DISEASE

Cystic fibrosis is an autosomal recessive trait on chromosome 7. This disorder affects chloride transport resulting in abnormal mucus production. This lifelong illness usually gets more severe with age and can affect both males and females. Symptoms and severity differ from person to person. Cystic fibrosis is the most common fatal inherited disease among whites and the major cause of chronic lung disease in children. 50% of people are expected to live to be 30, but a majority die before age thirteen. 1:2000 whites have cystic fibrosis, 1:17000 blacks, 1:6000 live births, 1:2500 Americans, and 1:20 is a carrier.

The genes are inherited in pairs, with one gene coming from each parent to make the pair. Cystic fibrosis occurs when both genes have mutations. A person with cystic fibrosis receives one cystic fibrosis gene from each parent. The parents of a child, with cystic fibrosis, each carry one nonworking copy of the gene and one working copy of the gene. The parents are called cystic fibrosis carriers, and because they have one working gene they have no symptoms. Carrier parents have 1:4 chance to have a child who is a noncarrier of cystic fibrosis, a 1:2 chance to have a child who carries the gene, and a 1:4 chance with each pregnancy to have an affected child. If you have a son or daughter with cystic fibrosis, then you have a 1:1 chance of being a carrier. If you have a brother or sister with CF, you have a 2:3 chance of being a carrier. If you have a niece or nephew with CF, you have a 1:2 chance of being a carrier. If you have an aunt or uncle with CF, you have a 1:3 chance of being a carrier and a 1:4 chance if you have a 1st cousin with CF.

Cystic fibrosis affects the lungs in particular. The secretions are thick and sticky rather than thin and watery. This interferes with the removal of dust and germs. It can lead to lung infections and even chronic lung damage. Air passages become clogged with mucus and there is often widespread obstruction of the bronchioles. Expiration is especially difficult. More and more air becomes trapped in the lungs, which results in obstructive emphysema. Atelectasis can occur leaving small areas collapsed. Eventually the chest assumes a barrel shape. The right ventricle, which supplies the lungs, may become strained and enlarged. Clubbing of the finger and toes may occur due to the compensation response indicating the chronic lack of oxygen.

Cystic fibrosis affects the pancreas. The mucus clogs the duct and blocks the transfer of enzymes from the pancreas to the intestines. These enzymes are needed to break down food that is necessary for proper growth and weight gain. The mucus in the digestive tract blocks the absorption of necessary nutrients. This is why there is often no weight gain despite good appetites. This can be associated with failure to thrive. The buttocks and thighs atrophy or waste away due to the fat disappearing from main deposit sites. People usually have light colored stools. There is also decreased blood cholesterol due to the poor absorption of fats from the intestine.

Cystic fibrosis can also affect the reproductive systems. Men are usually sterile due to the mucus blockage or absence of the vas deferens. Women usually have difficult conceiving, because the mucus interferes with the passage of sperm.

Cystic fibrosis is usually diagnosed in childhood. Mild cases may not be detected until adulthood. Common symptoms include chronic cough, wheezing, cyanosis, difficulty breathing, irritability, excessive mucus production, sinus infections, nasal polyps, recurrent pneumonia, poor growth, frequent loose foul-smelling stools, enlarged fingertips, and skin that is salty to the taste. The sweat test is usually used to detect high levels of salt. More than 60m Eq/L of chloride in sweat up to age 20 is diagnostic of CF when 1 or more criteria are present. Levels of 40-60 are highly suggestive. Direct genetic testing or reverse dot-blot can also be used. Amniocentesis is performed between weeks 15-22. Chorionic villus sampling (CVS) can be used to take a piece of placental tissue between weeks 9-12. Labs are also used in diagnosing CF. There is decreased

pancreatic enzymes trypsin, lipase, and amylase. Absence of trypsin alone is indicative of CF.

One complication of CF is a rare condition known as meconium ileus. The intestine of the newborn becomes obstructed with abnormally thick meconium due to the absence of pancreatic enzymes. The intestine can rupture resulting in shock. Signs and symptoms develop within hours after birth and include absence of stools, vomiting, and abdominal distention. X-rays are used to confirm this and surgery is used to correct the problem. The death rate is high including premature births and most who survive will manifest CF. Nurses in the nursery must be on guard for early detection. Rectal prolapse occurs in infants and children due to poor muscle tone in the rectal area and excessive leanness. It may be related to difficulty passing the frequent bulky stools. Fecal impaction and intussusception or telescoping of the bowel are other bowel complications in infants and toddlers. The liver becomes hard, nodular, and enlarged with progression. There is often edema in the extremities. There may be damage to the eye as a result from swelling and inflammation of the optic nerve. The retina may also hemorrhage. Improper lung function can cause heart strain resulting in death. Osteoporosis results from poor utilization of the fat-soluble vitamin D, which is necessary for proper calcium metabolism. The bones become porous and brittle. Deficiency of vitamin A occurs from the body's inability to absorb fats from which vitamin A is obtained. Sexual development may be delayed and women may experience secondary amenorrhea during exacerbations.

