

# **Cystic Fibrosis** *our focus*

## **Cystic fibrosis: late diagnosis**

Factsheet – August 2020

# Cystic fibrosis: late diagnosis

This factsheet is for people who were not diagnosed with cystic fibrosis (CF) as a baby or young child, but when they were older.

Finding out that you have CF is bound to come as a shock. You may find that a diagnosis of CF makes you feel differently about yourself, and that these feelings are confusing. 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. It also features quotes from other people who were not diagnosed until they were older.

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## What is cystic fibrosis?

Cystic fibrosis is an inherited condition that affects lots of different parts of the body. Most people with the condition experience a build-up of thick, sticky mucus in the lungs, digestive system and other organs.

In the lungs this can make breathing difficult and can lead to lung infections. In the digestive system this can affect how well the body absorbs nutrients and can cause digestive problems.

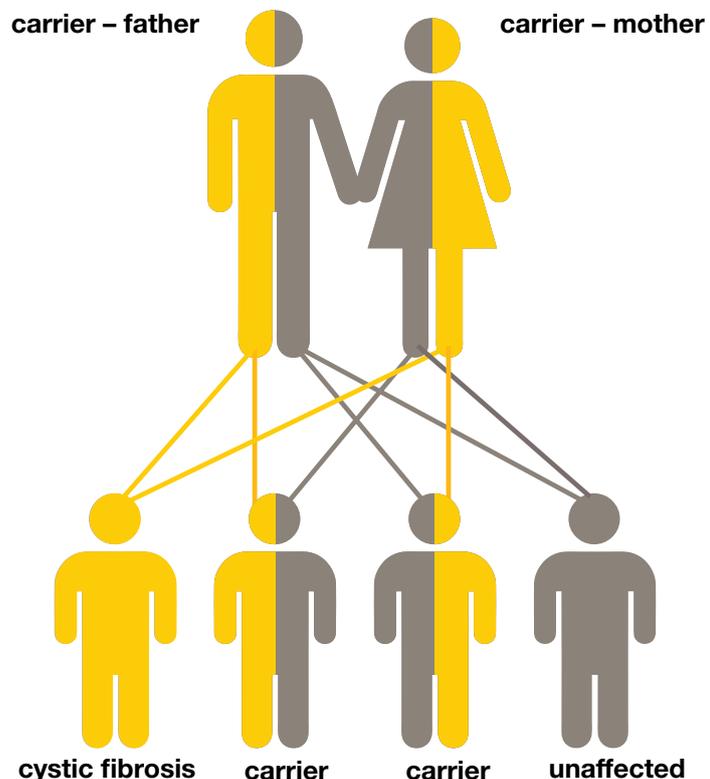
Cystic fibrosis varies from person to person and everyone with the condition is likely to be affected differently. This is important to remember as you begin to find out more about the condition and what it might mean for you.

**“I had heard of CF a long time ago when I was in secondary school. I remember learning about it in Biology class when we were studying genetics. I also recall watching a documentary about children with CF and how intense their treatments were.”** – Michelle, diagnosed aged 27

## Why me?

Cystic fibrosis is one of the UK's most common genetic conditions. More than 10,500 people in the UK have the condition, which is caused by a faulty gene that is inherited from both parents. As with other genetic conditions, you are born with CF, even if you are not diagnosed until you're older.

One in 25 people carry the faulty gene that causes cystic fibrosis. For a baby to have CF, both parents must have one copy of this gene, making them 'carriers'. However, even if both parents are carriers, this doesn't mean that every baby they have will have the condition.



If both parents are carriers (eg each parent 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 CF; and
- a one in four chance of not being a carrier and not having cystic fibrosis.

The chances are the same for each pregnancy.

**“I knew about CF as I had a brother who sadly died in 1959 aged seven, which was a good age for back then. It was openly discussed in the family if the subject ever arose, so I always knew about CF.”**

– Derek, diagnosed aged 30

## **Why wasn't it picked up earlier?**

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

The gene that causes CF was not identified until 1989. Until that time tests used to diagnose people with CF 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 tests for CF do not test for all these mutations, and you may have a rarer mutation that isn't tested for.

Because of the many different mutations, the symptoms of CF can be very different for different people. This can sometimes make it difficult to diagnose. It can even mimic other lung conditions such as asthma or bronchitis.

