What you need to know about...

CYSTIC FIBROSIS

AETIOLOGY
■ Cystic fibrosis is an inherited disorder of the exocrine glands. Both sexes can be affected.
■ It is an autosomal recessive disorder, which means that two copies of the abnormal gene are required to develop cystic fibrosis.
■ People with one affected gene and one unaffected gene are known as carriers; they will not develop cystic fibrosis themselves.
■ If a person carrying an affected gene has a child with another carrier, then the chance of the child inheriting two affected genes is 25 per cent. This happens once in every 2,500 live births, resulting in approximately 250 cystic fibrosis births in England and Wales each year.
■ Note that there is also a 25 per cent chance that a child of two carriers will inherit two normal genes and not be affected in any way.

PREVALENCE
■ Cystic fibrosis is usually recognised in infancy or early childhood.
■ It occurs mainly in Caucasians.
■ There are around 20,000 Europeans with cystic fibrosis.
■ In Britain, 4–5 per cent of the population are carriers of the cystic fibrosis gene.
■ There is no known cure for cystic fibrosis, although life expectancy has improved over the last few decades.
■ With early diagnosis and treatment most patients can expect to reach early adulthood. For those born in the 1990s the life expectancy is estimated to be around 40 years.

SIGNS AND SYMPTOMS
■ Cystic fibrosis causes the glands to produce abnormally thick secretions of mucus. The glands most affected are in the pancreas, the respiratory system, and the sweat glands.
■ The first sign of cystic fibrosis may be when a newborn develops meconium ileus – an obstruction of the small bowel by viscid stool.
■ Chronic cough.
■ Frequent, foul-smelling stools.
■ Excessive mucus in the lungs leads to persistent upper respiratory infections, emphysema and atelectasis.

DIAGNOSIS
■ The most reliable tool is the sweat test. This will show elevated levels of sodium and chloride.

TREATMENT
■ The main aim of treatment is to prevent respiratory infections; these are the most common cause of death.
■ Mucolytic agents, bronchodilators and mist tents are used to help liquefy the thick tenacious secretions.
■ Physiotherapy, such as postural drainage and breathing exercises to help dislodge secretions, is vital.
■ Broad-spectrum antibiotics and new antipseudomonal antibiotics may be used prophylactically.
■ Heart-lung transplantation has become an established treatment for end-stage cystic fibrosis.

NURSING IMPLICATIONS
■ The nurse’s role in health promotion, including exercise and nutrition, is crucial. Exercise helps to loosen the mucus – stimulating coughing to clear it out of the lungs – and improves the patient’s overall condition.
■ Nutritional advice is needed to help the patient overcome the malabsorption and malabsorption associated with cystic fibrosis.
■ Explanation of prenatal screening programmes is important as couples are offered a termination if they have a prenatal diagnosis of cystic fibrosis. The tests are expensive and have certain risks to the mother, so they are not used routinely.

RESEARCH
■ The discovery of the cystic fibrosis gene means that research is aimed at correcting the genetic defect.

WEBSITES
The Cystic Fibrosis Trust: www.cftrust.org.uk

Chest X-ray of the lungs of a patient with cystic fibrosis