

## **Cystic Fibrosis (CF)**

### **What is CF?**

CF is a disorder of exocrine glands. This results in thicker than average secretions being produced which primarily affects two systems the lungs and the pancreas.

### **What problems can be caused in the lungs?**

Thick secretions in the lungs causes blockage within the small air tubes and can lead to repeated chest infections, which over time can severely damage the lung. To help prevent the airway obstruction these children require antibiotics and chest physiotherapy.

### **What problems can be caused in the Pancreas?**

Thick secretions also cause blockage within the pancreas. The pancreas serves two functions, the production of insulin which controls our blood sugar and the production of enzymes which help to digest our food. In CF blockage of the passages through which the enzymes enter the bowel means that they are unable to digest their food properly. As undigested food cannot be absorbed these children are unable to absorb sufficient calories to grow and therefore tend to be small. If recognised enzyme supplements can be added to their food to help overcome this problem. Individuals with CF are also more prone to developing diabetes

### **How do babies get CF?**

CF is inherited as an autosomal disorder. In order to have a child with CF the parents both have to carry the condition. Normally we have two copies of every gene which we inherit one from each parent. If for whatever reason we have a duff or non functioning gene then as long as the other one is normal we do not have a problem. When we have children we have to pass on one copy of our genes. If we have a child with a partner who carries a mistake in the same gene then there are 4 possible options.

- 1) We both pass on normal copies of the gene
- 2) dad passes on a normal copy and mum the abnormal gene.
- 3) mum passes the normal copy and dad the abnormal gene
- 4) Both mum and dad pass on the abnormal gene.

It is only in the 4<sup>th</sup> scenario that the child acquires the disease. Each time this couple have a child there is a 1 in 4 risk. Unfortunately the body does not remember what has happened previously so it does not work that having had an affected child that the next three will be fine. The risk each time is the same.

One in every 25 of the Yorkshire population carries a risk that one or other of their CF genes will be abnormal. This means that  $1/25 \times 1/25 \times 1/4$  or one in every 2500 of the population will be born with CF. Currently all children in Yorkshire are tested for CF shortly after birth by a heel prick test.

**Can CF be detected before birth?**

We can test for this condition before birth through either a chorion villus biopsy or an amniocentesis. However we can also identify many but not all of the carriers from a blood test from the parents. If we take the blood test and neither parent appears to be a carrier than it makes it highly unlikely that the baby will have CF. the blood test takes between 10 – 14 days. CF does not usually cause any ultrasound abnormalities on scan, but occasionally a baby will have echogenic bowel (See page in fetal abnormality database on Soft Markers). If this is the case then the risk of a baby having CF is in the order of about 1 in 50.

**Is there a cure for CF?**

There is currently no cure for CF, but treatments controlling the problems associated with the disease have improved dramatically in the last decade and children born with CF at the current time can expect a good quality of life in most cases. For further information on the condition the UK CF website address is [www.cftrust.org.uk](http://www.cftrust.org.uk)