Since October 2007, newborn screening for CF 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 if the baby has a high chance of having cystic fibrosis. Other tests are then carried out to confirm a CF diagnosis or rule it out. The introduction of newborn screening for CF has meant that babies are being diagnosed earlier than ever before.

**“I always got told it was asthma and irritable bowel syndrome but I knew it was something more.”**

– Vicky, diagnosed aged 32

**“I had been referred to hospital to get a second opinion on care for an earlier diagnosis of Bronchiectasis. As part of the investigation to see why I had Bronchiectasis they checked for CF and the results showed I had it.”** – Michelle

**“After I learned more about the condition, I found it hard to process because what I was reading was not exactly what I was seeing in Michelle. I remember scouring the web to find information on milder cases of CF...”** – Shaun, Michelle’s husband

**“GPs and hospital doctors would tell my mother that I was faking my illness, that I was making things up to try and get attention. This despite the fact that my weight was dropping, and I would be off sick from school particularly during the winter terms.”**

– Shad, diagnosed aged 23

## **What is the treatment?**

There is no cure for cystic fibrosis, but there is a lot of research taking place to develop new treatments, and the outcomes for people with the condition continue to improve.

Daily treatments can help people with CF to control their symptoms. Depending on your situation, these might include:

- physiotherapy, inhalers and nebulisers to help clear the lungs,
- high-energy foods and enzyme supplements to help with digestion, and
- antibiotics to treat or prevent lung infections.

Treatments called ‘precision medicines’ that treat the underlying cause of CF have recently become available in the UK. However, they do not work for everyone with the condition. Your CF team should speak to you about the treatments available to you.

If you become particularly unwell, you may need to spend time in hospital on a ‘CF ward’. While you’re on the ward, you will stay in your own room and may be given antibiotics or other treatments through a drip in your arm. You’ll be able to have visits from friends and family, and bring your own clothes and things to keep you entertained while you’re there.

For some people, conventional treatments are not able to stop them from becoming very unwell, and they may need a lung transplant. This is an option that is not taken lightly. Your CF team will speak to you if they think that this is something you should be considering.

Once you are diagnosed, it’s important that 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.

Some people who are diagnosed later in life may find it difficult to complete their treatments. The burden these treatments places on people with CF and their families can be huge, so it is nothing to be ashamed of. Speak to your doctor or CF team if you are struggling, and they should be able to help.

**“It was hard managing my treatment as there is so much to do, but in time I have gotten into a routine and it is second nature.” – Vicky**

**“Whether it’s serving dinner with a side of Creon, entertaining our son whilst she nebulises, popping to the pharmacy to pick up a prescription or celebrating a high lung-function result, I try my best to support her.”**

– Shaun, Michelle’s husband

## **Where will I be treated?**

There are specialist CF centres in hospitals across the UK. These centres have teams of experts who specialise in the treatment of cystic fibrosis. This team can include doctors, nurses, physiotherapists, dietitians, psychologists and social workers.

We recommend that you ask your local hospital or GP to refer you to the CF centre nearest your home. If you have a question about your CF care, please contact the Cystic Fibrosis Trust Helpline on **0300 373 1000** or at [helpline@cysticfibrosis.org.uk](mailto:helpline@cysticfibrosis.org.uk).

## **What is the life expectancy for people with cystic fibrosis?**

Life expectancy for people with CF has increased dramatically over recent decades. In the UK there are now more adults than children with the condition, and half of those born with CF today are expected to live into their late 40s. However, this figure doesn’t account for advances in CF care, including new treatments that are being developed all the time.

Remember, any discussion of life expectancy won’t take into account your own personal circumstances, and it is important to focus on your own health.

Since the CF gene was discovered, research has come a long way. Today, research is taking place across the world to develop new treatments and to improve existing ones. For information about the research we fund, visit [cysticfibrosis.org.uk/research](http://cysticfibrosis.org.uk/research)

**“The consultant told me that they had recently diagnosed someone in their 70s with the condition and therefore it did not mean I was necessarily going to die soon.” – Michelle**

## Relationships, sex and fertility

A diagnosis of CF shouldn't affect your current relationships or stop you from starting new ones. It should also not impact your sex life. However, the condition can impact fertility.

Women with CF are often able to become pregnant, though there are some factors that might impact fertility. If you have CF, becoming pregnant can have an impact on your health. It's important to speak to your CF team if you are planning to have a baby.

Most men with CF will not be able to have a child without fertility treatments. This is because the tube that carries sperm from the testicles to the penis (called the vas deferens) is either missing or blocked.

