Cystic Fibrosis Carrier Testing

- Cystic fibrosis carrier testing for couples is a screening test for the common CF mutation called F508del
- 1 in 25 people in the Caucasian population are CF carriers
- A simple mouthwash will identify 75% of people who are carriers
- Carriers do not have symptoms of CF
- If both partners are carriers, there is a 1 in 4 chance in each pregnancy, of having a child with CF.
- If one partner is a carrier of the F508del mutation, extended mutation testing is offered to the other partner.
- If neither Caucasian partner carries the F508del mutation, the chance of having a child with CF is 1 in 40,000.
- Couple testing is offered prior to or in the early stages of pregnancy.
- Test results take 2-3 weeks

Appointments can be made during working hours by contacting Hunter Genetics - (02) 4985 3100.
A referral from your doctor is required.

Hunter Genetics
Cnr Turton & Tinonee Roads
(P.O. Box 84)
WARATAH NSW 2298
Tel: (02) 4985 3100
Fax: (02) 4985 3105
Email: hunter.genetics@hnehealth.nsw.gov.au
# What is Cystic Fibrosis

Cystic Fibrosis (CF) is the most common life-threatening inherited disease that affects Australians. CF causes the body to produce thick sticky mucous. This mucous blocks the tiny airways in the lungs and traps bacteria, causing repeated infections and lung damage. The mucous can also cause problems with digestion. The mucous may block the pancreatic duct, preventing enzymes needed for food digestion from flowing freely from the pancreas to the intestine. Enzyme supplements may need to be taken with each meal.

CF is a serious but variable disease and requires regular and continuous treatment. The severity of the lung disease is the key to the quality and length of life.

## How common is CF?

One in 2500 babies in our population are born with CF.

## How do you inherit CF?

In almost every cell of our body we have a set of 46 chromosomes. These chromosomes come in pairs. We inherit one copy of each pair from our mother and the other from our father. Genes are found on chromosomes. Genes are instructions used by the body to make all the proteins needed for normal growth and development. Sometimes mistakes occur in a gene and the gene no longer works correctly. These changes are called mutations. The gene involved with CF is found on chromosome 7. A person with CF has 2 non-working copies of this gene. They usually have inherited a non-working copy from each of their parents.

## What is a CF carrier?

A CF carrier is someone who does not have CF but carries one non-working copy of a CF gene and one working copy of the gene. Carriers do not have any symptoms of CF.

One in 25 people in the Caucasian population are CF carriers. Most CF carriers have the CF mutation called F508del.

If a person who is a CF carrier has a partner who is also a carrier, they have a 1 in 4 chance in each pregnancy of having a child with CF.

## What does carrier testing for couples involve?

If you are interested in having carrier testing for CF, you will need to get a referral from your doctor.

Appointments can be made by contacting Hunter Genetics on (02) 4985 3100.

You will be seen by our trained staff who will record your family history and discuss CF and carrier testing in detail. If you wish to proceed with carrier testing, a consent is completed and a simple mouthwash taken.

Testing for F508del costs $50.00 per person. An invoice will be sent to you from the laboratory.

Test results take two to three weeks.

## Who should have testing?

Couples who are planning a family or are in the early stages of pregnancy (before 15 weeks) are encouraged to consider carrier testing for CF.

Most children born with CF do not have a family history of the disease. This is because people can be a carrier of a CF mutation and not know it. Carrier testing can provide you with this important information.

If you have a family history of CF, or have a relative who has been identified as a CF carrier, then your risk of being a carrier can be greater. Let your doctor know if this is the case.