



GET IN TOUCH



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Beta-Thalassemia

HBB- gene testing

Description

In Beta-thalassemia, hemoglobin an iron containing protein is reduced due to mutations in the HBB gene. Reduced levels of hemoglobin leads to oxygen deficiency in many parts of the body.

Beta thalassemia is also known as

- ✦ Erythroblastic anemia
- ✦ Mediterranean anemia
- ✦ beta type-microcytemia
- ✦ beta type-thalassemia

Types of Beta-thalassemia

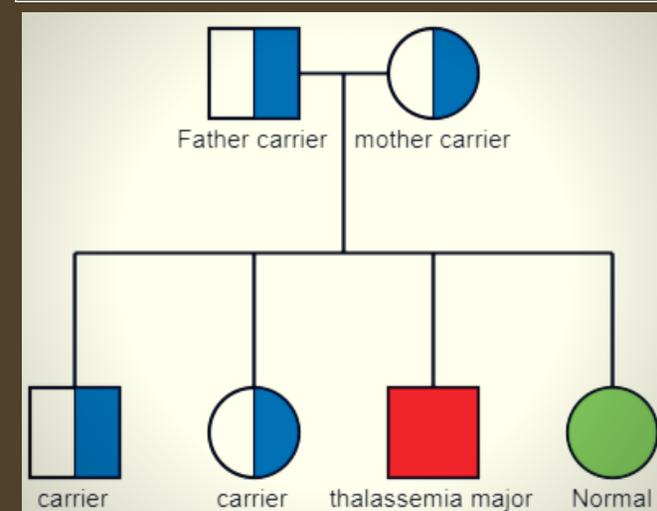
- ▶ Beta-thalassemia major
- ▶ Beta-thalassemia intermedia
- ▶ Beta-thalassemia minor

Effect of mutations

- ▶ beta globin production is prevented in some types of mutations
- ▶ beta globin production is reduced in some types of mutations

Inheritance

RECESSIVE



Most of the time thalassemia is inherited in a recessive pattern means that two copies of the gene need to be mutated to produce a disease

If father and mother are carriers, offspring probability

- 50% - thalassemia minor
- 25% - thalassemia major
- 25% - Normal

DOMINANT

Gene Testing

ARMS-PCR

SEQUENCING

- IVS 1-5
- IVS 1-1
- cd 41/42
- cd 8/9
- 619 bp deletion
- HbE (common mutations)

Test code: KIVFMG-002

Sample/Specimen:

8-10 ml Amniotic Fluid in two sterile 15 ml centrifuge tubes(pre-natal)/CVS(pre-natal)/2ml of EDTA blood

Shipment: Ship refrigerated immediately with cool packs do not freeze

Turn around Time: 72 hours from the day sample received in the laboratory

Full gene sequencing-Sanger method using 3500

Sequencing of specific mutation by Sanger method using 3500

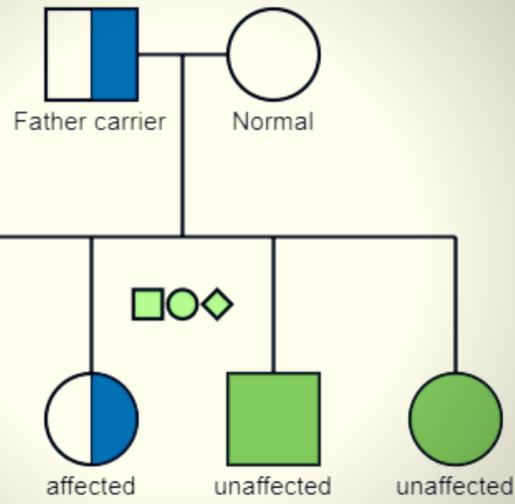
Test code: KIVFMG-003

Sample/Specimen:

8-10 ml Amniotic Fluid in two sterile 15 ml centrifuge tubes(pre-natal)/CVS(pre-natal)/2ml of EDTA blood

Shipment: Ship refrigerated immediately with cool packs do not freeze

Turn around Time: 3-4 days from the day sample received in the laboratory

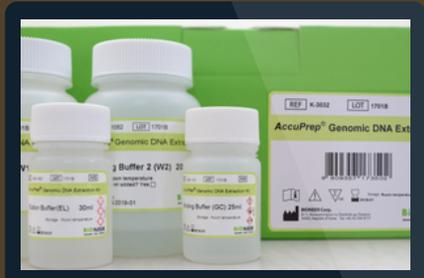


In some families it is inherited in dominant pattern. In this pattern one copy of mutated gene is sufficient to cause the disease.

Note: please contact the lab before sending prenatal samples

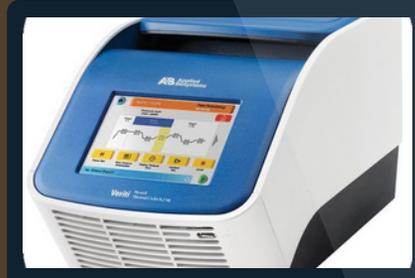
Test Procedure

ARMS-PCR



E

DNA Extraction



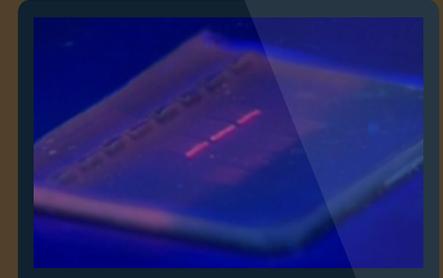
A

Amplification



E

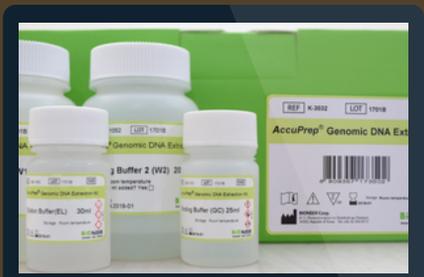
Electrophoresis



R

Results

SEQUENCING



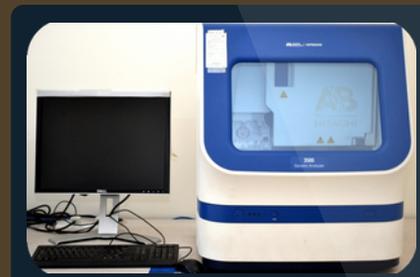
E

DNA Extraction



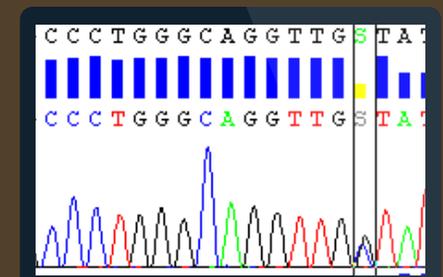
A

Amplification



S

Sequencing



R

Results