Cystic Fibrosis

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CYSTIC FIBROSIS

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WHAT IS CYSTIC FIBROSIS?

• Cystic fibrosis is an inherited genetic disease that affects the lungs and the digestive system, this condition is life threatening.

• The disease affects the cells that produce mucus, sweat and digestive juices.

• In cystic fibrosis a gene malfunction causes body secretions to become thick and sticky making hard for the body to get rid of any secretion.

• The secretions of the body don’t act as lubricants instead the secretions plug up the pancreas and the lungs.
GASTROINTESTINAL ISSUES

- Thick secretions from the pancreas block the exocrine movement of the digestive enzymes needed to absorb nutrients.
- The pancreatic ducts are totally plugged in serious advance cases of CF.
- The lack of digestive enzymes leads to malabsorption.
- Malabsorption leads to malnutrition making it hard for individuals to gain weight and developmental issues.
- CF individuals have difficulties absorbing various essential vitamins like A, D, E and K.
- In severe cases CF individual suffer of intestinal blockages due to thickened feces.
OTHER HEALTH ISSUES

• Having CF puts individuals at risk of diabetes 2

• Individual with CF can suffer from osteoporosis at young age due to the lack of absorption of the vitamin D which is involved in calcium and phosphate regulation.

• Infertility is common among people with CF however they are not sterile and they can reproduce with advanced reproductive techniques.
WHO CAN INHERIT CYSTIC FIBROSIS?

• Cystic fibrosis is most common in Caucasians of northern European descent. The condition occurs in one out of every 3,000 live births. About 25 percent of this ethnic group is a carrier of the condition.

• Cystic fibrosis is less common among persons of any other ancestry.

• An individual inherit cystic fibrosis when the father and mother pass two mutated genes to their offspring. The cystic fibrosis gene is located in chromosome 7.

• Cystic fibrosis is a recessive disorder. Which means that CF will only develop if there are two mutated genes from each parent, if the individual only carries one mutated gene then the individual is only a carrier of the gene of cystic fibrosis.
SYMPTOMS

- No bowel movements in the first 24-48 hours of life.
- Salty tasting skin.
- Failure to gain weight.
- Delayed growth.
- Watery stools that have foul smell.
- Weight loss.
- Recurrent respiratory infections.
HOW CAN CYSTIC FIBROSIS BE DIAGNOSED?

• A simple way to test for CF is to administer a sweat test. This will indicate the amount of salt in the sweat. 90% of people with cystic fibrosis have saltier sweat than a person without CF.

• Genetic test are also used to determine if an individual has CF. Doctors check for the mutated gene of cystic fibrosis located in chromosome 7

• Newborn screening. Newborn babies are tested using blood to determine if they have cystic fibrosis.
THE SWEAT TEST

- Measures the concentration of chloride and sodium that is excreted in the sweat.
- Two reliable positive results on two different days is a diagnostic for CF
- This test can be repeated as many times needed.
NEWBORN SCREENING

- Newborns can be easily tested for Cystic fibrosis with a blood test
- Elevated levels of Trypsinogen indicates the presence of CF.
- Screenings identify 10 percent of cases at birth.
- Most states and hospitals across the nation don’t screen for CF at birth since this diseases is so rare.
• Genetic testing is available for the most mutation of Cystic fibrosis are available but they are considered to be the most expensive of all the ways to test for CF

• The type of defective CF gene can affect the type of symptoms the individual experiences.

• Genetic testing can’t determine how severe the symptoms will be for the individual suffering from CF
TREATMENTS FOR CF

• Pancreatic enzymes to replace the ones that are not being produced.
• Vitamin supplements (A, D, E, K) these vitamins are especially needed in CF.
• Inhaled medicines to open the lungs airways
• Chest percussion
• Antibiotics in case of respiratory infection.
MANAGEMENT

• Lung transplantation becomes necessary in some cases for individuals with CF
• Exercise is a way to help the airways open up and is recommended to stay healthy
• Diet is important to meet the needs of individuals with CF
• Individuals with CF can eat greasy food to try to get fat from the food they eat since they have issues absorbing fats as well
LIVING WITH CYSTIC FIBROSIS

• New advance therapies and specialized care have permitted more patients to live a easier life.
• Children and adult are living longer over the last three decades.
• Today more tools and information are available to help patients and their families.
• 45 percent of the CF population is over the age of 18. Evidence that CF patients are living longer.
• There are more programs nationwide to support patients living with this disease.
GOALS TO CONTROL CF

• Prevent and control lung infections by exercising and taking medications
• Loose and remove sticky mucus from lungs using percussion techniques.
• Prevent blockages in the intestines by keeping a healthy diet
• Prevent dehydration by eating salty food and drinks that contain electrolytes
• Provide a balanced diet to keep a healthy weight and height
LIFE EXPECTANCY FOR CF

• There is no way to predict how long a person with CF will live but over the last three decades patients live longer than they used to before new medical techniques to keep the lungs clean.

• More of the 45 percent of people living with CF are 18 years older

• In 2009 the median predicted age of survival was in the mid 30’s.
FACTS ABOUT CYSTIC FIBROSIS

• The name comes from the fibrous scar tissue which develops in the pancreas.
• In 1938 American physician Dorothy H Andersen identified Cystic fibrosis disease.
• CF patients are most of the time hungry but even though they eat large quantity of food they still have troubles gaining weight and height.
• The genetic disease of Cystic fibrosis affect the whole body: lungs, endocrine system, pancreas, reproductive system, liver, digestive system.
• CF is a rare disease to inherit both of parents need to be carriers of the CF gene.
• Unaffected parents can still have kids that have cystic fibrosis even if the parents are only carriers
• Males and females are equally likely to be diagnosed.
CURE FOR CF?

- CF is commonly under diagnosed or missed in the majority of cases.
- Gene therapy is one most aspiring approaches to curing Cystic fibrosis.
- Gene therapy could replace the defective copies of the CF gene and replaced them with non-defective genes.
- That will result in stopping mucus from building up and being less sticky.
- So far no attempt have not successful.
IMAGES REFERENCES


