GENE Seeker

You know the risk of your child being a carrier of a genetic disease.

Beta thalassaemia

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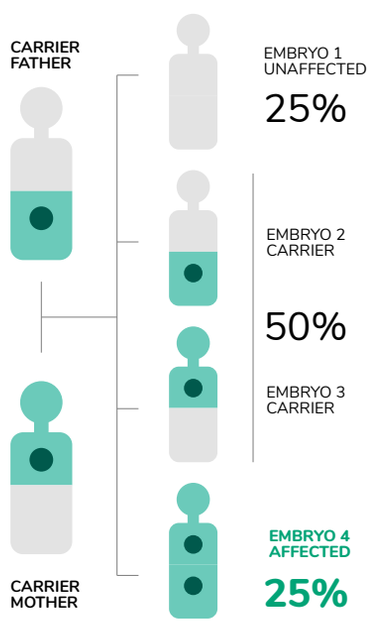
What is Beta thalassaemia?

Beta-thalassemia (BT) major is a severe early-onset form of BT (see this term) characterized by severe anemia requiring regular red blood cell transfusions.

Onset is during infancy with severe anemia, failure to thrive and progressive pallor. Feeding problems, diarrhea, irritability, recurrent bouts of fever, and progressive enlargement of the abdomen caused by splenomegaly and hepatomegaly may occur. Untreated or poorly transfused patients show growth retardation, pallor, jaundice, poor musculature, genu valgum, leg ulcers, formation of masses due to extramedullary hematopoiesis, and skeletal changes including deformities in the long bones of the legs and typical craniofacial changes such as bossing of the skull, prominent malar eminence, depression of the bridge of the nose, tendency to a mongoloid slant of the eye, and maxillae hypertrophy, which tends to expose upper teeth. In regularly transfused patients, growth and development tend to be normal but complications related to iron overload may develop, including growth retardation and failure or delay of sexual maturation. Later-onset iron overload complications include dilated myocardiopathy, arrhythmias, liver fibrosis and cirrhosis, diabetes mellitus, and insufficiency of the parathyroid, thyroid, pituitary, and, less commonly, adrenal glands. Other complications are hypersplenism, venous thrombosis and osteoporosis.



What is the next step if I'm a carrier of Beta thalassaemia?

If you are found to be a carrier of Beta thalassaemia, it is important that your partner be tested for the same genetic disorder.

What if my partner is not a carrier?

If your partner's test for Beta thalassaemiais negative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Beta thalassaemia?

When both parents are carriers of Beta thalassaemia, the probability of having a child with Beta thalassaemia is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.

If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

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