CONGENITAL ABNORMALITIES Cystic fibrosis

CONGENITAL ABNORMALITIES Cystic fibrosis

This is inherited as an autosomal recessive condition. It occurs most frequently among white people, in whom it is the most common inherited disorder (incidence of 1:2000 live births in the UK). Cystic fibrosis (CF) develops when there is a mutation in the CFTR (cystic fibrosis transmembrane conductance regulator) gene on chromosome 7. This gene creates a cell membrane protein that helps to control the movement of chloride across the cell membrane. CF is a multisystem disorder of exocrine glands that a ff ects the lungs, intestines, pancreas and liver and is characterised by elevated sodium and chloride ion concentrations in sweat. The mother may notice that the child is salty w hen kissed. Most of the organ damage is due to blockage of narrow passages by thickened secretions. Chronic pulmonary disease arises from plugging of bronchi and bronchioles. CF is the most common cause of chronic lung disease among childr in resource-rich countries. Cor pulmonale may develop later. At birth, the meconium may set in a sticky mass and produce intestinal obstruction (meconium ileus) (see Chapter 18 Secretions precipitate in the lumen of the pancreatic duct causing blockage, which results in duct ectasia and fatty replacement of exocrine acinar tissue. Pancreatic exocrine insu ffi ciency leads to fat malabsorption. Steatorrhoea is usually present from birth, resulting in stools that are bulky , oily and o ff ensive. The islets of Langerhans usually appear normal, but diabetes mellitus can occur in older patients. The liver may become cirrhotic as a result of bile duct plugging, and signs of portal hypertension may appear. Infertility is common owing to the absence of the vas deferens in men and thick cervical mucus in women. Outside the newborn period, the earliest clinical signs of CF are poor growth, poor appetite, rancid greasy stools, abdominal distension, chronic respiratory disease and finger clubbing. The appearance of secondary sexual characteris tics may be delayed. The diagnosis can be made by prenatal genetic testing, by the newborn blood spot (heel prick) test done on newborns in the UK and by the sweat test. Levels of sodium and chloride ions in the sweat above 90 /uni00A0 mmol/L confirm the diagnosis. Treatment is aimed at control of the secondary conse quences of the disease. Pulmonary function is preserved with aggressive physiotherapy and antibiotics. Malabsorption is treated by administration of oral pancrea tic enzyme prepa rations. The diet should be low in fat but contain added salt to replace the high losses in the sweat. With early diagnosis John Langdon Haydon Down (sometimes given as Haydon-Down), 1828–1896, physician, The London Hospital, London, and Superintendent, Earlswood Asylum for Idiots, Redhill, UK, described this syndrome in 1866. Johann Friedrich Meckel (the younger), 1781–1833, Professor of Anatomy and Surgery , Halle, Germany , described the embryological origin of the eponymous diverticulum in 1809. now expect to survive to their mid-thirties. Those with end- - stage lung

disease ma y be considered for lung transplanta - tion. Heterozygous carriers of the various gene muta tions are asymptomatic but can be identified by DNA analysis. There is a suggestion that such patients may develop pancreatitis later in life .

Revision #1
Created 2025-12-31 15:26:58 UTC by Omar Ayman
Updated 2025-12-31 15:26:58 UTC by Omar Ayman