostly leads to intrauterine death or severe connatal hydrops), symptoms start in infancy and early childhood. Lethal outcomes are observed at an early stage; erythroblastosis (megalo- and paraerythroblasts, reticulocytosis, basophil punctures), leukocytosis, delayed growth, hepatosplenomegaly, turricephaly or round skull (radio-graph picture: »hair-on-end appearance«). The patients require transfu-sions. The increased metabolism of erythrocytes leads to hemosiderosis and hemochromatosis with mental and physical retardation and pubertas tarda. Only few of the affected patients reach adolescence. Early manifestation of the disorder is associated with poor prognosis.

Classification:

α-Thalassemia: gene locus HBA1, HBA2, chromosome 16 pter-p13.3, normal HBA2 can point to alpha-thalassemia, hypochromic microcytic anemia.

β-Thalassemia: gene locus HBB, chromosome 11 p15.5 increase of HbA2, hypochromic microcytic anemia.

delta-Thalassemia (Lepore): Diagnosis: hemoglobin electrophoresis or HPLC, genetic clarification recommended.

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^ non-accredited parameter

*performed in a collaborating laboratory (Germany)

The following tests are available:

- **α-Thalassemia, genetic test**

Indication: Anemia, hypochromia, microcytosis
Material: 3-5 ml EDTA blood
TAT: upto 2 weeks*
Method: EXT
Ref.- range: see report

- **β-Thalassemia**

see **Hemoglobin electrophoresis**

Indication: microcytic iron refractory anemia, HBA2-, HBF-increase
Material: 3-5 ml EDTA blood
Stability: 7 days at 2 to 8°C
TAT: 3 days, FML
Method: HPLC
Ref.- range: see report

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

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