There is no cure to date. They have made progress towards a cure. They isolated the gene at U of M in 1989. This was the first human genetic disease to be cloned. They thought it was linked to the trace mineral Boron. Copies of the normal gene were made in 1990. They realized that the protein product of the gene, transmembrane conductance regulator or CFTR, influences chloride transport but were unsure how that led to CF. Gene therapy was experimented in 1993 along with the first drug called Pulmozyme. Ibuprofen was ruled effective in decreasing lung problems in children in 1995. They ruled in 1996 that the bacteria killing agent doesn't function in people with CF due to the excessive salt outside the epithelial cells. This allows pseudomonas and staphlococcus to cause chronic bacterial infections.

Treatment of CF includes taking Pancrease, an oral enteric coated pancreatic enzyme preparation, with meals and snacks to help aid in digestion. Fluids should be increased and liberal amounts of salt intake. Fluids are forced to prevent dehydration from frequent stools and excessive sweating. Salt tablets are often used in older children. Frequent high calorie meals and snacks are used to help maintain weight. Don't pile food on a child's tray. Make it attractive and the size should correlate with the child's size. Make mealtime a social time and encourage the child to eat. If he is in a private room, then stay with him or have someone in the room. The nurse feeding an infant must be calm and unhurried. Calories should be increased by 50% along with protein. Fat intake should decrease. Supplement vitamins A,D, and E are used double the recommended daily dose. Skim milk is often added to formula in infancy. Vitamin K is often given. Complex sugars should decrease and simple sugars should increase. Many doctors allow the child to eat what he wants and just increases pancreatic enzymes to provide a "normal" atmosphere. Weights are taken daily. Respiratory relief comes from postural drainage, pursed-lip breathing, general exercise to stimulate cough, deep breathing and coughing exercises, bronchodilators, expectorants, antibiotic use, intermittent aerosol therapy, and the controversial mist tent therapy. Injections should be avoided due to the excessive leanness but if necessary the sites must be monitored and alternated. Pay special attention to the skin. Cleanse the diaper area after each bowel movement. Ointment is often used to protect skin from stools. Expose the buttocks to air when a rash occurs. Pay special attention to the bony areas in order to prevent decubitus ulcers. Change position frequently due to the lack of fat and muscle. This helps to prevent skin breakdown and pneumonia. Don't leave the person staring at a blank wall. Air deodorant is advisable to prevent

lingering of offensive odors. Light clothing is recommended to prevent overheating. Loose clothing allows freedom of movement. Good oral hygiene is necessary especially due to dietary deficiencies. Make sure oral hygiene is also performed after postural drainage. Make sure immunizations are up-to-date and the influenza vaccine is also recommended. CF patients are usually in isolation to help prevent secondary infections. Allow for rest. This is very important as is prevention as a whole.

CF is hard on children. They often feel different from other children and tire easily. It is hard for them to accept restricted activity. They get really embarrassed about their stools. Give the child straight forward answers regarding his illness to prevent further anxiety. Uninvolved diagrams can be helpful. If he understands, he is more apt to be cooperative. Visiting hours should be flexible for parents. You should be considerate and encouraging. Showing undue concern can, however, cause the child to exaggerate and be demanding for attention.

Parents may have knowledge deficit and may need a lot of teaching and explanation. One of the misconception parents have is that their child's intelligence is greatly decreased. Intelligence is not affected. Parents often feel guilty, since this is an inherited disease. The child spends the majority of his time at home due to this lengthy illness. The child is also hospitalized for complications although stays are short to prevent exposure to other infections and illnesses. This puts a financial, physical, and emotional burden on the family. When do the parents find time for each other, themselves, or other children? How do they distribute their time and energy equally and fairly? Parents need encouragement and reassurance. They also need explicit instructions. Parent groups can help along with the Nat'l CF Research Foundation and the 1-800-FIGHT-CF hotline. Parents usually need help from a social worker and financial help for special equipment. Insist parents to get help from other family members or friends and encourage them to get away from it all periodically. Alarm clocks can remind them of medication times.