

44. CYSTIC FIBROSIS

Related conditions considered in other chapters Cardiac and Respiratory Conditions Chapter 11

44.2 Introduction

44.2.1 Cystic fibrosis is an inherited condition due to an abnormal gene. The clinical features are due to an alteration in the mucus produced in the cells lining the lungs and in many organs which secrete mucus, notably the pancreas.

44.2.2 In cystic fibrosis there is increased salt in sweat, the secretions from the lining of the airways in the lungs are more viscous (stickier) and there is pancreatic insufficiency. Pancreatic insufficiency causes decreased secretion of certain enzymes which digest food in the gut, and leads to malabsorption of food with the passage of frequent, loose, offensive stools. The thick secretions in the airways result in an increase in recurrent lung infections, which eventually lead to lung tissue destruction. Late complications include diabetes and liver damage.

44.2.3 The prognosis has consistently improved; 90% of children with cystic fibrosis now survive well in to their teens and many well into young adulthood. This advance is due to improved treatment of bronchial infections and better maintenance of nutrition, together with meticulous attention to measures such as chest physiotherapy. Heart-lung transplantation is increasingly being undertaken with consequent limitation in the severity of disabilities.

44.2.4 At any particular time the condition of a child with cystic fibrosis can be classified as:

- (i)** Normal clinically - although the child has the disease, symptoms are controlled by regular treatment, and growth and development are normal.
- (ii)** Mild - the child has symptoms referable to the chest and/or digestive system, but is free from sputum (mucus from the lungs sometimes containing blood or pus). Gaining weight and enjoying normal activities are achieved on regular treatment.
- (iii)** Moderate - the child has increasing sputum, problems with the digestive system, limitation of general activities and variable impairment of growth and development.
- (iv)** Severe - the child has repeated flare ups of chest infection, persistent cough with copious sputum, weight loss, shortness of breath and greatly reduced exercise tolerance. A proportion have additional complications.

44.2.5 Most adults with cystic fibrosis have serious respiratory disease. Respiratory failure is the most frequent cause of death. Recurrent chest infections result in breathlessness and copious, thick sputum, at times blood stained. As the disease progresses, the chest infections become more frequent and there may be wheezing, finger clubbing and blue discolouration of the skin due to poorly oxygenated blood (cyanosis).

44.2.6 Treatment includes physiotherapy (mainly postural drainage) at least twice a day, antibiotics (both oral nebulised - in the form of a spray - and intermittent courses intravenously), inhaled bronchodilators and a high energy and protein diet with supplements of pancreatic enzymes and vitamins. It has long been recognised that close attention to treatment has a significant effect on prognosis. However, it is now apparent that prognosis can be further improved when the management is supervised by specialist centres which give very intensive therapy in the form of physiotherapy, intermittent intravenous antibiotics, nebulised bronchodilators and nutritional support.

44.3 Care Needs

44.3.1 The needs of those with cystic fibrosis are dependent on the severity of the disease, the nature of any infection present and their age. History and clinical findings, including growth, will help assess these factors.

44.3.2 It is possible to maintain some children in a symptom free and generally healthy state by use of physiotherapy and other measures outlined in **44.2.6**. Precisely the same measures are used to treat children during acute infections, although they may be given more intensively, especially the physiotherapy. The need for the attention to keep a child in a healthy state is just as necessary as that required during acute infections. It is therefore important to note that children may only appear healthy because of the amount of attention being given, and that without this attention there would be a deterioration in the condition.

44.3.3 The amount of attention required by a child who receives physiotherapy twice daily, maintains normal height and weight, remains symptom free and engages in normal activities is likely to take at least 30 minutes twice a day. Some children are only maintained in this state with additional physiotherapy together with the frequent use of nebulised bronchodilators and antibiotics, with which they will require help. Nebulised antibiotics are usually given twice a day and will require assistance from a parent or carer until the child is mature enough and sufficiently independent to handle these matters alone. Some children may not be receiving the amount of attention their condition requires. A child who is not thriving, has repeated chest infections and restricted activity, despite receiving twice daily physiotherapy, in fact actually needs a programme of more intensive and frequent attention throughout the day.

44.3.4 There is an increasing trend towards the use of intermittent courses of intravenous antibiotics at home. During these courses, which usually last several days, administration of the antibiotics will be about three or four times throughout the day and will need to be given by an adult. Although these courses are given intermittently, they will nevertheless affect the overall amount of attention needed by the child over a period of time.

44.3.5 Adults and young adults, unless they are in a very poor condition, will be able to carry out postural drainage and administer bronchodilator drugs/antibiotics unaided. The age at which this level of independence is achieved will depend on clinical condition and maturity. It is unlikely that a child

would be able to manage treatment without assistance before puberty. Many older children also require assistance with the physiotherapy from time to time, especially when unwell.

44.3.6 Most people with all but the milder forms of cystic fibrosis require the support of a carer at home for the intensive and time consuming regimes that are so necessary. In mild or moderate cases it is not usual for attention to be required at night. However, if the general condition is poor, a session of physiotherapy may be required at night. Likewise, physiotherapy at night may be required during periods of acute infection. In these circumstances, sleep may be disturbed by distressing coughing or shortness of breath, and attention in connection with these may be needed.

44.4 Mobility Considerations

44.4.1 Cystic fibrosis in itself does not give rise to a need for assistance when walking out of doors. However, it can give rise to walking problems because of shortness of breath, particularly when it has reached the stage of being severe. The effects of cystic fibrosis on a person's mobility will need to be assessed on the evidence available in an individual case.

44.5 Heart/Lung Transplantation

44.5.1 In recent years, an increasing number of heart/lung transplants have been taking place. A significant number of these have been in patients suffering from cystic fibrosis. In uncomplicated cases, patients are discharged from hospital four to six weeks after surgery. Post operative patients are required to follow a complicated and closely monitored drug regime. In addition to this, the temperature has to be recorded twice daily and a daily record kept of the weight and certain measures of lung function. Consequently, it is to be expected that such people will continue to need a considerable amount of attention by day for a further year, although this time span will vary according to the circumstances of the individual case. At that time, continuing attention needs will depend on whether the transplant has been a success and on the particular circumstances of the case.

44.6 Duration of Needs

44.6.1 Once attention needs are established, in the absence of a heart/lung transplantation, they are likely to continue indefinitely.

44.7 Further Evidence

44.7.1 A factual report from the relevant hospital is likely to be of help in determining the history, clinical findings, height, weight, treatment and response to it and other factors which would determine the amount of treatment and attention required.