GENE Seeker

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

Haemoglobinopathies

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What is Haemoglobinopathies?

A rare hemoglobinopathy characterized by the presence of hemoglobin variants with structural abnormalities in the globin portion of the molecule which lead to auto-oxidation of heme iron, resulting in methemoglobinemia. Patients present with cyanosis for which no treatment is necessary. Mode of inheritance is autosomal dominant.

Methemoglobinemia is a clinical condition in which more than 1% of hemoglobin is oxidized to methemoglobin, a type of hemoglobin that contains the ferric (Fe3+) form of iron. Patients with hemoglobin M are cyanotic but otherwise asymptomatic. If the mutation occurs in the hemoglobin alpha subunit, cyanosis is apparent at birth, whereas if the beta chain is affected, cyanosis appears later or intensifies when beta subunit production increases. If a newborn carries a fetal M hemoglobin (gamma subunit), cyanosis disappears when the complete gamma-beta-switch occurs.

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

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

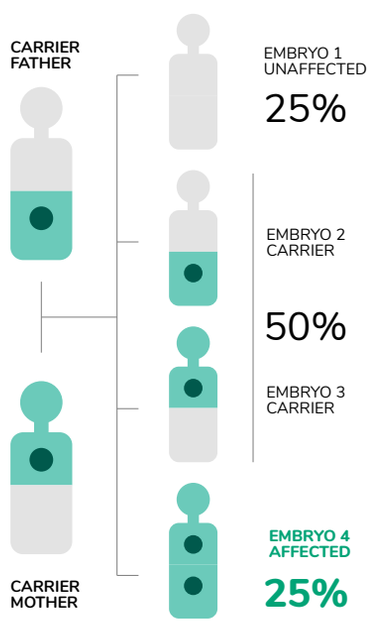
What if my partner is not a carrier?

If your partner's test for Haemoglobinopathies is 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 Haemoglobinopathies?

When both parents are carriers of Haemoglobinopathies , the probability of having a child with Haemoglobinopathies 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.

1 frequency

50 people

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