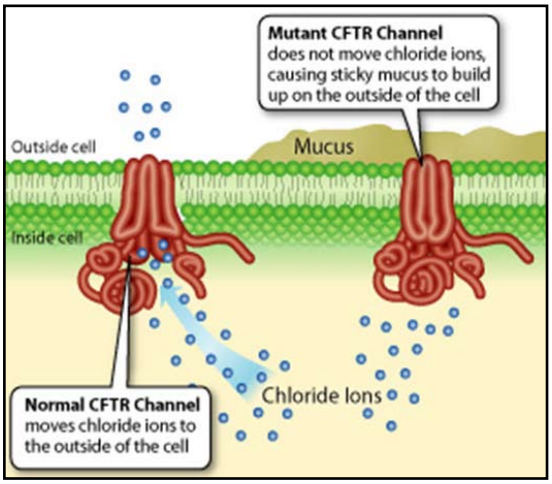
Cystic Fibrosis Screening
Information for Parents/Carers

What is Newborn Screening ?

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 Newborn Screening Laboratory in Potchefstroom (Ampath and North-West University Newborn Screening Programme) where it was tested for several disorders. Cystic fibrosis (CF) was one of the disorders.

What is Cystic Fibrosis?



Cystic fibrosis is a genetic disorder that affects the respiratory and digestive systems. People with cystic fibrosis inherit a defective gene on chromosome 7 called CFTR (cystic fibrosis transmembrane conductance regulator). The protein produced by this gene normally helps salt (sodium chloride) move in and out of cells. If the protein doesn't work correctly, that movement is blocked and an abnormally thick sticky mucous is produced on the outside of the cell. The cells most seriously affected by this are the lung cells. This mucous clogs the airways in the lungs, and increases the risk of infection by bacteria.

The thick mucous also blocks ducts in the pancreas, so digestive enzymes can't get into the intestines. Without these enzymes, the intestines cannot properly digest food. People who have the disorder often do not get the nutrition they need to grow normally.

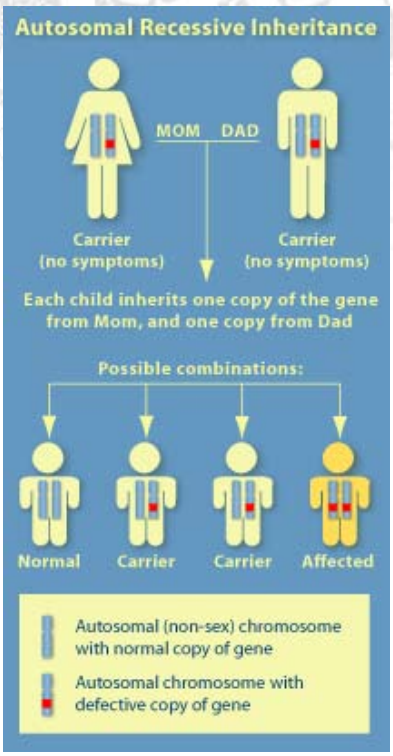
Finally, cystic fibrosis affects the sweat glands. Too much salt is lost through sweat, which can disrupt the delicate balance of minerals in the body

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

Cystic fibrosis occurs in about 1 in every 3 000 white babies, 1 in every 5 000 black babies and one in every 12 000 coloured babies and is one of the most common inherited diseases.

How do people get cystic fibrosis?

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm



Cystic fibrosis is a recessive disorder, which means that both parents must pass on the defective gene for any of their children to get the disease. If a child inherits only one copy of the faulty gene, he or she will be a carrier. Carriers don't actually have the disease, but they can pass it on to their children

