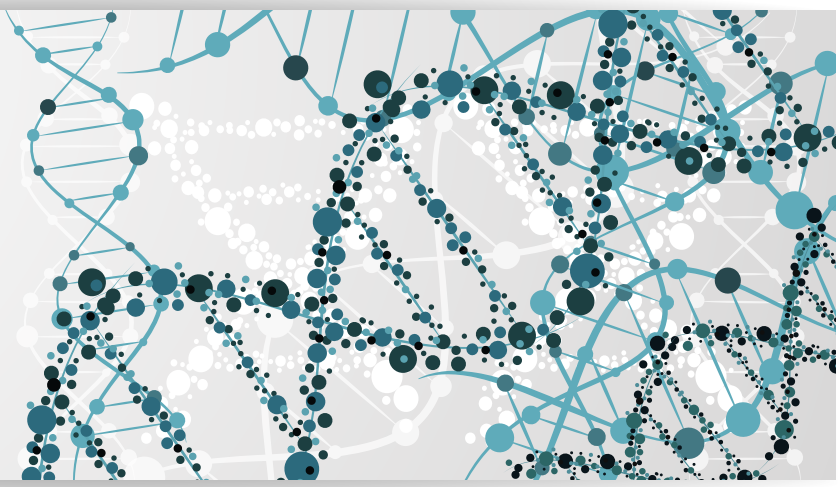


# GENE Seeker

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



## Alpha thalassaemia

[junogenetics.com](http://junogenetics.com)

### What is Alpha thalassaemia?

A rare developmental defect during embryogenesis, a contiguous gene deletion syndrome, is a form of alpha-thalassemia characterized by microcytosis, hypochromia, normal hemoglobin (Hb) level or mild anemia, associated with developmental abnormalities.

ATR-16 is a congenital disease. Patients present with either alpha-thalassemia trait or mild hemoglobin H disease (HbH disease) associated with a mild to profound (in most cases) intellectual disability and, in some cases, with mild, nonspecific dysmorphic features (mild hypertelorism, down slanted palpebral fissures, broad or prominent nasal bridge, small ears, short neck), microcephaly and short stature. Genital abnormalities (hypospadias and cryptorchidism) have been reported in males. Club foot is common.

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

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

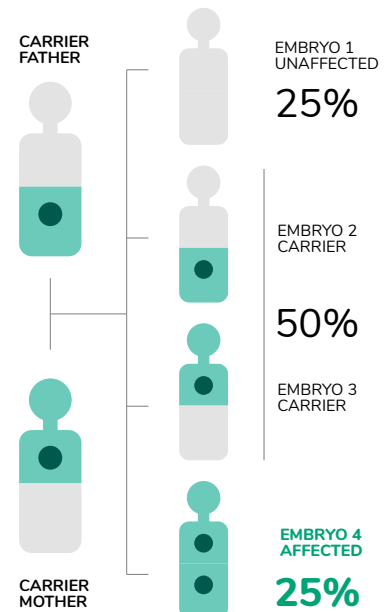
### What if my partner is not a carrier?

If your partner's test for Alpha thalassaemia 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 Alpha thalassaemia?

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

