Cystic Fibrosis our focus

Cystic fibrosis: diagnosis in adulthood

Factsheet – July 2016

Fighting for a Life Unlimited

Cystic fibrosis: diagnosis in adulthood

The information in this factsheet has been written for adults who have recently been diagnosed with cystic fibrosis.

Finding out that you have cystic fibrosis (CF) is bound to come as a shock. On the other hand, it can be a relief to know what is wrong and to discover that there is so much available in the way of treatment and support.

This factsheet will give you some idea of what to expect and where to go for advice and support.

Contents

What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited condition that affects vital organs in the body, particularly the lungs and pancreas, by clogging them with thick, sticky mucus. The mucus makes it difficult for people with CF to breathe and digest food properly, which makes them prone to chest infections and digestive problems.

Cystic fibrosis is an unpredictable disease and every person with the condition is likely to be affected differently. This is important to remember as you begin to find out more about the condition and what it might mean for you.

Why me?

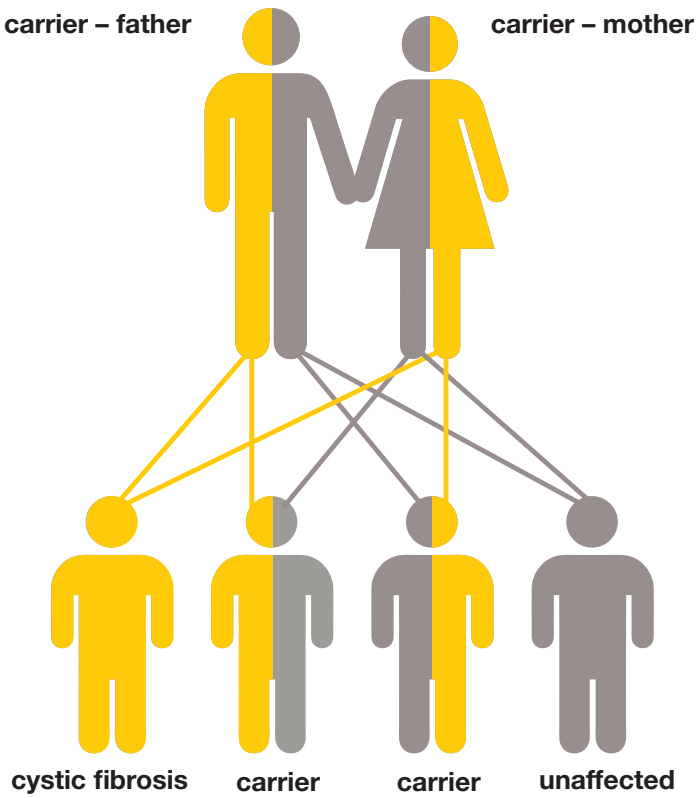
Cystic fibrosis is one of the UK's most common life-shortening inherited conditions. More than 10,500 people in the UK have the condition, which is caused by a genetic fault that is inherited from both parents. As with other genetic conditions, cystic fibrosis will have been present since birth, even if it is diagnosed later in life.

One in 25 people carry the faulty gene that causes cystic fibrosis. To have cystic fibrosis, both parents must be carriers of the faulty cystic fibrosis gene. Even if both parents have the faulty gene, it doesn't automatically mean that every baby they have will have the condition.

If both parents are carriers (eg each has only one copy of the faulty gene, rather than two), a child has:

- a one in four chance of being born with two faulty genes and therefore having cystic fibrosis;
- a two in four chance of being a carrier but not having the condition; and
- a one in four chance of being completely free of it, i.e. not having CF nor being a carrier of the faulty CF gene.

The chances are the same for each pregnancy.



Why wasn't it picked up earlier?

There are several reasons that could explain why you were not diagnosed earlier.

The gene responsible for cystic fibrosis was not identified until l989, and until that time tests used to confirm diagnosis in adults were not so reliable.

The faulty gene that causes CF can be faulty in many different ways. In fact there are now known to be more than 2,000 mutations (faults) that cause cystic fibrosis. The blood tests for CF do not test for all these mutations. You may have one of the rarer gene mutations, which is harder to diagnose.

Because of the many different mutations that occur in the CF gene, cystic fibrosis can vary widely in its severity and symptoms. This can sometimes make it difficult to diagnose, and it can mimic other lung diseases such as asthma or bronchitis.

Since October 2007, newborn screening for cystic fibrosis has been available for all newborn babies in the UK. A sample of blood is taken when babies are around five days old, as part of the routine heel prick test. These spots of blood are tested in the laboratory for signs of several medical conditions, including cystic fibrosis. The test identifies where the baby has a high chance of having CF; other tests are then carried out to confirm diagnosis. This has meant diagnosis can be made at an earlier stage.

