Cystic fibrosis

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of cystic fibrosis and where to get help.

What is cystic fibrosis?

Cystic fibrosis (CF) is an inherited disease primarily affecting the lungs and digestive system

- five babies are born with CF each week in the UK
- two young lives are lost to CF each week
- approximately half of those living with CF are likely to live past their late 30s

CF affects many internal organs, but in particular the lungs and digestive system by clogging them with thick sticky mucus.

What causes cystic fibrosis?

It happens because the gene that is responsible for making a protein which regulates the amount of chloride and sodium going in and out of the cells is faulty. This makes the mucus that lines the airways and other organs thick and sticky and therefore less able to clear secretions and protect against infection.

Human beings have about 30 to 40,000 different genes, each of which has a function in making an individual person. The genes are arranged in pairs (one of the pair from each parent) on 23 chromosomes. Inevitably, some of these genes are faulty; a normal gene can overcome a faulty one, but if both genes in the pair are faulty, the genetic instructions cannot work. About one person in every 25 carries the faulty CF gene.

Most people carry different faulty genes but in CF (and other recessive conditions) parents, though healthy themselves, carry the same faulty gene, and risk passing them on to their children. Each pregnancy carries a 25 per cent chance of the child being affected.

What are the signs and symptoms of cystic fibrosis?

Often in newborn babies there are no immediate signs of any problems and so coming to terms with the diagnosis can be difficult. It is important, however, for appropriate treatment to be started at the earliest opportunity so that we can keep the baby as well as possible for as long as possible and delay the onset of symptoms.

When symptoms appear these may include a cough, chest infections, difficulty absorbing fat which results

in poor weight gain. A combination of medication and physiotherapy (physical activity and airway clearance) can help control lung infections and prevent lung damage.

The pancreas is a gland in the abdomen and one of its functions is to produce digestive juices (enzymes) that help digest and absorb the food we eat. In most babies with CF, the small channels through which the enzymes flow become blocked with sticky mucus. This means that they cannot digest and absorb fat as well as they should. To help this, we will give your baby pancreatic enzymes with each feed. If your baby needs these enzymes, the dietitian will explain this in more detail to you. Taking pancreatic enzymes will not affect breastfeeding.

We all have mucus in our lungs, which helps them to function, but in babies with CF, the mucus is abnormally thick. This can block the smaller airways and lead to infection. To help prevent this happening, we usually start babies on antibiotics and teach parents about airway clearance (chest physiotherapy). Physical activity is a very important part of CF care and we encourage this from very early on – the physiotherapists will explain more about this when you meet them

Occasionally babies with CF display signs within the first couple of days of life with an obstruction of the bowel (called meconium ileus). The baby fails to open their bowels to pass meconium (a thick black material present in the bowels of all newborn babies) because the meconium is so thick that it blocks the bowel. Babies with meconium ileus may need an urgent operation to relieve the blockage, although in some cases it can be managed without surgery.

People with CF are prone to developing bone disease (weak bones) due to the nutritional and other problems involved with the disease. This often causes problems in early adulthood and it is therefore very important that careful attention is paid to good nutrition and exercise to keep the bones healthy and strong.

CF is associated with fertility problems, particularly in men where the tubes that carry sperm are blocked. Women with CF do produce healthy, fertile eggs so effective contraception is necessary.

CF can also cause blockage of small ducts in the liver. This only happens to approximately eight per cent of people who have CF, but it is serious and people with CF are regularly monitored for liver complications.

How is cystic fibrosis diagnosed?

Newborn screening for CF is routinely undertaken across the UK. The test is part of the heel prick sample of blood that is taken in the first few days after a baby is born. The sooner CF is diagnosed, the earlier the appropriate treatment can commence.

Although CF is now mostly diagnosed through this screening process, there are some babies and older children (and even adults) who are diagnosed following unexplained illness. In these cases the test used to confirm the diagnosis is either a sweat test (as those with CF have a greater than normal amount of salt in

their sweat) or genetic testing with a blood sample or a swab of the inside of the cheek).

How is cystic fibrosis treated?

Medication for the chest may include:

- antibiotics to help treat or to control chest infections
- medicines to help clear mucus from the lungs
- other medications, such as steroids or bronchodilators, may also be necessary depending on the individual clinical situation

Medicines for the digestive system include pancreatic enzymes, extra vitamins and nutritional supplements.

Physiotherapy is an important part of the management of CF. Regular physical activity helps to keep the body fit (especially the lungs and heart), lungs clear, bones healthy and muscles strong. In addition airway clearance techniques are used to help clear mucus from the airways.

Other medications and treatment may also be necessary depending on each individual situation.

What happens next?

At present, there is no cure for CF but there are many treatments currently available and exciting advances are also being made in this field.

Further information and support

The **Cystic Fibrosis Trust** is the main organisation in the UK offering support and advice to anyone affected by CF. Call their helpline on 0300 373 1000 or visit their website at www.cysticfibrosis.org.uk

Notes

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