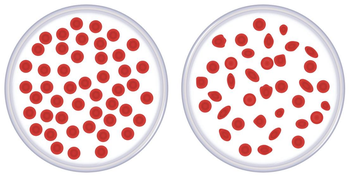
Thalassemia Reference Standard

Background

Thalassemia, also known as globin dysplasia anemia, is an inherited single-gene disorder caused by insufficient or absent synthesis of globin chains due to congenital defects in human globin genes. Based on the type of globin chain affected, thalassemia is primarily classified into α, β, γ, δ, and other subtypes, with α-thalassemia and β-thalassemia being the most prevalent forms.

Thalassemia has a high global prevalence, posing a significant threat to public health. It was first identified in regions including the Mediterranean, Middle East, Caucasus, Central Asia, the Indian subcontinent, and Southeast Asia.



Normal blood Thalassemia

Currently, the effective clinical prevention and diagnosis of thalassemia require blood sampling for globin chain analysis and genetic testing. The stepwise screening process typically involves routine blood tests, hemoglobin electrophoresis, and genetic testing, the latter being the gold standard for confirming a diagnosis of thalassemia.

Introduction

CB-Gene has launched Thalassemia Reference Standards, which include specialized controls for both α-Thalassemia and β-Thalassemia. These reference standards are developed to function as positive and negative controls in Thalassemia gene test kits, ensuring accuracy and reliability in diagnostic applications.

Additionally, they are designed for routine quality control and performance evaluation of the Thalassemia testing process. By supporting consistency and precision in genetic testing, these standards enhance the credibility of results, particularly in detecting Thalassemia mutations and improving overall laboratory workflows.

This innovation highlights CB-Gene's commitment to advancing the quality of molecular diagnostics and supporting effective disease management.

Product data

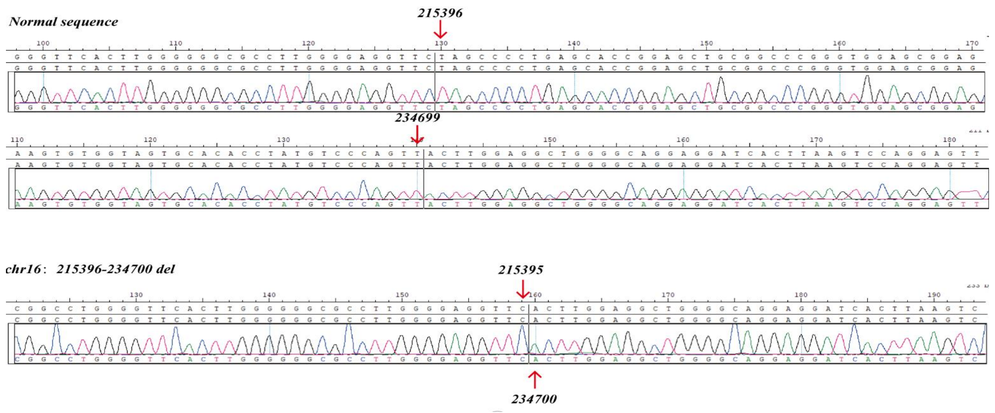


Figure 1: ***α-thalassemia αα/--SEA Reference Standard CBPD0029***

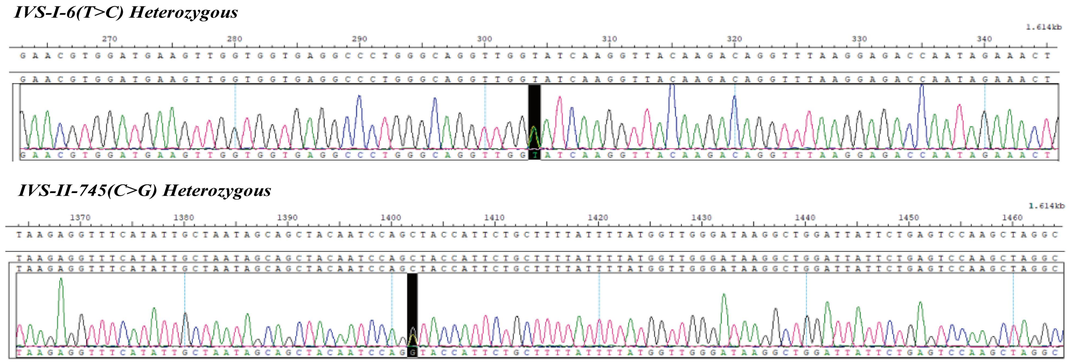


Figure 2: ***β-thalassemia IVS-I-6(T>C)&IVS-II-745(C>G) double mutation Reference Standard CBPD0010***