What is the treatment?

There is no cure yet for CF, but the prognosis continues to improve.

Meanwhile, a daily regimen of treatment helps people with CF to control their symptoms and to lead fulfilling lives. Depending on your individual situation, treatment may include:

- physiotherapy at least once a day to help clear the lungs,
- high-energy foods and enzyme supplements to aid digestion, and
- antibiotics to fight infections.

It's important that, following diagnosis, you start treatment as soon as possible. Not only should it make you feel better, but it will improve the quality of your life in the long term.

New and ever-more effective techniques are being developed to treat the condition, and your specialist CF centre should be able to advise you.

Most people respond well to treatment for many years and if, eventually, conventional methods fail to be effective, some may go forward for lung transplantation.

Where should I be treated?

There are specialist CF centres in hospitals throughout the UK. These centres have multidisciplinary teams of expert clinicians, including doctors, nurses, physiotherapists, dietitians, psychologists and social workers, who specialise in the treatment of cystic fibrosis.

Research shows that these centres offer the best treatment for cystic fibrosis. We recommend that you ask your local hospital or GP to refer you to the CF centre nearest your home. If you need more information about CF centres, please contact the Cystic Fibrosis Trust helpline on 0300 373 1000 or at helpline@cysticfibrosis.org.uk.

What is the outlook?

Life expectancy for people with CF has increased dramatically over recent decades. With modern treatments, nearly all children with CF survive into adulthood and in the UK there are now more adults than children with the condition. Half of those living with cystic fibrosis will survive into their 40s, and there are many people living with cystic fibrosis in their 50s, 60s and older.

Since the faulty gene was discovered in 1989, research has come a long way. Research is taking place across the world to develop new, more effective treatments for cystic fibrosis, and there are a number of promising treatments in development. For more information about the research that the Cystic Fibrosis Trust funds please look at our website at cysticfibrosis.org.uk/research.

Coming to terms with living with cystic fibrosis

Coming to terms with the diagnosis of cystic fibrosis can be difficult, not just for the person diagnosed but also for partners, families and friends.

From a practical point of view, many people who have been diagnosed later in life find it challenging to incorporate the regimen of medical care into everyday life. It's not easy, but it is important to maintain the treatment regimen and to discuss any problems with your doctor, or other members of the CF team.

If you have been recently diagnosed, you might have very mixed emotions about your future. Naturally, you may be worried about the impact that CF might have on you, your family, your job, your studies, your social life, your friends; indeed, every aspect of your life. Some people initially try to deny the reality, which is a common reaction particularly if the symptoms are, or have been, mild.

Coming to terms with CF can be difficult and consequently some people experience problems with sleeping, lack of concentration, loss of appetite or even suffer with depression.

Alternatively you might feel angry with medical professionals or with people close to you. These emotions are all natural reactions to the diagnosis of an illness like cystic fibrosis. In time these feelings are likely to subside. Some people find that the process of coming to terms with a diagnosis of CF is helped by talking to a psychologist or social worker from the CF team.

You can also contact the Cystic Fibrosis Trust helpline on 0300 373 1000 or helpline@cysticfibrosis.org.uk, whether you have specific questions or would just like to talk to someone about the feelings you may be experiencing. Another option is the Cystic Fibrosis Trust forum, which is an online community where people affected by CF can share their thoughts and experiences.

You can find the forum at cysticfibrosis.org.uk/forum.

Relationships, sex and fertility

A diagnosis of cystic fibrosis shouldn't stop you developing new, or continuing with existing, relationships. However, the condition can impact on family planning.

Women with CF may be able to have children; it is thought that CF could reduce a women's fertility but this is not always the case. Therefore, it's advisable to use contraception unless you are planning a pregnancy. Pregnancy and motherhood can have health implications for a woman with cystic fibrosis, so it's important to consult with your CF team to ensure you're in the best possible health prior to trying for a baby.

Men with CF can enjoy a normal sex life, but are likely to be infertile because the tubes from the testes are either absent or blocked by mucus. Infertility can be one of the symptoms that contributes to a diagnosis of cystic fibrosis in adulthood. However, it is still possible to father a child using fertility treatments.

If you are planning a family, it is advisable to consider carrier testing for your partner and genetic counselling prior to pregnancy. If your partner is also a carrier of the CF gene this means there is a 50% chance your baby would be born with cystic fibrosis. Genetic testing is a personal decision; for some the outcome of carrier testing will inform their family plans whereas for others the outcome is not relevant.

