

20. juli 2016

## **Information for parents to a new-born child, who is healthy but carries a mutation for cystic fibrosis**

This letter informs you about the result of the analysis for cystic fibrosis in the heel prick test.

### **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 carries a single mutation for cystic fibrosis. This means that:

**Your child is healthy and your child will not develop 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, but carries a mutation for cystic fibrosis. There is no need for further evaluation.

### **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. Because your child has only one mutation she or he does NOT have cystic fibrosis, - just like the parent, from whom the child has inherited the mutation, does not have cystic fibrosis. Among all Danes, approximately 1 out of 30 people will be a healthy carrier of a mutation for cystic fibrosis.

### **What does it mean to be a carrier of a mutation for cystic fibrosis?**

The main reasons for informing you that your child is a carrier of a mutation for cystic fibrosis is:

- 1. Increased risk in future pregnancies:** The mutation must be inherited from one of the parents. If you wish to get pregnant again it is important that you know that the risk of getting a child with cystic fibrosis is increased compared to other parents. The risk for other parents is 1 out of 4000 pregnancies, while your risk is 1 out of 250 pregnancies.
- 2. Increased risk for your family:** If you are 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 of this: In other words tell your family that you have discovered that your child is a carrier of a mutation for cystic fibrosis and that one of you therefore is a carrier as well.
- 3. Increased risk for your child getting children with cystic fibrosis:** Your child could pass on their mutation to their own children. Your child should before considering getting children be told that he or she is a carrier.

### **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