

CYSTIC FIBROSIS CARRIER SCREENING

Purpose of carrier screening:

- To see if a couple is at an increased risk for giving birth to a child with CF.
- CF cannot be treated before birth, so the information would be for preparation or termination.
- CF carrier screening is completely optional. It is a personal decision.

What is CF?

- It is a genetic condition causing lifelong breathing and digestion problems.
- The severity varies and cannot always be predicted based on the DNA test result.
- It is autosomal recessive, so it occurs in both males and females and both parents must be carriers to have an affected child.
- Carriers do not have any health problems.

Meanings and limitations of results:

- CF carrier screening cannot detect everyone who is a carrier.
- The results would provide a couple with a new risk for giving birth to a child with CF. This new risk figure assumes correct paternity and no family history. This new risk figure only applies to the current couple and may change with other partners.
- CF carrier screening may find that a male offspring may have an increased risk for one type of infertility.
- If both partners are found to carry CF, then they are offered the option of prenatal testing via amniocentesis or chorionic villus sampling.

Currently our local hospitals – York, Memorial, Gettysburg, Hanover, Lancaster General, and Women and Babies – test all newborns to see if they are affected with CF on the expanded Newborn Screening Panel.

Features of CF:

Chronic pulmonary disease
Pancreatic insufficiency
Failure to thrive, meconium ileus in infants
Sinusitis, nasal polyps
Infertility in males
Death within the 30's

Treatment of CF:

Pulmonary therapy (antibiotics, bronchodilators, mucolytics, chest PT, lung transplant)
Digestive therapy (pancreatic enzyme replacement, dietary supplements)
Reproductive therapy (genetic counseling, sperm aspiration, ICSI)
Gene therapy is not currently available.

Information provided by the genetic counselor at Maternal Fetal Medicine, York Hospital.

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