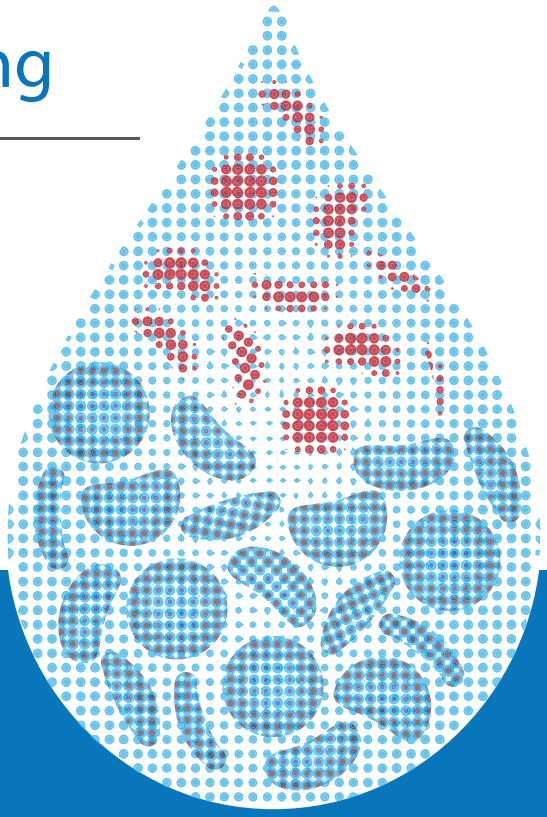
NOT JUST COMMON MUTATIONS!

MEDGENOME

Comprehensive Thalassemia Genetic Testing



Beta-thalassemia (β-thalassemia)

- It is characterized by reduced synthesis of the hemoglobin subunit beta (hemoglobin beta chain)
- This results in microcytic hypochromic anemia, an abnormal peripheral blood smear with nucleated red blood cells, and reduced amounts of hemoglobin A (HbA) on hemoglobin analysis.
- Beta-thalassemia (OMIM#613985) can be caused by homozygous orcompound heterozygous mutations in the HBB gene (OMIM*141900).

MedGenome's Genetic Testing for Beta-Thalassemia

- The sequencing of complete HBB gene can detect all the common point mutations and small indels * as well as novel and rare mutations.
- NGS based amplicon sequencing to analyse complete HBB gene.
- Covers UTR, promoter exonic and intronic regions along with 619bp deletion in HBB gene.

Test Code	Test Name	Mutation Type	Region covered	Methodology
MGM044	Beta thalassemia [HBB] gene analysis	SNVs / Indels	Complete HBB gene including common mutations.	NGS
MGM1763	Comprehensive Alpha and Beta thalassemia gene panel	SNVs / Indels / Common Mutations / 619 bp deletion / large CNVs	Complete HBB, HBA1 & HBA2 genes. Along with Partial sequencing of HBG1, HBG2 and HBD genes & promoter regions for CNV analysis	NGS

*List of common mutations covered:

Variant	cDNA position	Methodology
IVS1-5	c.92+5G>C	NGS
Codon41/42(-TCTT)	c.125_128delTCTT	NGS
Codon 30 (G>C)	c.92G>C	NGS
IVS1-1 (G>T/A)	c.92+1G>A	NGS
619-bp deletion	-	NGS
Codon 8/9(+G)	c.27dupG	NGS
Codon 15 (G>A)	c.47 G>A	NGS
Codon 16 (-C)	c.51delC	NGS
Poly A Site (T>C)	c.*110T>C	NGS
Codon 15 (-T)	c.46delT	NGS

List of CNV's covered:

Deletions covered	Methodology
290bp-deletion	NGS
Indian 619 bp β°-thal	NGS
Indian G γA γ (δβ)°-thal	NGS
SEA-HPFH	NGS
Hb-Lepore	NGS
Dutch I 12.6 kb β°-thal	NGS
Belgian 50 kb G γ(A γδβ)°-thal	NGS
Chinese G γ(A γδβ)°-thal	NGS
Filipino G γ(A γδβ)°-thal	NGS
Yunnanese G γ(A γδβ)°-thal	NGS

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Alpha-Thalassemia

- Alpha-thalassemia (α-thalassemia) has two clinically significant forms:
  1. Hemoglobin Bart hydrops fetalis (Hb Bart) syndrome- Caused by deletion of all four -globin genes.
  2. Hemoglobin H (HbH) disease- Most frequently caused by deletion of three -globin genes.
- Hb Bart syndrome, the more severe form, is characterized by fetal onset of generalized edema, pleural and pericardial effusions, and severe hypochromic anemia, in the absence of ABO or Rh blood group incompatibility.
- HbH disease is characterized by microcytic hypochromic hemolytic anemia, splenomegaly, mild jaundice, and sometimes thalassemia-like bone changes. Individuals with HbH disease may develop gallstones and experience acute episodes of hemolysis in response to oxidant drugs and infections.
- Alpha-thalassemia (OMIM#604131) is caused by mutations in the HBA1 (OMIM*141800) and HBA2 OMIM*141850) genes.

MedGenome's Genetic Testing for Alpha-Thalassemia

Test Code	Test Name	Mutation Type	Region covered	Methodology
MGM1769	Comprehensive alpha thalassemia gene analysis [HBA1 & HBA2]	SNVs / Indels / CNVs	HBA1 & HBA2 genes	NGS
MGM1763	Comprehensive Alpha and Beta thalassemia gene panel	SNVs / Indels / CNVs	Complete HBB, HBA1 & HBA2 genes. Along with Partial sequencing of HBG1, HBG2 and HBD genes & promoter regions for CNV analysis	NGS

List of common mutations covered:

Variant	Methodology
Common two α-globin-gene deletion	NGS
Common single α-globin-gene deletion	NGS
c.2T>C	NGS
c.94_95delAG	NGS
c.95+2_95+6delTGAGG	NGS
c.207C>G	NGS
c.207C>A	NGS
c.223G>C	NGS
c.[339C>G; 340_351delCTCCCCGCCGAG]	NGS
c.377T>C	NGS
c.*94A>G	NGS

List of CNV's covered:

Deletions	CNV detection method
--SEA	Direct Detection
--FIL	Direct Detection
--THAI	Direct Detection
-(α)20.5	Direct Detection
--MED	Direct Detection
-(α)21.9	Direct Detection
-(α)27.6	Direct Detection
-(α)3.7	Coverage based
-(α)4.2	Coverage based
HS-40	Coverage based
Other deletions	Coverage based

Get in touch

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