Testing for Cystic Fibrosis

before and during pregnancy

NEWCASTLE ULTRASOUND *for* WOMEN

NUFW is a leading centre for peri-conceptual care. NUFW are able to offer CF-carrier testing as they have a Clinical Geneticist, a Counsellor and six other specialists in foetal maternal medicine and infertility who can provide expert advice on the complex issues surrounding Cystic Fibrosis.

What is Cystic Fibrosis?

Cystic fibrosis (CF) is a common inherited condition, affecting one in every 2,500 babies born in Australia. CF mainly affects the lungs and digestive system, due to an alteration (mutation) of the genes responsible for production of a salt transporting protein. As a result, thick mucus is produced in the lungs and the gut, leading to recurrent respiratory infections and poor absorption of nutrients.

There is no cure for this disorder and treatment involves daily physical therapy and medication. Life expectancy varies depending on the severity of the illness. Some people will not survive past teenage years and others may live to around 50 years of age. Infertility in men is common due to absence of the sperm transport tubes; the vas deferens.

Interpretation of Results

There are a number of possible test outcomes

Cystic fibrosis follows an autosomal recessive inheritance pattern. This means that if you and your partner are both found to be carriers of a mutation, then there is a 25% chance of a child being affected by CF in every pregnancy. There will also be a 50% chance of a child being a carrier like yourself (and unaffected), and a 25% chance the child will neither be a carrier nor affected. If one partner is found to be a CF carrier but theother is not, then there is a 50% chance in every pregnancy that the altered gene will be passed on to the child, who will then also be a carrier but will not be affected by CF. The risk of having a child affected by CF is greatly reduced when only one parent carries a detectable mutation. However, the risk is never zero, as not all CF mutations can be screened for. Similarly, if neither partner is found to be a carrier on the simple test, the risk is not zero but is significantly reduced for both being a carrier or having a CF-affected child. The reductions in risk are larger again for a negative multi-test result.

What if I Am Found to Be a CF Carrier?

If you and your partner are both found to be carriers of a CF mutation then you will be offered prenatal diagnosis in each of your pregnancies. Prenatal diagnostic tests involve analysing the DNA from a small sample of placenta (obtained by chorionic villus sampling) or amniotic fluid (obtained by amniocentesis) to determine whether the mutations have been passed on to the fetus.

What is the Chance that I May Carry a Gene Mutation?

This varies greatly. We know that in Australia 1 in 25 people from Caucasian (Northern European) ancestry will be a carrier of the mutated gene. The mutation is also common in those from Southern European and Middle Eastern populations, but is rarely present in those of Oriental or Asian background. If there is a family history of CF, then the chance of carrying a mutated gene may well be increased. For example, if you have a sibling affected by cystic fibrosis, you will have a 2 in 3 chance that you are a carrier.

How Can I Tell if I am a Carrier?

Carriers are usually perfectly healthy and are unaware that one of their genes has undergone mutation. Carrier status can only be identified by specialised analysis of a sample of your DNA. However, before embarking on carrier testing, it is important to receive appropriate counselling about the condition, the chances of being a carrier, the nature of the CF test, the limitations and costs of CF testing, the option of prenatal testing if both parents are carriers, and the options available if a fetus is diagnosed with CF.

At Sydney Ultrasound for Women (SUFW), these complex issues will be discussed with you by one of our experienced staff, which includes two Clinical Geneticists, a Genetic Counsellor and five other specialists in prenatal diagnosis.

How is Cystic Fibrosis Inherited?

Everyone has two copies of the salt transporter gene. You inherited one copy of the gene from your mother and the other copy from your father. In turn, you and your partner will each pass on just one of your two genes to your children. If both copies of your gene are unaltered, then you will only be able to pass on normal genes to your children. If one of your genes is mutated and the other gene unaltered, then you are a carrier of the CF mutation. Carriers are not affected by CF but will have a 50% chance of passing on the mutated gene to their children. It is only when both parents are carriers of a CF mutation that there is a chance that a child may receive a mutated gene from both of them. When this occurs, the child has two mutated genes and will be affected by cystic fibrosis.

Cystic Fibrosis Carrier Testing

Carrier testing is available to all couples who are concerned about having a CF-affected child, and is particularly important in those with a family history of CF. Very ccurate carrier risk assessment is possible if the family mutations are known. Testing is ideally undertaken prior to establishing a pregnancy, but is also readily performed early in pregnancy. It is not possible to test for all the mutations that may result in cystic fibrosis.

However, the mutations involved in up to 90% of those affected by CF can be tested for. Furthermore, certain mutations are more common in particular ethnic groups, and recommendations for testing will take ethnicity into consideration.

There are two options available for CF carrier testing, the simple test and the multi-test.

1. The simple test screens for one mutation only, Delta F508, but this mutation accounts for 75% of those affected by CF. This test is particularly suitable for couples of Caucasian ancestry, as the prevalence of the tested mutation is highest in this group.

2. The multi-test screens for 31 different mutations, which account for 90% of those affected by CF.

Testing is done on a sample of DNA extracted from a buccal cell sample (mouth swab). Initially, either the woman or the couple is tested depending on the urgency of the result. Results usually take two to three working days, and you will be notified by phone or letter depending on the result. A laboratory fee is charged for CF testing, with the multi-testing being more expensive. There is no Medicare rebate for this laboratory test.

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