Capital Bio
TECHNOLOGY

Thalassemia Gene Mutations Detection Kit (Microassay)

The multiple allele-specific PCR-based universal array (ASPUA) technology is adopted to test the commonest thalassemia gene mutation site and deleted fragment in human body. It can synchronously complete 25 mutation tests of α-thalassemia and β-thalassemia gene at once, and the test result can assist for clinical diagnosis and be used for field of epidemiological survey,premarital examination and neonatal test.



Fast

Completing all tests with 4.5 h

Comprehensive

25 deletion, non-deletion α-thalassemia and point mutation β-thalassemia can be tested within one experiment

Simple

Operate automatically and test result can be automatically judged by software

Flexible

Whole blood, oral swabs and blood spot sample can be tested

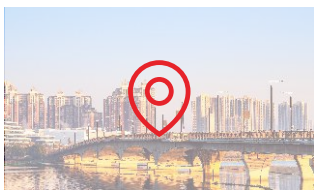
Applicable Population



Group with family history of thalassemia



Group whose spouse is the carrier or patient of thalassemia



Group in thalassemia high-incidence areas



Group who pay attention to premarital, progestation and antenatal health examination

Thalassemia Symptom

Thalassemia is hemolytic anemia caused by abnormal synthesis of red blood cell hemoglobin due to genetic defects, easy rupture of red blood cells, and shortened service life. Common types of thalassemia: α–thalassemia and β–thalassemia The thalassemia can be divided into heavy, intermediate, light and static models according to the severity of illness.

Type		Clinical Manifestation
α -thalassemia	Silent	No symptom; erythrocyte is normal; MCV value and MCH value is lower or normal.
	Slight	No symptom or slight anemia; the size erythrocyte is various, thecentral part is lightly stained; MCV value and MCH value is low.
	Intermediate	It names as Hemoglobin H disease as well, with large clinical difference, different anemia duration and different degrees. Most of symptoms gradually appear after infancy stage and special face similar with major β–thalassemia will appear to the elderly patient.
	Major	It names as Hb Bart's fetal edema syndrome. The fetus presented with anasarca, anemia, look pale, hepatosplenomegaly and died later after miscarriage or birth.
β -thalassemia	Slight	No symptom or slight anemia, the patient can live to old age.
	Intermediate	The symptoms (slight or moderate hepatosplenomegaly, moderateanemia) appear in toddler stage mostly.
	Major	The newborn has no symptoms at birth and suffers the disease from 3 to 12 months, such as chronic progressive anemia, presenting with look pale, hepatosplenomegaly or dysontogenesis. Regularly blood transfusion and treatment of iron chelating agent can maintain the life to about 20 years old while most of child patients will die before 5 years old if no treatment.

Test Process

Sample preparation 01



DNA extraction



PCR amplification

2.5 h

Chip reaction 02



Hybridization reaction



Chip cleaning

1 h

Detection and analysis 03



Chip scanning



Data analysis

5 min

Test Content

Type	Gene Defect (25 kinds)	
α–thalassemia	Deletion	––SEA; –α3.7; –α4.2
	Non–deletion	aQS; aCS; aWS
β–thalassemia	Point mutation	nt29 A>G, CD71–72(+A), Cap (–ACCC), ATG>AGG, CD14–15(+G), CD17A>T, CD26G>A, CD27–28 (+C), IVS–I–1 G>T, IVS–I–5 G>C, CD41–42(–TCTT), CD43G>T, CD30 A>G, nt28 A>G, VS–II–654 C>T, nt30 T>C, VS–II–5 G>C, CD37 G>A, nt32 C>A

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