Some people will find out that they have CF because they have a baby that is diagnosed with the condition. Infertility can also be one of the symptoms that causes someone to be diagnosed in adulthood.

Despite the potential impact of CF on fertility for men and women, it's important to use contraception if you don't wish to become pregnant.

If you are planning a family, you should consider carrier testing for your partner. If they are also a carrier of the CF gene this means there is a 50% chance of your baby being born with cystic fibrosis. Carrier testing is a personal decision. For some people the outcome of carrier testing will inform their family planning, whereas for others the outcome is not important.

For more information on fertility and cystic fibrosis, visit [cysticfibrosis.org.uk/fertility](https://cysticfibrosis.org.uk/fertility)

**“The consultant said most women with CF can have children, but we needed to see whether my partner carried the gene to check if there was a possibility our children could be born with CF.”** – Michelle

**“No one took me through what CF was at the very beginning. My only memory from the early conversations was a doctor telling me that I wouldn't be able to have children. I remember crying on the tube home.”** – Shad

## Coming to terms with living with cystic fibrosis

If you have been recently diagnosed, you might be experiencing very mixed emotions. You may be worrying about the impact that CF might have on your family and friends, job, studies and social life.

A common reaction to receiving a late diagnosis is to feel angry that you weren't diagnosed earlier. You may even not want to accept your diagnosis, particularly if the symptoms are mild or have been in the past.

Or, you might be relieved to have a diagnosis and be able to start treatment. Many people who are diagnosed later in life have experienced years of health problems. Some may have even been misdiagnosed with other conditions. For this reason, a diagnosis can also feel like a positive thing.

All of these emotions are normal, and in time they will probably become less intense. Some people find that talking to a CF psychologist or social worker helps them to come to terms with their diagnosis.

If you have specific questions or would just like to talk to someone about how you're feeling you can contact our Helpline on **0300 373 1000** or at [helpline@cysticfibrosis.org.uk](mailto:helpline@cysticfibrosis.org.uk).

You can also visit our Forum, an online community where people affected by CF can share their thoughts and experiences. You can find the Forum at [cysticfibrosis.org.uk/forum](http://cysticfibrosis.org.uk/forum)

**“It was a long journey from the age of six and countless accusations of faking my illness, to a five-minute sweat test at the age of 23.”** – Shad

**“I felt shocked and angry that this hadn't been picked up before. Questions ran through my head. The main one: will I die? I was scared and didn't know what to expect. I also felt relieved that I had a diagnosis and the things I was experiencing weren't in my head as I had been made to feel previously.”** – Vicky

**“I do still have negative feelings about it at times, but they tend to be worries about my family. I have a nine-month-old son and I want to be here for him and be fit and healthy for as long as possible, so actually it just encourages me to stay focused and consistent.”**

– Michelle

**“It took me a while to finally accept her diagnosis and come to terms with the worst-case scenarios. Processing those thoughts and feelings was very hard and at times it felt like I was grieving.”**

– Shaun, Michelle's husband

## What do I tell my family and friends?

What you tell your family and friends about your diagnosis is up to you. If you have a good relationship with them it could be helpful to reach out for support.

As you know, CF is a genetic condition, which means there might be implications for other people in your family. It could be helpful to sit down with your family and speak about the feelings that everyone has about the diagnosis.

If this is not something you feel comfortable doing, you could instead provide them with information from your CF team or direct them to the Cystic Fibrosis Trust website. We have information resources about all aspects of CF, which you can read at [cysticfibrosis.org.uk/publications](https://cysticfibrosis.org.uk/publications)

Some people have told us that they tell their friends and families not to search for information online. This is because information on the internet can be out of date or scary when taken out of context.

If you have children, it's important that you speak to them about your diagnosis. The way you approach this will obviously depend on the age of your child and how CF impacts your life. Your CF team might be able to support you with having these conversations. We have written two books for children who have parents with CF, which you can download or order at [cysticfibrosis.org.uk/rosieandseb](https://cysticfibrosis.org.uk/rosieandseb)

**“One time at a party hosted by my cousins, I was being introduced as the one who has ‘the adult version of CF.’” – Shad**

**“I was so scared about how people would change around me or think about CF because of what is written about it online, especially since a lot of the common symptoms didn't actually apply to me. However, that wasn't an issue. Many people have taken the time to understand the condition and how it affects me personally.” – Michelle**

**“At first, I would worry about Michelle a lot, constantly asking how she was feeling and if she was ok, which wasn't what she wanted to be reminded of.”**

– Shaun, Michelle's husband

## Further education and employment

You might be worried that your diagnosis could impact your education or employment. However, there is support available to you.

