Cystic fibrosis is a hereditary glandular disease in which the body produces abnormally thick mucus that obstructs the breathing passages, causing chronic lung disease. There is a deficiency of pancreatic enzymes and a general failure to gain weight. The disease is treated with antibiotics, pancreatic enzymes and a high-protein diet; sufferers must undergo vigorous physiotherapy to keep the chest as clear as possible.

In 1989 teams of researchers in Michigan, USA, and Toronto, Canada, discovered that cystic fibrosis is caused by mutations to a gene called the cystic fibrosis transmembrane conductance regulator (CTFR). Each human carries two copies of this gene. A person is called a cystic fibrosis carrier if one of their CTFR genes has the mutation. In order for a person to develop the disease, both their CTFR genes must have the mutation. This discovery has enabled the development of a screening test for carriers of the disease, which can also be detected in an unborn child.

There were approximately 9,000 cystic fibrosis sufferers in Britain in 2012.

Inheriting the disease Among people of European descent, one person in 22 is a carrier of the disease. If two carriers have children, each child has a one-in-four chance of having the disease, so that it occurs in about one in 2,000 pregnancies. Around 10% of newborns with cystic fibrosis develop an intestinal blockage (meconium ileus) which requires surgery. It is the commonest fatal hereditary disease amongst white people.

Treatment Cystic fibrosis was once universally fatal at an early age; now, although there is no definitive cure, treatments have raised both the quality and expectancy of life, so that sufferers may survive into their thirties or even forties. Management of cystic fibrosis is by diet and drugs, physiotherapy to keep the chest clear, and use of antibiotics to combat infection and minimize damage to the lungs. Some sufferers have benefited from heart-lung transplants.

Gene therapy Cystic fibrosis is seen as a promising test case for gene therapy, although there is currently no viable gene therapy-based treatment clinically proven to improve the health of sufferers.

In 1993, UK researchers at the Imperial Cancer Research Fund, Oxford, and the Wellcome Trust, Cambridge, successfully introduced a corrective version of the gene for cystic fibrosis into the lungs of mice with induced cystic fibrosis, restoring normal function.

In 1997, US researchers successfully cured mice with cystic fibrosis by administering gene therapy in utero. The fetus breathes in the corrective gene attached to an adenovirus and it becomes...
incorporated in the developing cells. All the mice were born healthy and survived to old age.

In 2001, the UK Cystic Fibrosis Gene Therapy Consortium was formed to develop a viable medical treatment for the disease using gene therapy. By 2005, the consortium had identified a promising gene therapy product using a liposome to transfer the modified gene via a selected DNA molecule (plasmid) into a human patient's cells. The consortium announced the start of the world's largest clinical trial of gene therapy for the treatment of cystic fibrosis, based on their product, in 2012.

cystic fibrosis

gene therapy for cystic fibrosis

APA

Chicago

Harvard

MLA


© RM, 2018. All rights reserved.
APA

Chicago

Harvard

MLA