Medicine for Managers

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Cystic Fibrosis

Cystic Fibrosis is an inherited disorder. It cannot be acquired after birth. It is associated with the existence of a faulty gene which is associated with salt and water control and its transfer between the body cells and the extracellular fluid. The gene is carried harmlessly by about one in twenty-five of the population. It is estimated that about one in every 2,500 births is a cystic fibrosis child.

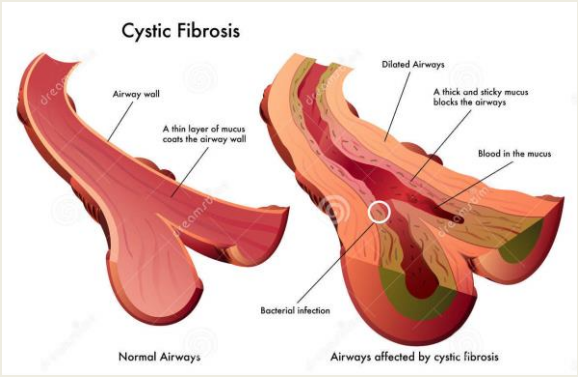
The cardinal clinical feature of the condition is the build up of thick, sticky mucus which occurs in the lungs, in the digestive tract and in other parts of the body causing a variety of symptoms.

The condition reduces life expectancy but over the last seventy years the situation has changed dramatically. In the 1940s, a child born with cystic fibrosis would have been unlikely to live beyond their first birthday.

Nowadays, they live well into adulthood and survival rates continue to improve with an average lifespan of 45 years or more. 80% of deaths are due to lung complications.

Symptoms are seen in the early years of life and the condition may first reveal itself in infancy if a baby develops ***meconium ileus*** (a condition where the contents of a baby's bowel are extremely sticky and cause the bowel to become blocked).

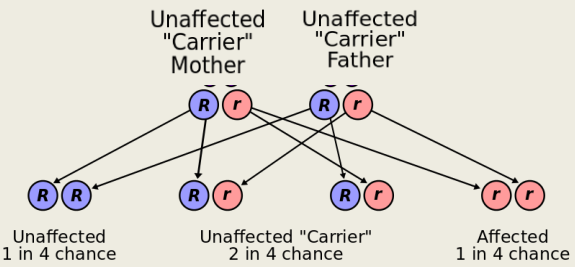
In childhood the main symptoms are recurrent chest infections together with persistent, 'mucusy', sometimes hacking coughs associated with difficulty in clearing the thick mucus. There may be associated wheezing and shortage of breath. Other non-respiratory symptoms include diabetes, osteoporosis, liver dysfunction and



poor weight gain (which is the result of chronic infection, poor absorbtion of nutrients from the gut and increased metabolic demand due to chronic ill health).

Sinus infection and facial pain, nasal discharge and polyps are common.

About 95% of males who reach adulthood are infertile as are about 20% of females in whom thickened cervical mucus blocks ovulation and disrupts menstruation.



For a child to be born with cystic fibrosis, the affected gene must be acquired from ***both parents*** although neither of them will normally be aware that they carry the gene.
The offspring children will have a twenty-five percent chance of having the disease, acquiring the gene from both parents Two chances in four will be the birth of an unaffected and asymptomatic 'carrier' of the gene and there will be a one in four chance that the child will be completely unaffected. Many different mutations can produce the features of cystic fibrosis and the variations in genetic cause account for the variation in severity.

Most cases of cystic fibrosis are now diagnosed promptly after birth because of the use of the neonatal blood spot test. The test is simple and involves collecting a drop of blood from a heel prick. The specimen is then tested for cystic fibrosis, and also for sickle-cell disease, congenital hypothyroidism and a number of inherited metabolic diseases (diseases where babies may develop serious illnesses as a result) which can all be successfully treated with diet and medication.

If the blood test raises the suspicion of cystic fibrosis, further tests may be carried out:

1. ***Sweat test*** to measure the amount of salt in the sweat. Cystic fibrosis sufferers have a higher level of salt in the sweat than the normal.
2. ***Genetic test*** which is taken as a sample of saliva to identify the faulty gene which causes cystic fibrosis.

Unfortunately there is no cure for cystic fibrosis but enormous advances in treatment and management have been made since the war. Cystic fibrosis patients acquire chest infections due to a variety of organisms and these days they can be identified and treated efficiently with appropriate antibiotics. Drugs are available to reduce the viscosity of the mucus so that it can be coughed up more easily. This is helped by physiotherapy techniques provided by skilled and specialist staff to develop methods of coughing up the sputum through postural techniques, ***active cycle breathing techniques*** (ACBT) and the use of devices. In addition there is modern medication to reduce the associated inflammation in the airways and to produce bronchodilation (opening up of the airways). Dietary control is important together with the use of supplements to ensure that digestion is effective and that there is good absorbtion of nutrients to avoid the problems of weight loss and malnutrition.
For those sufferers in whom the lungs have been badly damaged lung transplant may be needed eventually.

During my medical practice lifetime there have usually been one or two cystic fibrosis children in the practice at any one time and for me the change has been amazing. Children born in the 1970s, who would have died in childhood, now still alive and able to live a reasonably normal life through the wonders of modern medicine.

The condition was only formally identified in 1938 by Dorothy Hansen although historical

texts suggest that the condition was recognised as far back as the sixteenth century and there are some indications that it was much earlier. The genetic basis really only started to be clarified in the 1980s.

The ***Cystic Fibrosis Trust*** provides information about the condition and they can provide a parent pack of information. The trust may be contacted through:

https://www.cysticfibrosis.org.uk

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