Interesting facts about cystic fibrosis

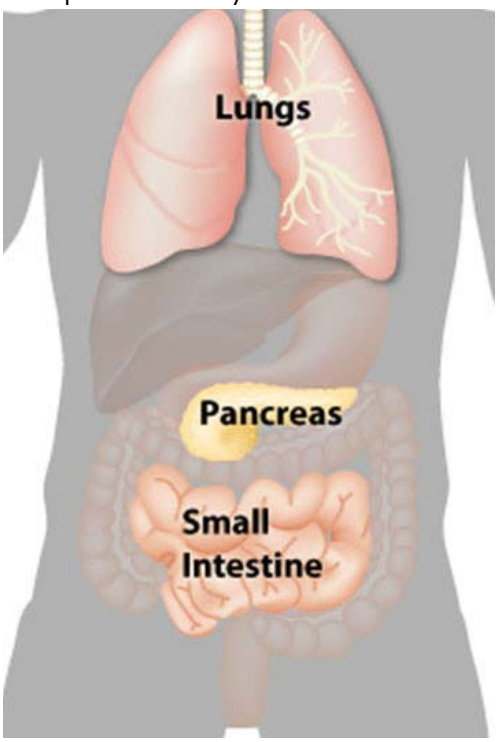
More than 1,000 different mutations in the CFTR gene have been identified in cystic fibrosis patients. The most common mutation (observed in 70% of cystic fibrosis patients) is a three-base deletion in the DNA sequence, causing an absence of a single amino acid in the protein product.

What are the symptoms of cystic fibrosis?

Symptoms of cystic fibrosis can include: coughing or wheezing, respiratory illnesses (such as pneumonia or bronchitis), weight loss, salty-tasting skin, and greasy stools. Because the lungs are clogged and repeatedly infected, lung cells don't last as long as they should. Therefore, most cystic fibrosis patients only live to be slightly more than thirty years old.

How do doctors diagnose cystic fibrosis?

People with cystic fibrosis have between 2 and 5 times the normal amount of salt in their sweat. Thus, doctors can use a sweat test to measure the amount of salt (sodium chloride) in a person's sweat. Sweat is collected from the person's arm or leg and taken to a laboratory to be analyzed. In newborns, doctors can measure the amount of a protein called trypsinogen in the blood. The level of this protein is higher than normal in people with cystic fibrosis. Finally, genetic tests can identify a faulty CFTR gene using a sample of the patient's blood.



How is cystic fibrosis treated?

Although there is no cure for cystic fibrosis, new treatments are helping people with the disease live longer than before. Most treatments work by clearing mucous from the lungs and preventing lung infections. The common treatments include:

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm

Chest physical therapy, in which the patient is repeatedly clapped on the back to free up mucous in the chest

Inhaled antibiotics to kill the bacterial that case lung infections. Bronchodilators (also used by people with asthma) that help keep the airways open

Pancreatic enzyme replacement therapy to allow proper food digestion. Gene therapy (a treatment currently in clinical trials), in which the healthy CFTR gene is inserted into the lung cells of a patient to correct the defective gene.

What tests are done to screen for CF?

Two types of 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 (IRT) secreted by the pancreas. In those babies who do have a high level, a second test, a DNA (gene) test, shows whether the baby carries any copies of the most common mutation associated with CF, called ΔF508 in white babies and 3120+1G➔A in black babies. Babies with two copies of ΔF508 or 3120+1G➔A definitely have CF.

A repeat of the IRT testing will be necessary at 24-27 days of age if the first IRT value was elevated and is helpful if no mutations are detected.

Our doctor has told us that our baby has one copy of the mutation ΔF508 (or 3120+1G➔A) and could have CF and must have a further test. What does this mean?

It is possible that your baby might have CF. It is most likely that your baby has one copy of the 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 ΔF508 (or 3120+1G➔A) gene mutation, and also one copy of another rare CF mutation, which we do not test for directly. There are about

1000 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.

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm

Our doctor has told us that our baby has an elevated second IRT test and could have CF and must have a further test. What does this mean?

An elevated IRT value between day 24 an 27 of life might mean that your baby has CF. (Approximately one out of every 10 babies with a second sample elevated IRT level has 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. 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, the skin may become slightly reddened.

Your baby's result may 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 protocol used by the sweat testing laboratory and the weight of your baby. Your pediatrician will send you to the appropriate facility to have the sweat test done.

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 important that the family has a genetic consultation. Your doctor can arrange this visit.