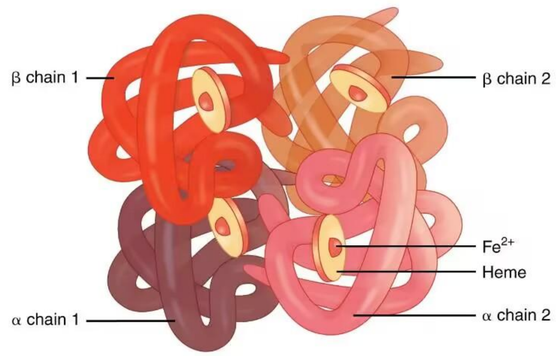
Product	Cat.No	Variation site	Allelic Frequency	Transcript
β-thalassemia Codon 39(C>T)&IVS-I-110(G>A) double mutation Reference Standard	CBPD0001	Codon 39(C>T)	50%	NM_000518.5
		IVS-I-110(G>A)	50%	NM_000518.5
β-thalassemia IVS-I-110(G>A) mutation Reference Standard	CBPD0002	IVS-I-110(G>A)	100%	NM_000518.5
β-thalassemia -87(C>G)&IVS-I-110(G>A) double mutation Reference Standard	CBPD0003	-87(C>G)	50%	NM_000518.5
		IVS-I-110(G>A)	100%	NM_000518.5
β-thalassemia Codon 6(A>T) mutation Reference Standard	CBPD0004	Codon 6(A>T)	50%	NM_000518.5
β-thalassemia Codon 39(C>T)&IVS-II-1(G>A) double mutation Reference Standard	CBPD0005	Codon 39(C>T)	50%	NM_000518.5
		IVS-II-1(G>A)	50%	NM_000518.5
β-thalassemia Codon 26(GAG>AAG)&IVS-I-1 (G>T) double mutation Reference Standard	CBPD0006	IVS-II-1(G>A)	100%	NM_000518.5
β-thalassemia IVS-II-654(C>T)&IVS-II-654(C>T) double mutation Reference Standard	CBPD0007	Codon 71/72(+A)	50%	NM_000518.5
		IVS-II-654(C>T)	50%	NM_000518.5
β-thalassemia Codon 27(GAG>AAG)&IVS-I-1 (G>T) double mutation Reference Standard	CBPD0008	Codon 26(GAG>AAG)	50%	NM_000518.5
		IVS-I-1 (G>T)	50%	NM_000518.5
β-thalassemia IVS-I-1(G>A)&IVS-I-6(T>C) double mutation Reference Standard	CBPD0009	IVS-I-1(G>A)	50%	NM_000518.5
		IVS-I-6(T>C)	50%	NM_000518.5
β-thalassemia IVS-I-6(T>C)&IVS-II-745(C>G) double mutation Reference Standard	CBPD0010	IVS-I-6(T>C)	50%	NM_000518.5
		IVS-II-745(C>G)	50%	NM_000518.5
α-thalassemia αα/--SEA Reference Standard	CBPD0029	chr16:215396-234700 del	50%	N/A

Product advantages

For **α-Thalassemia samples**, a combined probe-anchored polymerization sequencing method was utilized to detect genetic variations. This method was validated using Sanger sequencing, ensuring the accuracy and reliability of the results.

For **β-Thalassemia samples**, Sanger sequencing alone was employed as the primary method of analysis due to its high precision in detecting point mutations and small insertions/deletions.

All the above samples are derived from natural sources and closely mimic clinical conditions, ensuring their relevance and applicability for diagnostic and research purposes.



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