PATERNITY For Life
DNA TESTING SPECIALISTS

Focusing on
Personalised Medicine

CYSTIC FIBROSIS SCREENING

In the UK, around 10,400 people have Cystic Fibrosis (CF); that's roughly 1 in every 2,500 babies. CF affects around 100,000 people in the world. On average 1 in 25 people carry the CF gene - most of whom are unaware that they are carriers.

What is Cystic Fibrosis?

Cystic fibrosis (CF), also known as mucoviscidosis, is an inherited disorder caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. The CFTR gene helps to regulate the flow of salt and water into and out of cells in our body. It plays an important role in the production of sputum (mucus), sweat and other digestive fluids. Normally, these secreted fluids are thin and slippery. However, for people with CF the defective gene causes the secretions to become thick and sticky. As a result, the secretions clog up tubes, ducts and passageways, particularly in the lung and digestive system.

The lungs make thicker sputum than normal, which can trap bacteria in the small airways and lead to infection. Symptoms that typically develop include persistent cough, wheezing, shortness of breath and breathing difficulties and repeated chest infections.

CF can also affect other parts of the body, including the liver, nose, sinuses and sweat glands. Men with CF are usually infertile, due to absence or blockages of the vas deferens, which are ducts that carry sperm.

Who is at risk?

In the UK, one in 2,500 babies are born with CF - approximately one every four days. On average 1 in 25 people carry a defective CF gene - most of whom are unaware that they are carriers. Because carriers of CF are unaffected and show no symptoms, they may not be aware that CF may be a real risk. That's approximately 1 million unaware carriers.

Carrier Testing

If have a history of CF in your family, a partner with CF (or a family history of it), or a child with the condition, you may choose to get carrier testing. A simple mouth swab can determine if you are a carrier of the faulty gene that causes cystic fibrosis. Carrier testing is often done for people who are thinking of starting a family and have a relative with cystic fibrosis

If someone carries the faulty gene that causes CF, will their child have the condition?

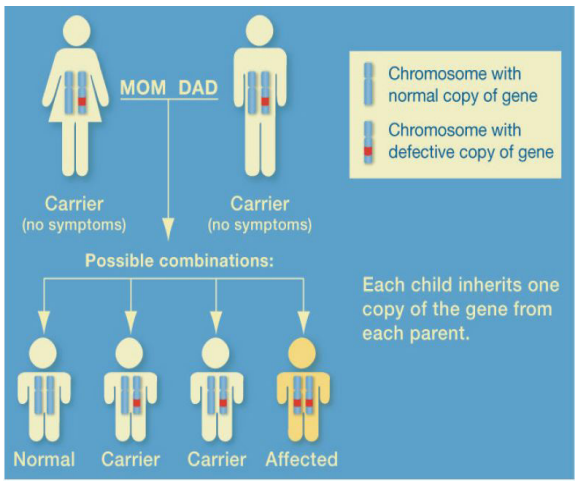
Cystic fibrosis is a recessive disorder, which means that both parents must pass on the defective gene for any of their children to get the disease. If a child inherits only one copy of the faulty gene, he or she will be a carrier.

People who have one normal CFTR gene and one faulty CFTR gene are CF carriers. CF carriers usually have no symptoms of CF and live normal lives. However, carriers can pass the faulty CFTR gene on to their children.

If both parents are carriers of the faulty gene, but don't have cystic fibrosis, a child has:

- a 25% chance of being born with cystic fibrosis,
- a 50% chance of being a carrier, like their parents, but not having the condition, and
- a 25% chance of being completely free of the condition – neither having CF nor being a carrier of the faulty gene.

Where an infant receives the CF gene from just one parent, he/she will not be born with CF, but will throughout life, be a symptomless carrier of the CFTR gene like his/her parents.



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How is the CFTR Test performed at Paternity For Life?

CFTR is the only gene known to be associated with CF. Over 1600 mutations occur in the CFTR gene; almost all are point mutations or small (1–84 bp) deletions where small pieces of DNA are missing. The most common mutation, ΔF508, is a deletion (Δ signifying deletion) of three nucleotides that results in a loss of the amino acid phenylalanine (F) at the 508th position on the protein.

A sample is collected using a simple and painless mouth swab. DNA is then extracted from the sample and screened for specific defects in the CFTR gene. Our testing is performed using massive parallel sequencing (next generation sequencing) which allows for the analysis of exons, intron-exon boundaries, and UTRs that contain common mutations in the CFTR gene. This methodology with sequence analysis of all exons, intron/exon borders, promoter regions, and specific intronic regions detects more than 98% of CFTR mutations.

How will I receive my results?

Your results will be sent directly to your nominated health professional, who will be able to discuss your results with you. The results will state whether the person tested has been found to be a carrier. It must be noted that a negative result does not absolutely rule out the possibility of being a carrier. Details of the chances of still being a carrier are given to people who test negative, and a statistical level of the risk of having a child with CF is given to couples.

How do I organise testing?

Paternity For Life aims to educate patients and their families about inherited diseases and their risk of inheritance, to empower them with the knowledge to take control of their health and treatment plans. As inherited disease testing and the results from such tests can be complex, we also recommend you seek Genetic Counselling both prior to and after testing to help you understand your results and the impacts they may have for both you and your family.

If you are ready to proceed with testing, a Testing Request Form can be downloaded from our website. We recommend you book an appointment with your doctor to discuss the testing and have your Request Form signed. You can then head to our website to submit your order and payment. Paternity For Life will then send a testing kit directly to your home address. All that is required for testing is a simple, painless mouth swab. Once we receive your sample we will begin testing. Results are typically available to your doctor within 21 working days.

Download Request Form and submit your order to Paternity For Life

Receive testing kit from Paternity for Life

Paternity For Life Receives sample for testing

Paternity For Life completes requested testing

Results sent to nominated health care professional

Email: admin@paternityforlife.co.uk | Web: www.paternityforlife.co.uk