Screening for Cystic Fibrosis

Information for parent

What is cystic fibrosis?

Cystic fibrosis (CF) is a genetic condition that affects many organs in the body by building up a sticky secretion in the body; especially the lungs, pancreas and sweat glands. These will result in respiratory problem, incomplete digestion and growth.

Who will have CF?

CF is an inherited condition that occurs when a particular protein "cystic fibrosis transmembrane conductance regulator (CFTR) protein" is either missing or not working well. Everyone has a pair of CFTR genes that make the CFTR protein.

If both mother and father are carriers of the abnormal CFTR gene, there will be a 1 in 4 chances that their offspring will be affected by CF. CF is more common among the Caucasian population.

Screening test for CF

An enzyme called immunoreactive trypsinogen (IRT) is measured in baby's dried blood spot. If IRT is elevated, analysis of baby's CFTR gene will follow.

What to do if the screening is positive?

A positive CF screening test means the child may have CF. Further diagnostic tests including genetic testing and a "sweat' test are needed for confirmation. You will be contacted by medical personnel if the screening test is positive.

What is the outcome of CF patient?

Recent medical advances have improved the outcome of CF patient significantly. Early diagnosis and treatment are the important steps to ensure a better outcome.

JOSHUA HELLMANN FOUNDATION FOR ORPHAN DISEASE

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