



3. Could our baby have cystic fibrosis?

A guide to help women with cystic fibrosis make informed choices

Could our baby have cystic fibrosis?

The genetics of CF

When any couple is considering having children, one of the concerns is – will our baby be healthy? This may be one of the questions passing through your mind, particularly since you probably already know that cystic fibrosis is inherited. The following information is likely to apply to you in general terms but it is often very useful to go and discuss the situation fully with your local Clinical Genetic Service.

What are chromosomes and genes?

Chromosomes are made up of strands of a molecule called DNA and genes are segments of this DNA. Genes provide a genetic code, or blueprint, for the body to develop and function



correctly. We inherit one set of genes from each of our parents, so that we have two copies of each gene, and we pass one set on to our children. If the genetic code alters in some way, eg. the order gets muddled or part of it is

removed, it will affect the ability of the body to work properly. It is not unusual for errors to occur in the genetic code – many do not cause a problem, but some do, and cystic fibrosis is one of these. If this happens the alteration (or mutation) can then be passed on to our children; there are several different ways in which this can occur. In cystic fibrosis, illness can occur only if an altered gene is inherited from both parents; this is called recessive inheritance. A person who has one altered gene and one with the normal code is said to be a 'carrier'.

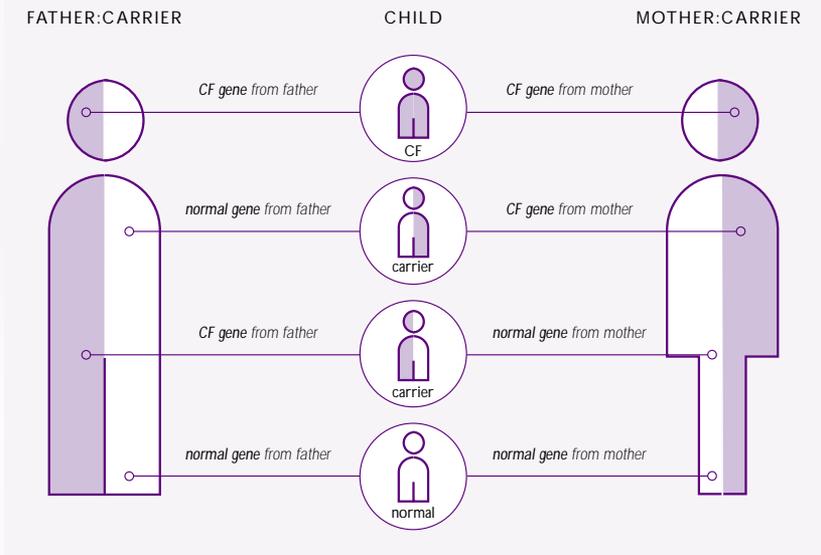
How does this apply to me and my children?

CF can only occur if a person has two altered copies of the CF gene. There are several hundred different alterations known within the gene, but only a few of them are commonly found in the UK population. You may already know which gene alterations you have. If this test has not yet been done you can discuss it with the CF team or ask to be referred to the genetic service.

Many people with CF in the UK share the same gene alteration called delta F508; 60% will have 2 of these and a

If both parents are carriers, a child has:

- a one in four chance of being born with CF
- a two in four chance of being a carrier but not having the disease
- a one in four chance of being completely free of CF, i.e. not



further 30-35% will have one copy of delta F508 plus another CF gene. To some extent, the particular mutations you have play a part in what kind of trouble cystic fibrosis causes you. Knowing your own gene alterations also helps other people in your family if they want to know if they are carriers, because the laboratory knows exactly what to look for.

What happens if I have children?

Whether we are male or female, the cell division producing eggs or sperm results in only one of each pair of chromosomes, and hence genes, being present. This will mean that all of your eggs will have a single copy of the CF gene. Your children will receive their other set of genes from your partner. If your partner does not carry a CF gene, then your children will only inherit a single copy from you making them a carrier, but they will not have CF.

What is the chance of my partner being a carrier?

