



## INFORMATION FOR PARENTS/CARERS FOR CYSTIC FIBROSIS

### What is the Newborn Screening Programme?

Your baby had a blood sample taken from the heel at about three days of age. The blood was absorbed into a special card, sent to the NSW Newborn Screening Laboratory in Sydney where it was tested for several disorders. Cystic fibrosis (CF) was one of the disorders.

### What is Cystic Fibrosis?

Cystic fibrosis is one of the commonest inherited diseases affecting Australian children. One in every 2,500 babies, male or female, will be born with CF each year.

CF is a condition in which the body's secretions, especially in the lungs and gut, are much more sticky than usual. These thick secretions encourage infection in the lungs, and abnormal secretions from the pancreas frequently make food digestion and absorption incomplete. The baby may have large bulky smelly bowel actions.

CF is an autosomal recessive genetic disorder. This means that each cell in the body carries two copies of each gene, one inherited from each parent. In all of us, there are "mutations" in a few of the genes. For a recessive gene mutation to result in a particular disorder both copies of the gene must carry a mutation.

People who carry a single CF gene are called carriers of CF. One person in every 25 is a carrier of CF. It is important to remember that carriers of CF are healthy and do not develop CF, but will require genetic counselling before having children.

### What tests are done to find CF?

Two tests may be done on the blood sample. The first test will show whether or not the baby has a high level of a protein called immunoreactive trypsin (or IRT) from the pancreas. In those babies

who do have a high level, a second test, a DNA test, shows whether the baby carries any copies of the most common mutation associated with CF called  $\Delta F508$ . Babies with two copies of the  $\Delta F508$  definitely have CF.

**Our doctor has told us that our baby has one copy of the mutation  $\Delta F508$  and could have CF and must have a further test. What does this mean?**

Your baby has only about a **one in five chance** of having CF. This means that out of five babies who are found by the laboratory to have a high level of IRT and one copy of the  $\Delta F508$  mutation, only one baby will have CF. It is most likely that your baby has one copy of the  $\Delta F508$  mutation, and one entirely normal gene and is a healthy carrier of CF the same as one parent.

However, your baby could have one copy of the  $\Delta F508$  gene mutation, and also one copy of another rare CF mutation, which we do not test for directly. There are about 800 rare CF mutations. If this occurs your baby could have CF. Because of this possibility your baby must have further investigations to exclude CF. One of these investigations is a sweat test.

### What is a sweat test?

Your baby's sweat is analysed for salt content. Usually the sweat from your baby's arm or leg is collected on a pad. Sweating is encouraged by a chemical called pilocarpine which is applied to your baby's arm or leg. The test causes only a minor amount of discomfort and takes about an hour to complete. However, because the baby's skin heats up over a small area during the test, there is a risk that the skin will become red.

Your baby's result will be ready on the day of the test. On rare occasions not enough sweat is

produced to give a result. This does not indicate an abnormal result. The test will have to be repeated.

### **When can our baby have a sweat test?**

At around four to six weeks of age. This depends on the sweat testing laboratory. The test must be done at a laboratory which regularly performs sweat tests. This may mean that the most appropriate laboratory is in a children's hospital. You may need to travel to have a sweat test.

### **What happens if our baby's sweat test is normal?**

The doctor will tell you that your baby's sweat test is **NEGATIVE**. This means that the salt content in your baby's sweat is normal and your baby almost certainly does not have CF.

If your baby does not have CF, your baby is a healthy carrier of a single gene mistake. However, it is very important that both parents visit a genetic counsellor to discuss further testing of each parent. Either you or your doctor can arrange this visit (*see back page for telephone numbers of counsellors*).

It is best if both parents are tested for the most common gene mutation  $\Delta F508$ . The result could be important if you wish to have more children. Since your baby carries a CF gene mutation, one parent will certainly carry a CF gene mutation also. If by chance both parents carry a CF gene mutation, it is possible (one in four chance with every pregnancy) that you could have a baby with CF. Prenatal testing is available for CF. All of these issues will be discussed with you by the genetic counsellor.

### **Our baby has only one copy of the $\Delta F508$ mutation but the sweat test is **POSITIVE**. Does our baby have cystic fibrosis?**

Yes. The sweat test is a diagnostic test for CF and is very unlikely to be incorrect if it has been performed at a laboratory which regularly performs sweat tests. Both you and your partner carry a CF gene. One of you carries the mutation  $\Delta F508$  and the other carries one of the rarer mutations which we do not test for directly. Your baby must have inherited both CF genes, and therefore has cystic fibrosis.

### **Our doctor has told us that our baby has two copies of the mutation $\Delta F508$ and has cystic fibrosis. Could this be wrong?**

No. What your doctor has told you is correct. The laboratory tested your baby's blood on two different days and both times it has shown two copies of the mutation  $\Delta F508$ . This is evidence of CF.

### **Our baby has been diagnosed to have CF but is quite well - what are the symptoms of CF?**

Your baby may develop a cough or wheeze, be slow to gain weight or have frequent, bulky, smelly bowel actions. Your baby may also sweat a lot in hot weather and the skin may taste more salty than normal.

