

# Thalassemia

see **Hemoglobin electrophoresis**

General:

Thalassemia is an inherited hemoglobinopathy with increased HbA<sub>2</sub> and HbF leading to a distinct hypochromic anemia in most cases. Thalassemias are classified according to clinical symptoms and to distinct hemoglobin defects.

Clinical classification:

**Thalassemia minor:** Heterozygous carriers, slight  $\beta$ -thalassemia, mostly increased HbA<sub>2</sub>; temporary or minimal splenomegaly, 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  $\beta$ -thalassemia with overproduction of HbF (up to > 95%), (whereas the homozygous  $\alpha$ -form mostly leads to intrauterine death or severe congenital hydrops), symptoms start in infancy and early childhood. Lethal outcomes are observed at an early stage; erythroblastosis (megaloblasts and paraerythroblasts, reticulocytosis, basophil punctures), leukocytosis, delayed growth, hepatosplenomegaly, turricephaly or round skull (radio-graph picture: »hair-on-end appearance«). The patients require transfusions. 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:

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

**$\beta$ -Thalassemia:** gene locus HBB, chromosome 11 p15.5 increase of HbA<sub>2</sub>, hypochromic microcytic anemia.

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

The following tests are available:

- **$\alpha$ -Thalassemia, genetic test**

Indication: Anemia, hypochromia, microcytosis

Material: 3-5 ml EDTA blood

TAT: upto 2 weeks\*

Method: EXT

Ref.- range: see report

- **$\beta$ -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/>