In the UK, it is comparatively common to carry a CF gene, about 1 in every 25 people do. We can work out the chance of you and your partner having a baby with CF from this information:

- *Your own chance of passing on a CF gene is 1 (ie. 100% certain)*
- *Your partner's chance of being a carrier is 1 in 25*
- *If he were a carrier, the chance of passing on a CF gene and your baby having CF would be 1 in 2*
- *By multiplying these figures we arrive at a statistical risk of you having a baby with CF of 1 in 50*

The incidence of CF is different in different parts of the world so that the likelihood of a person being a carrier will vary



The incidence of CF is different in different parts of the world so that the likelihood of a person being a carrier will vary and needs to be sorted out individually. This is best done through your local Clinical Genetics Service.

Many people who are actually contemplating a pregnancy want to obtain as much information as they can beforehand, including checking to see if their partner carries a CF gene alteration. This can be done quite easily, usually from a blood test, but it is usually better to make sure you both understand the implications by discussing this with a genetic counsellor.

If the test does not show the presence of one of the common gene alterations, your partner's chance of being a carrier is much reduced. It will not, however, be zero because it is not possible to check for the many hundred rare mutations. The actual risk figure remaining will be in the order of 1 in 200 (if your partner comes from the UK).

We can now modify the above calculation regarding the chance of the baby having 2 CF genes:

- *Your chance of contributing a CF gene remains 1*
- *Your partner's chance of being carrier has been reduced to 1 in 200*
- *If your partner is a carrier the chance of passing on the gene is 1 in 2*

If we again multiply this it shows that the chance of a baby with CF is now



about 1 in 400 which many people find quite reassuring.

Of course, your partner's test result may show him to be a carrier of the CF gene and in this situation the risk of you having a baby with CF is much higher—1 in 2.

If you found yourselves in a high risk situation like this you might want to consider what options are open to you.

What are the options if my partner is a carrier of the CF gene?

- *Remaining childless – See section in leaflet 4*
- *Going ahead with a pregnancy and ignoring the risks. If you do become pregnant then you have a 50/50 chance that the baby will have CF. All your babies will be carriers of the CF gene. You may be happy to accept these risks – but remember if your child does have CF you may face difficult decisions later on because of the risks of cross-infection between you and your child*
- *Going ahead with a pregnancy and having pre-natal testing*

Prenatal testing

Prenatal testing can be undertaken at about 9-11 weeks of pregnancy (Chorionic Villus Biopsy – CVB).



This involves taking a small piece of developing placenta tissue. The genes here are the same as those of the baby. There is a small risk of miscarriage – about 1 in 50 – which you should be aware of. For this reason, a prenatal test is only offered if there is a high chance of CF in the baby. If the test confirms CF in the baby, you are faced with the decision of whether or not to continue the pregnancy.

- *Having In-Vitro Fertilisation (IVF) treatment together with Pre-Implantation genetic diagnosis*

Pre-implantation genetic diagnosis (PGD)

If you have In Vitro Fertilisation (IVF) treatment to help you to become pregnant (see full explanation in leaflet 6), it might be possible to use techniques which would allow a cell from each developing embryo to be removed and tested for CF gene alterations before any are placed in the womb. The results are known after a few days and a discussion takes place with you about which embryos should be selected for transfer to your womb or frozen for storage. This would enable selection of embryos which do not have two faulty CF genes and avoids the need to intervene in the pregnancy later on. Pre-implantation genetic diagnosis is a new technique and the details about success rates are limited at present. This would only be an option if your partner was a known CF carrier.



IVF treatment means it might be possible to allow a single cell from the embryo to be tested for CF gene alterations before they are placed in the womb

Costs of this treatment: PGD adds approximately £1000 per IVF cycle.

As you may already know these procedures are very expensive and are not usually offered on the NHS.

- *Using the sperm or eggs of a donor who does not carry the CF gene – see sections in leaflet 6 entitled: Donor insemination, Egg donation, and Surrogacy*
- *Fostering or adoption – see section in leaflet 6*

There is obviously a lot to consider when planning a pregnancy. Our understanding about the genetics of CF is developing all the time so it can be useful to be referred for genetic counselling at the planning stage. You may also wish to have the help of a counsellor if you are faced with making difficult decisions.



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Leaflets in the series:

- Leaflet 1:** Can I have a baby?
- Leaflet 2:** How can I have sex safely?
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- Leaflet 4:** Should we have a baby?
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- Leaflet 6:** How could we become parents?
- Leaflet 7:** How does it feel to go through infertility treatment? – A patient's perspective
- Leaflet 8:** What is it like to be a parent?

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