Cystic Fibrosis

Cystic Fibrosis (CF) is an inherited multi-system disease that primarily affects the lungs and digestive system. CF is caused by a defective gene that creates a protein called cystic fibrosis transmembrane conductance regulator (CFTR). When this protein works normally, it helps regulate the movement of chloride and water within the body. A mutated form of this gene causes the body to produce abnormally thick and sticky fluid, called mucus. This mucus accumulates inside the lungs and in the pancreas and liver, organs that help with digestion. Buildup of viscous mucus leads to life-threatening lung infections, chronic bronchitis, and difficulty breaking down and absorbing nutrients from food. Symptoms of CF include coughing and difficulty breathing (due to mucus buildup in lungs), increased sweat with a very salty taste, recurrent lung and sinus infections, reduced growth (due to nutritional deficiencies), and infertility.

CF is one of the most common chronic lung diseases in children and young adults, affecting 70,000 people worldwide. It is most prevalent in people of Northern or Central European descent, with one in 25 Caucasians carrying a copy of the defective gene. Most children with CF are diagnosed by two years old, though a minority are not diagnosed until adolescence or older. CF is a life-threatening disorder, but modern treatments have enhanced and extended life expectancies for children and adults with CF, who now often live into their 30s, 40s and beyond.

Risk Factors

CF is an autosomal recessive disorder, which means that a person needs to inherit two copies of the mutated CFTR gene, one from each parent, to develop the disease. Millions of people carry only one copy of the defective gene—this is called a heterozygous carrier. These people will not show any signs or symptoms of CF. If two carriers have children together, approximately one quarter of their children are likely to have CF disorder, half will likely be carriers like their parents, and one quarter will likely not carry the defective CFTR gene.

Diagnosis and Management

It is very important to diagnose cystic fibrosis early, as quickly beginning treatment can improve both survival and quality of life. CF can be diagnosed by genetic testing before birth, by a blood test in newborns, or by a sweat test in children. In order to rule out false positives, any babies diagnosed by genetic or blood testing are also given a sweat test. High levels of sodium and chlorine in the individual's sweat are diagnostic for CF and are responsible for giving the skin a characteristic salty taste.

It is important for the patient to live a healthy lifestyle, including good nutrition, staying hydrated, regular exercise, frequent health check-ups, and routine vaccinations. Follow-up and monitoring by doctors who are familiar with CF are also critical, as individuals with CF will have a number of daily medications and medical treatments tailored to their needs. While there is no cure for CF, there are several treatment methods that significantly delay the decline in organ function.

The primary site of action of CF is the lungs. Medications includes high-dose antibiotics to prevent and treat infections, inhaled drugs to help open the airways, and DNAse enzyme therapy to thin mucus and make it easier to cough up.

Treatment also includes mechanical techniques such as exercise or percussion of the lungs to break up mucus and induce coughing. This is often accomplished with a vibrating vest that the patient wears for a period of time each day. These therapies can be extremely time-consuming for the patient, and one of the most difficult challenges for CF patients is finding the time to comply with prescribed treatments while balancing a normal life. As lung disease gets worse, oxygen therapy may be needed. Double-lung transplantation is sometimes an option late in the progression of the disease.

Most CF patients will also experience symptoms affecting the pancreas and gastrointestinal tract. Treatments include replacement pancreatic enzymes to aid in proper absorption of nutrients, a special diet high in protein and calories, and vitamin A, D, E, and K supplements. Complications of CF can include cystic fibrosis-related diabetes (CFRD), osteoporosis, and infertility, and these problems can be managed as well.

Cystic Fibrosis in Arab Populations

Although cystic fibrosis is not as common in Arab populations as it is in Europe and the United States, there are still very high rates of children born with CF in the Middle East. For every 100,000 live births, CF will affect approximately 7 babies in Oman and Qatar, 17 in Bahrain, 24 in Saudi Arabia, and 33-39 in Jordan. Cases of CF have also been reported in the UAE, Kuwait, Egypt, Lebanon, Palestine, Iraq, Iran, and Pakistan. Unfortunately, many of these babies are diagnosed after one year or more, and many die in childhood. A comprehensive neonatal screening program in Middle Eastern countries could identify babies born with CF in their first days of life and treat them sooner, thereby reducing morbidity and mortality of the disease in the region.

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Children with cystic fibrosis tend to have several nutritional issues. They might be unable to absorb nutrients from food efficiently. In addition, frequent infections leave them requiring foods with high calorie quotients. A high-fat high-energy diet is recommended for such children. High fat foods such as oil, butter, full cream milk, cream, mayonnaise and cheese are recommended. Always remember to compensate for the extra fat by taking extra pancreatic enzymes.