FACT SHEET
Healthcare Provider

Cystic Fibrosis (CF)

Description:
Cystic Fibrosis (CF) is an autosomal recessive disorder. CF is caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) protein, a complex chloride channel found in all exocrine tissues. CFTR controls the flow of water and certain salts in and out of the body's cells. As the movement of salt and water in and out of cells is altered, mucus becomes thickened. The thickened mucus can affect many organs and body systems including the lungs, sinuses, pancreas, liver, intestine, and reproductive tract, and to increased salt content in sweat gland secretions. This was considered a childhood disease in the past with the median survival age of 8. As of 2005, the median age is approximately 35.

General Population Incidence:
CF occurs most commonly in Caucasians, in about 1 in 2,500 Caucasian live births. CF occurs in about 1 in 8,500 for Hispanics, 1 in 15,000 for African Americans, and 1 in 31,000 for Asians. CF has also been reported in other ethnic groups, including Native American, Middle East, and Pacific Islander populations.

Symptoms:
- Chronic diarrhea
- Poor growth/ failure to thrive
- Foul-smelling stools
- Meconium ileus
- Rectal prolapse
- Abdominal pain
- Chronic pancreatitis
- Frequent episodes of wheezing
- Persistent cough
- Recurrent pneumonia
- Salty-tasting skin
- Chronic sinus infection
- Nasal polyps

Newborn Screening Technology:
CF is detected by an immunoflourescent assay (IFA) technique for Immununoreceative trypsinogen (IRT). IFA first measures the level of IRT in the blood. For infants whose IRT is either the highest 5% of results or above 60 ng/lml, the test result is confirmed by repeat testing. The interpretation of generated IRT result is then based on the infant's age. If meconium ileus is present, then the IRT could show a false normal result.

Diagnosis:
Diagnosis is based upon compatible clinical findings with biochemical or genetic confirmation. The laboratory evidence for CFTR dysfunction includes either a sweat chloride above 60 mEq/L for two tests, identification of two CF mutations, or an abnormal nasal potential difference measurement. Compatible clinical findings include presence of typical clinical features, a history of CF in a sibling, or a positive newborn screening test. It is important to note that normal sweat chloride does not absolutely rule out CF. An important fact is that 10% to 15% of patients with Cystic Fibrosis are pancreatic sufficient.

Treatment:

- Regular outpatient monitoring with a multi-disciplinary team including physician, nurses, dieticians, respiratory therapists, social workers, and school health personnel (e.g., school nurses), at least every 3 months, preferably by a Cystic Fibrosis Center
- Management of problems that cause lung obstruction, which may involve:
  - Chest physical therapy (to help loosen and clear lung secretions; this may include manual hand therapy, a therapy vest, and devices such as a percussor or flutter, which vibrate the chest wall and loosen secretions).
  - Exercise (to loosen mucus, stimulate coughing, and improve overall physical condition).
  - Medications (to thin, reduce mucus and help breathing, such as Deoxyribonuclease, bronchodilators, and anti-inflammatory medications).
  - Antibiotics (to treat infections).
- Management of digestive and weight problems, which may involve:
  - Appropriate diet.
  - Pancreatic enzymes to aid digestion.
  - Vitamin supplements.
  - Treatments for intestinal obstructions.
- Psychosocial support (dealing with issues such as independence, sterility and sexuality, financial issues, and relationships).

Immunizations:

Immunization schedules should be followed to ensure protection from all other childhood diseases. It is crucial that patients with Cystic Fibrosis receive the Influenza vaccine each year.

Growth and development:

It is crucial to monitor all growth parameters on a regular basis.

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