

## Using Genetic Testing for Diagnosis of Beta-Thalassemia

### Indications:

- Clinical or hematological diagnosis of beta thalassemia
- Known familial mutations in *HBB*

### Benefits:

Genetic testing for beta thalassemia

- Can confirm a suspected clinical or hematological diagnosis of beta thalassemia and identify the familial mutation(s)
- Can help to distinguish between beta thalassemia major and intermedia
- Can identify presymptomatic family members of patients and inform the prognosis, facilitating selection and timely initiation of the most effective treatment
- Can detect carrier state in family members of patients and assist in genetic counseling .

### Background:

- Beta thalassemia is an inherited form of severe anemia, with an estimated prevalence of 1:100,000 worldwide and 1:10,000 in the European Union, reflecting the increased prevalence in Mediterranean populations.<sup>1,2</sup>
- Mode of inheritance is typically recessive, but dominant cases have also been reported.<sup>1,2</sup>  
Based on disease severity, two types of symptomatic beta thalassemia are distinguished: beta thalassemia major (also known as Cooley's anemia) and beta thalassemia intermedia.<sup>1,2</sup>
- Beta thalassemia major is characterized by infancy-onset severe anemia and requires life-long blood transfusions for survival. By definition, the intermediate form requires only intermittent blood transfusions for survival.<sup>1,2</sup>
- Distinction between the major and intermediate forms is important for treatment decisions. While unnecessary transfusions should be avoided, . delaying the initiation of regular transfusions can increase the risk that a patient may develop multiple antibodies against donor red blood cells.<sup>1</sup>
- Bone marrow or chord blood transplantation offers a cure, especially if performed before lasting organ damage has developed.<sup>2</sup>
- Severity of beta thalassemia and mode of inheritance can partially be predicted from the nature of the causative mutations in *HBB*, the gene coding for beta globin.<sup>1-3</sup>

**References:** 1. Galanello R and Origa R (2010) Orphanet Journal of Rare Diseases 5:11. 2. Cao A and Galanello R. GeneReviews. <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=b-thal> Accessed 08/01/10 3. Thein SW (2005) Genetic modifiers of  $\beta$ -thalassemia. Haematologica 90:649-60.

**Ordering Information:** Please see other side.



## Ordering Information for Beta-Thalassemia Testing

### Indications for Testing

- Clinical or hematological diagnosis of beta thalassemia
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### Ordering Information

Gene(s)	Test Code
<i>HBB</i>	252823

### Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

### Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

**For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at [www.correlagen.com](http://www.correlagen.com).**