Inherited Life-threatening Disorder

What is Cystic Fibrosis?

Cystic fibrosis is an inherited life-threatening disorder that damages the lungs and digestive system which makes it difficult to breathe. Cystic fibrosis is about genetics, you can’t catch it. But if both your parents have the CF gene, there’s a one in four chance you’ll be born with the condition. (Cysticfibrosis.org.au, 2019)

Frequency of Disease

If 10% of the children that had Meconium ileus will have cystic fibrosis. If a child's intestine is thicker and stickier than normal meconium, creating a blockage in a part of the small intestine called the ileum. In Australia, 1 out of 25 people have Cystic Fibrosis, that's approximately 3,000 people who had this disease in a year. (Betterhealth.vic.gov.au, 2019)

Is the Disease more common in a particular community/country/race?

Cystic fibrosis or CFTR mutation most commonly occurs in Caucasians of northern European carrying about 1 in 29 Caucasians. People who are born with cystic fibrosis will get two copies of the gene and later now that disease will be passed on/inherited. (Cff.org, 2019)

For people if they have one of the following concerns, please see the doctor;

Symptoms

- Hyperinflation - over-breathing your lungs, caused by a pulmonary blockage.
- Crepitations - This is the cracking noise in your joints.
- Finger clubbing - Odd changes in the shape of your fingers and fingernails.
- Frequent lung infections or pneumonia - Lung damage caused by a viral infection, sometimes the lungs may become solid and unable to breathe properly.
- Wheezing - Is a high-pitched sound that occurs when a person is breathing.
- Failure to gain weight, even though the child eats normal amounts
- Very salty sweat - how salty your sweat is.
- Poor height growth - If over time your height is still constant (not growing).
- Frequent sinus infections - An infection that causes a nasal blockage, causing difficulty breathing.
- Fatigue - Mental health problems. (HealthEngine Blog, 2019)

Genetics of the Disease

Cystic fibrosis disease is an autosomal recessive disease in which the gene is located on chromosome 7. Most common mutations occur in 70% of CF chromosomes. Cystic fibrosis is

caused by a faulty gene, that disrupts the movement of salt and water in the body’s cells making the mucus and people with CF ticker and sticker. This sticky mucus causes problems, mainly in the lungs and digestive system but also affects other parts of the body too. The small airways in the lungs can get clogged up with this mucus, causing infections and over-time damage to the lungs. CF is an autosomal recessive disorder involving the CFTR gene which stands for cystic fibrosis transmembrane conductance regulator and the gene codes for the CFTR protein CF develops when there's a mutation in the CFTR gene but because it's autosomal recessive you need to inherit two mutated CFTR genes one from mom and one from dad. During Caucasian history, 1 in 29 has been affected by CF gene mutation. (En.wikipedia.org, 2019)

Reference of the image showing lists of X and Y chromosomes and circling chromosome 7 as it can produce a faulty gene. (En.wikipedia.org, 2019)

Prognosis

Nearly eleven thousand in the UK and in the UK each week two people die from CF and five more babies are born with it. About one in twenty-five people carries the faulty gene and most don’t even know it. Recent figures suggest that half the people with CF will live past their forty-seventh birthday. But it’s also thought that a baby born today with CF could live longer, as life expectancy continues to increase with the amazing advances in treatment and care. (Cysticfibrosiscare.org.uk, 2019)

Treatments

There's currently no cure for cystic fibrosis, but a number of treatments are available to help control the symptoms, prevent complications, and make the condition easier to live with the following treatments include: (Mayoclinic.org, 2019)

- Antibiotics to prevent and treat chest infections
- Medicines to make the mucus in the lungs thinner and easier to cough up
- Medicines to widen the airways and reduce inflammation
- Special techniques and devices to help clear mucus from the lungs
- Medicines that help the person absorb food better
- Following a special diet and taking supplements to prevent malnutrition

It may be an unhealthy relationship but there is a lot of daily treatments to tackle cystic fibrosis effectively which can make our union that bit more harmonious. A whole host of antibiotics to fight infections in your lungs, physiotherapy to help shift the mucus that builds up around the organs. Enzyme capsules to take with food. We need a high special diet to make sure our bodies get all the nutrients that it needs. More drugs to thin the sticky mucus. If conventional treatments no longer work, a lung transplant may eventually be needed if the lungs become greatly damaged. (En.wikipedia.org, 2019)

Cystic fibrosis presents daily compromises and challenges, but with careful management and the right treatment people with CF and lead full and happy lives.