### **Our baby has been diagnosed to have CF. What should happen now?**

Your doctor will make an appointment for your baby at your closest CF clinic. The major clinics are based in Sydney and Newcastle, so if you live in the country you will need to travel to visit the clinic for the first time. Probably after that your family doctor or paediatrician will look after your baby, and occasional visits to the CF clinic may be all that is necessary. If you live near the border of another state it may be easier for you to go to a CF clinic in Brisbane, Adelaide or Melbourne.

### **Will our baby get very sick?**

Because CF has been diagnosed early, your baby has a much better chance of keeping well. Recent medical and scientific advances have greatly improved the outlook for babies with CF.

### **Where can we get further information about CF?**

The CF Foundation has booklets, pamphlets and videos that may be of help to you.

Cystic Fibrosis Foundation  
51 Wicks Road  
**NORTH RYDE NSW 2113**  
Phone: (02) 9878 2075.

If you need any further information from the NSW Newborn Screening Laboratory please telephone (02) 9845 3255/3659.

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## Cystic Fibrosis Clinics in New South Wales

### SYDNEY AND NEWCASTLE AREA

- Westmead:** The Children's Hospital at Westmead  
Cnr Hawkesbury Rd & Hainsworth Street  
WESTMEAD NSW 2124  
Ph: (02) 9845 2525 clinic appt.  
Fax: (02) 9845 3283 sweat test appt.
- Randwick:** Sydney Children's Hospital  
High St  
RANDWICK NSW 2031  
Ph: (02) 9382 1470 clinic appt.  
Fax: (02) 9382 1484/1485 sweat test appt.
- Newcastle:** John Hunter Hospital  
New Lambton Heights NSW 2305  
Ph: (02) 49 213 750 clinic appt.  
Fax: (02) 49 214 405 sweat test appt.

## Genetic Units in New South Wales

The following telephone numbers will help you to reach a genetic counsellor:

### SYDNEY AND NEWCASTLE AREA

- Westmead:** The Children's Hospital at Westmead  
Cnr Hawkesbury Rd & Hainsworth Street  
WESTMEAD NSW 2124  
Ph: (02) 845 3273 Fax: (02) 9845 3204
- Kogarah:** St George Hospital  
Gray Street  
KOGARAH NSW 2217  
Ph: (02) 9350 2315 Fax: (02) 9350 3901
- Liverpool:** Liverpool Health Services  
PO Box 103I  
LIVERPOOL NSW 2170  
Ph: (02) 9828 4665 Fax: (02) 9828 4650
- Newcastle:** Hunter Genetics  
PO Box 84  
WARATAH NSW 2298  
Ph: (02) 49 853 100 Fax: (02) 49 853 105
- Penrith:** Nepean Hospital  
Department of Clinical Genetics  
Derby Street, PENRITH NSW 2751  
Ph: (02) 47 343 362 Fax: (02) 47 343 764
- Randwick:** Sydney Children's Hospital  
High St  
RANDWICK NSW 2031  
Ph: (02) 9382 1704 Fax: (02) 9382 1711

## Genetic Counsellors/Outreach Clinics

### OUTSIDE SYDNEY AND NEWCASTLE AREA

- Bathurst:** Bathurst Community Health Centre  
Ph: (02) 63 315 533 Fax: (02) 63 322 039
- Broken Hill:** Broken Hill Community Health Centre  
Ph: (08) 80 801 556 Fax: (08) 80 801 611
- Canberra:** The Canberra Hospital  
Ph: (02) 62 555 042 Fax: (02) 62 443 834
- Coffs Harbour:** Mid-North Coast Health Service  
Ph: (02) 66 599 700 Fax: (02) 66 599 703
- Forster:** Forster Community Health Care  
Ph: (02) 65 929 703 Fax: (02) 65 929 607
- Gosford:** Central Coast Health  
Ph: (02) 43 370 207 Fax: (02) 43 370 217
- Goulburn:** Child Development Unit  
Ph: (02) 48 273 951 Fax: (02) 48 273 958
- Katoomba:** Blue Mountains Hospital  
Ph: (02) 47 343 362 Fax: (02) 47 343 764
- Mudgee:** Macquarie Area Health Service  
Ph: (02) 63 726 455 Fax: (02) 63 727 341
- Muswellbrook:** Muswellbrook Community Health Ctre  
Ph: (02) 65 422 050 Fax: (02) 65 422 005
- Nth Coast:** Child and Family Health Centre  
Ph: (02) 66 250 111 Fax: (02) 66 250 102
- Port Macquarie:** Pt Macquarie Community Health Ctre  
Ph: (02) 65 882 882 Fax: (02) 65 882 800
- Tamworth:** Tamworth Community health Centre  
Ph: (02) 67 662 555 Fax: (02) 67 663 967
- Taree:** Mid North Coast Area Health  
Ph: (02) 65 929 703 Fax: (02) 65 922 607
- Wagga Wagga:** Wagga Wagga Base Hospital  
Ph: (02) 69 386 666 Fax: (02) 69 215 632
- Wollongong:** Institute of Maternal & Paediatric Services  
Ph: (02) 42 225 216 Fax: (02) 42 225 698

