GENE Seeker

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

Spinal muscular atrophy

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What is Spinal muscular atrophy?

A rare, genetic proximal spinal muscular atrophy characterized by degeneration of alpha motor neurons in the anterior horns of the spinal cord and lower brain stem manifesting with onset of severe and progressive muscle weakness in the first 6 months of life and presenting with severe, generalized hypotonia and weakness,. Dysphagia and respiratory impairment may also be present at presentation or appear at a later stage. Classically, before the advent of recent therapies, type 1 patients never achieved sitting without support.

Disease onset occurs before 6 months of age. The severe, symmetrical muscle weakness affects predominantly proximal limbs but often also involves the extremities. Cries are weak. Poor sucking ability and reduced swallowing are frequent, leading to feeding difficulties. Deep tendon reflexes are absent. Patients have paradoxical breathing, a bell shaped chest and develop respiratory failure. Mild contractures (of the knees and, more rarely, of the elbows), and scoliosis may be present. Classically, patients were not able to achieve sitting without support but this has changed following the availability of new treatments.

What is the next step if I'm a carrier of Spinal muscular atrophy?

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

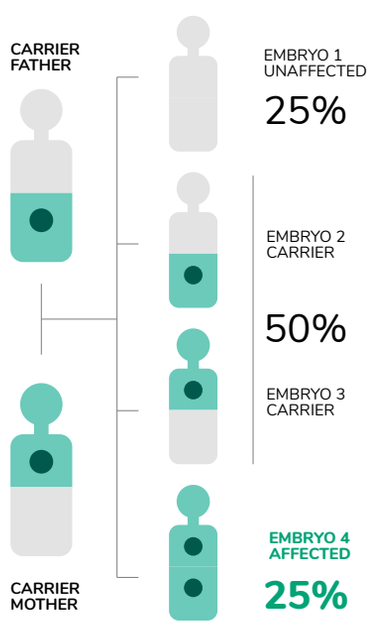
What if my partner is not a carrier?

If your partner's test for Spinal muscular atrophy 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 Spinal muscular atrophy?

When both parents are carriers of Spinal muscular atrophy, the probability of having a child with Spinal muscular atrophy 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

45 people

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