Fertility Design
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Cystic Fibrosis

Cystic fibrosis is an inherited disorder caused by the presence of mutations in both copies of the gene for the cystic fibrosis transmembrane conductance regulator (CFTR) protein. If two copies of the defective CF gene will be identified, one copy from each parent, then the child is ill. Cystic fibrosis is the most common lethal genetic disease in white populations. Cystic fibrosis is not contagious but hereditary. The birth frequency of affected children in Greece is 1/2500. If two carries of the CF get married, the possibility for their children to get infected is 1 to 4 or 25%. Carriers are healthy, without any clinical symptoms, while the genome is quite bigger and has many possibilities for mutations. Cystic fibrosis signs and symptoms vary, depending on the severity of the disease.

Description of CF contagion

In cystic fibrosis, a defect (mutation) in a gene changes a protein that regulates the movement of salt in and out of cells. The result is thick, sticky mucus in the respiratory, digestive and reproductive systems, as well as increased salt in sweat. It is important to mention that there have been numerous reports from around the world mentioning mutations in the gene of chromosome 7 known as *CFTR* (cystic fibrosis transmembrane conductance regulator) as well as over 1.000 mutations that affect the CFTR gene in different ways. Due to this fact of multiple and frequent mutations in populations/ethnicities, it is difficult to categorize and number the mutations that could be controlled globally. According to World health Organization, for the reasons above, we apply the monitoring for mutating gene of the carriers, handling at least the 85% of the Greek mutant population. The most common mutation OF CFTR is the F08del and is detected in 54% of the Greek CF carriers. Cystic fibrosis is transmitted to a child when both parents carry the recessive gene but do not have the disease. When such a couple has children, there is a 25 percent chance that one of their children will develop cystic fibrosis.

Note: for the examination of CF, in case of pregnancy, we don't examine directly the embryo, in contrast with the highest amount of genetic diseases.

Diagnosis

Diagnosis for CF can be divided into four stages:

- **Prenatally:** Parental prenatal screening is useful in identifying the 4% of the population that may carry a single copy of the CF allele. In pregnant women amniocentesis is the first choice during week 18 because the amniotic fluid surrounds the fetus can be tested for fetal intestinal enzymes (18-20 weeks), so a sample of amniotic fluid is extracted from the amniotic sac (the protective covering around the fetus) and analyzed.
- **Childhood /adulthood:** The most common test is the electrolyte sweat test. This test measures the amount of electrolytes (sodium [salt], potassium and chloride) in a person's sweat. This is done by applying a chemical (called pilocarpine) to the forearm and using a mild electric current to cause the area to sweat. If higher than normal amounts of sodium and chloride are evident, CF is present.
- **Genetic Testing:** In families with CF history, brothers, sisters, and first cousins of the CF patient should be tested to see if they carry a defective gene, especially if they seem to have a chronic lung or digestive problem.CF tests involve taking any type of tissue, such as white blood cells or cells located in the inside of the cheek, and analyzing the samples for mutant genes. A common molecular control includes the detection of 36 mutations in CF gene, which are developed in the 74% of the Greek population. can also be accomplished an expanded genetic control ,because it covers the 85% of mutations or even more an examination to the hole gene of CF can be done.

Infertility

Cystic fibrosis (CF) is one of the autosomal recessive diseases, caused by mutations in a gene known as cystic fibrosis transmembrane regulator (CFTR). The majority of adult males with CF (99%) is characterized by congenital bilateral absence of vas deferens (CBAVD). CBAVD is encountered in 1-2% of infertile males without CF. Females with CF are found to be less fertile than normal healthy women. In females with CF, delayed puberty and amenorrhea are common due to malnutrition. CFTR mutations are also associated with congenital absence of the uterus and vagina (CAUV). The National Institutes of Health recommend genetic counseling for any

couple seeking assisted reproductive techniques with a CF male or obstructive azoospermia which is positive for a CF mutation.

General information

- 1/700 couples are carriers of the disease and have possibilities of 25% to be their children infected
- When the mother has been tested ,and there are not any indications for CV, she has 1/17000 possibilities to birth an infected child
- If it has been discovered in mother the mutant gene CFTR, without fathers screening, the possibilities to give birth to an infected child is 1/100.But if the father has been detected with:
    - 85% without mutations ,the danger for the child to born infected is 1/700
    - 99% without any mutations, the danger for the child reduces to 1/4000.