Rigshospitalet

Information for parents to a new-born child, who has been tested for cystic fibrosis and carries a mutation for cystic fibrosis

This letter informs you about the result of the analysis for cystic fibrosis in the heel prick test that was carried out shortly after your child was born. The test showed that:

Your **child does not have cystic fibrosis** and **will not develop it**.

Result of the heel prick test for cystic fibrosis
All new-borns in Denmark are within 2-3 days of birth analysed for inherited diseases through the so-called heel prick test. Your child was among other diseases analysed for cystic fibrosis. This is an inherited disease that causes lung and nutrition problems.

The test showed that your child is a carrier of a single mutation for cystic fibrosis
This means that your child does NOT have cystic fibrosis, but that it carries a single mutation for cystic fibrosis and can pass this on to his or her own child.

How is cystic fibrosis inherited?
Cystic fibrosis is a genetic disease. A child can only develop the disease if it has two mutations for cystic fibrosis. Your child has only inherited one mutation from you, and therefore neither you nor your child has cystic fibrosis.

Carriers of a mutation for cystic fibrosis
To be a carrier of a mutation for cystic fibrosis has no health significance. Your child does not have cystic fibrosis and will not need treatment, but your child carries a mutation for cystic fibrosis. This is called being a healthy carrier of a mutation for cystic fibrosis.

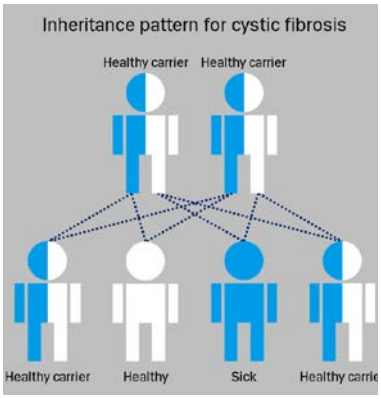
You need to be aware of:
The main reasons for informing you about your child being a carrier of a mutation for cystic fibrosis are that you need to be aware of:

Facts on cystic fibrosis

Cystic fibrosis, often referred to as CF, is an inherited disease primarily affecting the lungs.

In Denmark around 3 % of the population, or 1 in 34, are carriers of a mutation for cystic fibrosis.

The child is only at risk for inheriting the disease if both of the parents are carriers of a mutation for cystic fibrosis.



1. **Increased risk in future pregnancies:** If you wish to get pregnant again it is important that you know that the risk of getting a child with cystic fibrosis is slightly increased compared to other parents.
2. **Increased risk for your child getting children with cystic fibrosis:** Your child could pass on their mutation to their own children. Your child should therefore, before considering getting pregnant, be informed that he or she is a carrier.
3. **Increased risk for your family:** When your child is a carrier of cystic fibrosis, other members of your family have an increased risk of being carriers as well. Thus, they have an increased risk of getting a child with cystic fibrosis. We encourage you to inform your family members of this.

Genetic counseling for carriers of a mutation for cystic fibrosis
Should you or your family members have any questions to the above-mentioned or if you wish additional analyses to clarify who of you carries the mutation, we encourage you to get referred to the local department of clinical genetics through your own doctor. This is especially important if you wish to get pregnant again.

Sincerely

Department of Clinical Genetics, Copenhagen University Hospital, Rigshospitalet

A Danish version of this letter is available on the webpages for the Department of Clinical Genetics. www.rigshospitalet.dk/kgk