Am I a carrier for cystic fibrosis?

Who is at risk?

Millions of people carry a mutated CF gene, but do not have any symptoms[3]. About 1,000 new CF cases are diagnosed each year[1]. Historically, CF has been associated with those of European descent, but this is changing as the diversity of the global population increases. The risk of carrying the abnormal gene by ethnicity[4] is as follows:

- Ashkenazi Jewish: 1 in 25
- Mixed European: 1 in 25
- Eastern European: 1 in 25
- French Canadian: 1 in 25
- Northern European: 1 in 25
- Southern European: 1 in 25
- Hispanic: 1 in 46
- African-American: 1 in 65
- Asian-American: 1 in 90

About cystic fibrosis.

Cystic fibrosis (CF) is a life-threatening inherited chronic disease that affects multiple organs, including the lungs and digestive system. It is the most common genetic disorder of the Western world, with an estimated 70,000 children and adults worldwide having the disease[1]. CF significantly shortens the lifespan of people affected by it—median survival is approximately 37 years[2].

CF is a recessive genetic disease. In order to have CF, a person must inherit two mutated copies of the CF transmembrane conductance regulator (*CFTR*) gene. Individuals that have a single mutated *CFTR* gene are called "carriers." They are often asymptomatic and unaware that they carry a defective gene.

Why should I know my carrier status?

Although there are novel therapies for CF, there is no known cure. Even if you do not have a family history of the disease, you may still carry a mutated *CFTR* gene that can be passed on to your children. Ask your doctor about being screened for your CF carrier status. This is the only way to truly understand your risk of having a child with the disease.

I am a carrier. Now what?

The earlier you know your CF carrier status, the more options you may have for healthy family planning. If you and your partner test positive as CF carriers, you can look at options such as *in vitro* fertilization (IVF) and preimplantation genetic diagnosis (PGD) to improve the chances of having a healthy child.

If you know that you or your partner are CF carriers, make sure that your infant is screened for CF. Early diagnosis of CF and a comprehensive treatment plan has been shown to improve both survival and quality of life[3].

Illumina and CF.

Illumina is dedicated to providing solutions that advance the understanding of genetics and health in an effort to improve human wellness. CF is a common genetic disorder where screening and early diagnosis can make a difference. Illumina technology is helping healthcare professionals and patients to analyze the *CFTR* gene in a more comprehensive molecular manner, enabling therapeutic decisions that will lead to a better quality of life.

References:

1. Cystic Fibrosis Foundation (www.cff.org/AboutCF/Faqs/) 26 March 2013.
2. The American Congress of Obstetricians and Gynecologists (2011) Update on carrier screening for cystic fibrosis. Committee Opinion No. 486. 21 March 2011.
3. U.S. National Library of Medicine. PubMed Health (http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001167) 27 March 2013.
4. March of Dimes (http://www.marchofdimes.com/pregnancy/prenatalcare_cysticfibrosis.html) 17 April 2013.

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