It is best if both parents are tested for the most common gene mutation, which is ΔF508 in the white population, and 3120+1G→A in the black population. 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. 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. All of these issues will be discussed with you at the genetic consultation.

There is a chance that the sweat test result may have to be repeated, if the results are such that they do not show categorically that your baby does not have CF. The possibility of CF must be considered, and your doctor will want to repeat the sweat test in a few weeks.

Our baby has only one copy of the ΔF508 mutation but the sweat test is POSITIVE. Does our baby have cystic fibrosis?

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm

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 that regularly performs sweat tests. Both you and your partner carry a CF gene. One of you carries the ΔF508 or 3120+1G□A mutation and the other carries one of the rarer mutations. Your baby must have inherited both CF genes, and therefore has cystic fibrosis.

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 nearest CF clinic. 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.

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?

South African Cystic Fibrosis Association Contact Details:

http://www.sacfa.org.za

Tel: 011-803 4670 / 083 6808423

Attached are the Cystic Fibrosis Advisory Committee of SA:

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm

Cystic Fibrosis advisory committee of SA.

Dr. Cathy Baird
Jhb. Hospital
bruceleech@telkomsa.net
cathybaird@telkomsa.net
Tel: 0114883496
0114883498
Fax: 0114884488
Cell: 0833248326

Dr. Jonathan Egner
St Augustine's Medical Centre
Chelmsford Road
Durban 4001
jegner@paediatrician.co.za
Tel: 0312010214 (w)
Fax: 0312010563
Cell: 0828233392

Dr. Susan Klugman
Dept. Paediatrics
Jhb Hospital
Ward 284
sblieden@global.co.za
Tel: 0114883983 (w)
0117869851 (h)
Fax: 0114883983
Cell: 0834075091

Prof. Michele Ramsay
Div. Human Genetics
NHLS
P O Box 1038
Johannesburg
2000
Michele.ramsay@nhls.ac.za
Tel: 0114899214 (w)
0116464248 (h)
Fax: 0114899226
Cell: 0736472726

Dr. Tamatha Urquhart
Pretoria Academic Hospital
P O Box 104
Woodlands
Pretoria
0072
tamatha@woodlandsnet.co.za
Tel: 0123542135 (w)
0129970670 (h)
Fax: 0123545275
Cell: 0834126350

Dr. Bertram Henderson
PO Box 339 (G11)
Bloemfontein
9300
gnmgbdh.md@mail.uovs.ac.za
Tel: 0514053046
Fax: 051444495
Cell: 0828959080

Dr. Graham Ducasse
Dept. Paediatrics
Greys Hospital
Private Bag X9001
Pietermaritzburg 3200
graham.ducasse@kznhealth.gov.za
Tel: 0338973000 (w)
0333431929 (h)
Fax: 0338973409
Cell: 0833257569

Dr. Paul Gebers
St. George's Hospital
Port Elizabeth
info@kidspe.co.za
wjp@global.co.za
Tel: 0413743586
Fax: 0413743583
Cell: 0826512318

Prof. Mervyn Mer
Adult Respiratory Clinic
Jhb Hospital
merm@medicine.wits.ac.za
Tel: 0114884911 (w)
0118852789 (h)
Fax: 0114883923
Cell: 0834881200

Prof. Paul Willcox
E16 Respiratory Clinic
Groote Schuur Hospital
Observatory
Cape Town and
UCT Private Academic Hospital
Anzio Road
Observatory
Cape Town 7925
pwillcox@samedical.co.za
Tel: 0214421810
0214421817 (w)
0216714744 (h)
Fax: 0216839296
Cell: 0832612064

Prof. Tony Westwood
Red Cross Children's Hospital
Klipfontein Rd
Rondebosch
Cape Town
anthony.westwood@uct.ac.za
Tel: 0216585190
Fax: 0216891287
Tel: 0216713671
Cell: 0737927133

Prof. Robin Green
Pretoria Academic Hospital
robin.green@up.ac.za
Tel: 0123545272
Fax: 0123545275