What do I tell my family and friends?

It is up to you. If you have a good relationship with your family and friends, it can be helpful to discuss your diagnosis; they could be a good source of support. They may be concerned about you, or want to help, but not know how. As you know, CF is a genetic condition, which means there might be implications for others in your family. It can be helpful to show friends and family literature on cystic fibrosis, or direct them to the Cystic Fibrosis Trust website.

Some people like to involve those closest to them in all aspects of their care; others prefer to keep their treatment private; ultimately it's a personal choice.

If you have children, it's important that you tell them about cystic fibrosis. The way you approach this will obviously depend on the age of your child and the extent of your symptoms. Your CF team might be able to support you with this or you can contact the Cystic Fibrosis Trust helpline.

Further education and employment

Your diagnosis should not stop your education or employment plans.

If you are in, or are planning to start, further education it can be helpful to be open about your condition so that you can discuss the best way to fulfil your course requirements whilst maintaining your treatment regimen. If you are looking at different colleges and universities away from your home, it's worth considering where the nearest CF centre would be. The Cystic Fibrosis Trust produces a factsheet on further education that you might find helpful.

Many adults with CF are employed and have managed to fit their treatment regimen around their work. If you discuss your diagnosis with your employer and explain how your CF affects your life and work, you may be able to negotiate different working practices (or reduced hours) to fit in with your circumstances.

It is against the law for an employer to discriminate against someone because of a disability such as cystic fibrosis. For the purposes of the legislation, disability refers to is a physical or mental condition that has a long-term and substantial effect on your daily life. For more information about your employment rights you can contact our helpline on 0300 373 1000 or at helpline@cysticfibrosis.org.uk. Citizens Advice also have informative pages about your rights at work, please visit www.citizensadvice.org.uk/discrimination.

General lifestyle

The fact that your symptoms have been given a name should not necessarily affect your social life and general lifestyle, although you may have to make some changes to accommodate your daily treatment regimen. You need to be more aware of, and attempt to avoid, the risk of chest infections and particularly the risk of cross-infection from other people with cystic fibrosis. People with CF can carry bugs in their lungs that are not harmful to those without the condition but can be very harmful to others with cystic fibrosis. Your CF clinic will be managed in such a way that you will never be in the same area as another CF patient. It's important for your health, and the health of others with CF, that you don't mix with other people with the condition. The Cystic Fibrosis Trust forum (www.cysticfibrosis.org.uk/forum) is an ideal online community where you can chat and share experiences without any risk to your health.

There are some environmental risks that you should also be aware of. Cigarette smoke is harmful to your lungs and so smoky places should be avoided. There are some environments (including rotting vegetation, mud and stagnant water) that harbour bugs, such as bacteria, fungi or viruses, which may be harmful to the lungs of people with cystic fibrosis. Your CF team will be able to advise you further on infection risks.

What support is available?

You are not alone; there are over 5,000 adults with cystic fibrosis in the UK. The Cystic Fibrosis Trust Support Service is there to help you by providing practical, emotional and financial support. Our helpline is available Monday to Friday from 9am to 5pm, and provides advice about cystic fibrosis, or just a listening ear. Our services also include financial support, through health and wellbeing grants, and smaller emergency grants. We also offer advice on benefits, welfare and rights for people with cystic fibrosis and their families. Our CF Connect scheme also allows people affected by cystic fibrosis to gain support, over the telephone, from others who have been through similar experiences.

For further information about the support we can offer, please contact our helpline on 0300 373 1000 or helpline@cysticfibrosis.org.uk.

Further information

The Cystic Fibrosis Trust provides information about cystic fibrosis through our factsheets, leaflets and other publications.

Most of our publications are available through our helpline and can be downloaded from our website or ordered using our online publications order form. Visit cysticfibrosis.org.uk/publications.

The Cystic Fibrosis Trust helpline can help you with a range of issues, no matter how big or small. Our trained staff can provide a listening ear, practical advice, welfare/benefits information or direct you to other sources of support. The helpline can be contacted on 0300 373 1000 or helpline@cysticfibrosis.org.uk and is open Monday to Friday, 9am – 5pm.

Calls to 0300 numbers cost no more than 5p per minute from a standard BT residential landline. Charges from other landlines and mobile networks may vary, but will be no more than a standard geographic call and are included in all inclusive minutes and discount schemes. If you are worried about the cost of the call please let us know and we'll call you back.

You can also find more information at our website cysticfibrosis.org.uk.

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More factsheets available at:
cysticfibrosis.org.uk/publications