WANT TO BE TESTED ?

Take this pamphlet to your doctor to arrange testing.

TEST NAME: CF mutation screen

SPECIMEN : 5 mL blood (EDTA).

SEND TO:

The National Referral Laboratory,

4th Floor, Rogerson Building,

Women's and Children's Hospital.

72 King William Rd.,

North Adelaide, 5006.

INCLUDE ON THE REQUEST FORM:

- The name and date of birth of the person who has (or is a carrier of) CF. This information is important for the laboratory doing the carrier testing.
- The relationship of the person being tested to this person (include which side of the family they are on) eg mother, mother's sister, cousin on dads side of the family etc.

To arrange an appointment to see a genetic counsellor contact:

South Australian Clinical Genetics Service
Women's and Children's Hospital
Phone : (08) 8161 7375
Fax: (08) 8161 6088
Email: sacgs@mail.wch.sa.gov.au

For information about testing contact:

National Referral Laboratory
Women's & Children's Hospital
Phone : (08) 8161 7294
Fax: (08) 8161 7100
Email: michael.fietz@adelaide.edu.au

For further information on CF see:
www.cfsa.org.au

CHILDREN, YOUTH & WOMEN'S HEALTH SERVICE

Government of South Australia | Women's & Children's Hospital

Women's & Children's Hospital
72 King William Road
North Adelaide
South Australia 5006
Phone (08) 8161 7000
Fax (08) 8161 7459
Web: http://.wch.sa.gov.au

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CARRIER TESTING FOR CYSTIC FIBROSIS

Information for people who may be carriers of Cystic Fibrosis

WHO SHOULD BE TESTED ?

- People with a relative or partner with Cystic Fibrosis.
- People with a relative or partner who is a *carrier* of Cystic Fibrosis.
- Men with certain forms of fertility problems (including absence of the vas deferens).
- Pregnant women (and their partners) who have "echogenic gut", detected on ultrasound.
- Parents whose baby is screened at increased risk of Cystic Fibrosis on newborn screening.
- Couples who are cousins.

CYSTIC FIBROSIS – SOME QUICK FACTS

- Cystic fibrosis (**CF**) is a serious inherited disorder mainly affecting the lungs and pancreas.
- 1 in 2,500 babies born *has* CF.
- Most babies who have CF and some babies who are carriers of CF are detected on newborn screening.
- A sweat test can be used to confirm the diagnosis of CF in babies, children and adults suspected of having CF.
- Both parents must be carriers to have a child with CF.
- 1 in 25 people are *carriers* of CF.
- Some milder forms of CF can cause infertility in males and reduced fertility in females.
- Babies with "echogenic gut" detected on ultrasound during pregnancy may be at risk of CF.
- If you are related to your partner, you may choose to have carrier testing for CF to check whether you are both carriers.

CARRIER TESTING FOR CYSTIC FIBROSIS

What does it mean to be a carrier of cystic fibrosis?

Being a carrier of cystic fibrosis will not affect your health.

If I am a carrier, can my children be carriers?

Yes. If you are a carrier, each of your children has a 1 in 2 chance of being a carrier.

How can I be tested to see if I am a carrier of cystic fibrosis?

A genetic test is done on a small sample of your blood at the Women's and Children's Hospital. There is no charge for this test if you live in South Australia.

Can this test always detect carriers?

If you have a family history of CF, the laboratory will look at your test result in combination with all available family information to determine if you are a carrier.

If you have no family history, the test can detect 4 out of every 5 carriers.

How long does it take to get results?

2-3 weeks.

If I am a carrier, should my partner be tested?

Yes. If you and your partner are both carriers, you have a 1 in 4 chance of having a baby with cystic fibrosis. If you are pregnant or planning a pregnancy, it is important to know if you are a carrier.

What does it mean if I am a carrier and my partner's carrier test is negative?

If only one parent is found to be a carrier there is still a *small* chance of having a child with cystic fibrosis (about 1 in 500), because testing cannot exclude the hundreds of rare genetic mistakes that cause CF.

What happens if I plan to have children with a different partner?

If you are a carrier, you should have your new partner tested as well. If your new partner is a carrier, you have a 1 in 4 chance of having a child with cystic fibrosis.

If my partner and I are carriers, can we have a test during pregnancy to see if our baby has cystic fibrosis?

Yes. See your doctor or contact the South Australian Clinical Genetics Service. Discuss testing ***before*** planning a pregnancy. If you are already pregnant, discuss testing with your doctor or the South Australian Clinical Genetics Service ***as soon as possible***.