If you are in further education or planning to go into it, consider talking to your college or university about your condition. You should be able to work together so that you can fulfil your course requirements and complete your treatments. If you are thinking of moving to go to college or university, find out where your nearest CF centre would be. Take a look at our pack on leaving school at [cysticfibrosis.org.uk/leavingschool](https://cysticfibrosis.org.uk/leavingschool)

Many adults with CF are employed and manage to fit their treatments and hospital appointments around their work. With the support of your employer you should be able to do this too.

It's against the law for an employer to discriminate against someone because of a disability. This includes cystic fibrosis. However, if you don't make an employer aware that you have a disability, you may lose protection under discrimination law. Find out more about the rights of people with disabilities at [cysticfibrosis.org.uk/disability](https://cysticfibrosis.org.uk/disability)

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 that work for you. For more information about your employment rights, visit [cysticfibrosis.org.uk/work](https://cysticfibrosis.org.uk/work). You can also contact our Helpline on 0300 373 1000 or email [helpline@cysticfibrosis.org.uk](mailto:helpline@cysticfibrosis.org.uk).

**“I'm lucky in that prior to diagnosis I was on a 35-hour week with flex time, so the flexibility that offers is great. Now however, I am thinking of changing jobs because after 30 years the pressures of work are getting to me.” – Derek**

**“CF has impacted my energy levels, so my ability to work from an office with my peers has been greatly hindered.” – Shad**

## General lifestyle

Being diagnosed with CF does not change who you are as a person, and it doesn't have to change everything about how you live your life. However, there are some things that you might need to be more aware of.

People with CF can carry specific bugs in their lungs. These are harmless to people who don't have CF, but are very dangerous to others with the condition. If people with CF meet and share these bugs, this is called cross-infection. For this reason, you should never meet with other people with CF in person. Your CF clinic will be carefully managed to avoid cross-infection. You should also try to avoid people who have illnesses that you could catch, like coughs and colds.

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. Things like rotting vegetation, mud and stagnant water can harbour bugs that could pose a risk to your health.

Everyday life might take more planning than it used to. A common challenge that comes with having CF is making sure you have enough time and energy to do the things you want to do, whilst fitting in your treatments, clinic visits and maybe hospital stays.

You may also need to put more planning into going on holiday, as travel insurance can be more expensive for people with pre-existing health conditions like cystic fibrosis. There are also some activities that you may be advised to avoid. Visit [cysticfibrosis.org.uk/travel](https://cysticfibrosis.org.uk/travel) to find out more.

Remember, your CF team will be able to support you with any questions you might have about your condition and how it affects you as an individual.

**“Holidays take some planning, although I can currently get cover under a group policy through work.” – Derek**

**“Some of my medication has to be refrigerated, and it is actually not always common for hotels to have mini bars!” – Michelle**

## What support is available?

You are not alone, and there are almost 6,000 adults with CF in the UK. The Cystic Fibrosis Trust is here to help you by providing practical, emotional and financial support. This includes:

- Our Helpline, which can provide you with advice about CF or a listening ear if you need someone to talk to. Visit [cysticfibrosis.org.uk/helpline](https://cysticfibrosis.org.uk/helpline) for opening hours.
- Financial support like grants, benefits advice and income maximisation. Find out more at [cysticfibrosis.org.uk/financialsupport](https://cysticfibrosis.org.uk/financialsupport)
- CF Connect, a scheme that allows people affected by CF to gain connections over the telephone with others who have been through similar experiences.
- Information resources covering everything from physiotherapy and nutrition to information on employment. Visit [cysticfibrosis.org.uk/CFConnect](https://cysticfibrosis.org.uk/CFConnect) to find out more.

**“I have accepted my condition now and feel thankful for the team of people who help me. I am grateful that I am living and breathing and understand the reasons why I have had these symptoms most of my life.” – Vicky**

**“I envy the Michelle pre-diagnosis because she didn’t have to get up earlier and do physio, and take tablets with every meal. But on the other hand, I have learnt so much about my body, the ways to look after it and why it is so important to maintain good health.” – Michelle**

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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](https://cysticfibrosis.org.uk)

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