

Genetic Testing in New Zealand

Each newborn baby has a screening test for genetic conditions soon after birth, called the "Guthrie Heel Prick Test".

A positive result from this test may indicate the baby has CF. Most carriers of CF can also be identified by this test. A positive result is then confirmed by further testing.

Carrier Testing is available through regional genetic services.

It is free for:

- Relatives of people with CF
- People with a family history of CF
- Partners of people with CF
- Partners of carriers of CF

Carrier testing may also be done through your GP but there will be a cost involved. The test requires a blood sample or have a swab sample taken from your mouth.

Regional genetic services can discuss with couples at risk of having children with CF, the genetic risk and reproductive options. Including prenatal testing and pre-implantation genetic diagnosis (PGD).

Genetic Services in New Zealand

Northern Regional Genetic Service

Based at Auckland City Hospital,
Services the area Northland to Central North Island to Gisborne
Phone : 0800 476 123 or 09 307 4949 x25870

Central and Southern Regional Genetic Service

Based at Wellington and Christchurch Hospitals
Services the area New Plymouth to Invercargill
Wellington
Phone: 0508 364 436 or 04 385 5310
Christchurch
Phone: 0508 364 436 or 03 378 6574

Cystic Fibrosis Association of New Zealand

The Cystic Fibrosis Association of New Zealand aims to ensure increased life expectancy and improved quality of life for people with CF and their families; through research to achieve better control and ultimately a cure for the condition.

The Association's activities include providing: information about CF, support: social and financial, education, communication, advocacy, promoting public awareness, encouraging and funding research, and raising funds to ensure these services to the CF community continue into the future.

For more information about Cystic Fibrosis and the work of the Association, and to find out how you can access help or offer your help please contact our National Office or visit our website.

www.cfnz.org.nz



Cystic Fibrosis Association of New Zealand

Level 1, Broadway
62 Riccarton Road
PO Box 8241
CHRISTCHURCH 8440

Phone: 0800 651 122 or 03 341 8024
Fax: 03 341 8025
Email: info@cfnz.org.nz



Genetics of Cystic Fibrosis

- A Layperson's Guide



What is Cystic Fibrosis?

Cystic Fibrosis (CF) is an inherited genetic condition that is passed on by parents to their child in the child's genes. To be affected by CF, a child must inherit a CF gene from each parent.

People with CF are born with the condition and it will affect them throughout their life. CF shortens a person's life expectancy but there are continual advancements in treatments and therapies.

What is a gene?

A person's unique combination of genes is known as their 'genetic make-up'. Genes, made of DNA, are the instruction manuals for our bodies. They direct the production of proteins which make our bodies function. Faulty genes can cause parts of our bodies not to function correctly, as seen in CF.

Genes come in pairs: one copy is passed on from the person's mother, the other from their father. Cystic Fibrosis is a recessive disorder where a person has two copies of the CF gene: one copy from each parent.

What does it mean to be a 'carrier' of CF?

A person with one copy of the CF gene and one normal copy is called a 'carrier'. They generally have no symptoms of CF. Most carriers have no idea they are carriers as they are completely healthy. Prior to having a child with CF, carriers can only be identified through genetic testing.

In New Zealand, 1 in 25 Caucasian people will be a carrier of CF. You have a higher chance of being a carrier if you have a family history of CF or relatives who have CF.

What is the risk of passing CF onto your children?

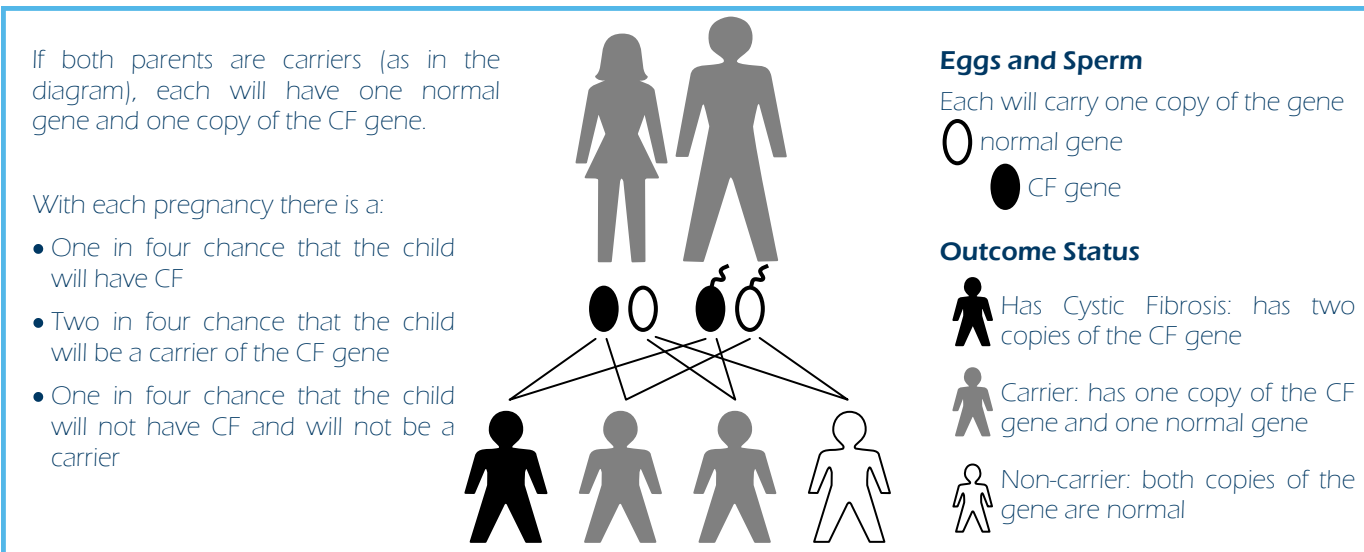
A child must receive two copies of the CF gene to have CF. An individual with CF will always pass on a CF gene whereas a carrier has a one in two chance of passing on the CF gene.

Two carriers can have children who have CF. In New Zealand, the incidence of two CF carriers in a couple occurs one in 625 couples.

Are all CF genes the same?

Researchers have identified many different variations to the CF gene, the most common is known as $\Delta F508$, and it accounts for between 50-80% of all cases. Most individuals in New Zealand have a least one copy of this gene.

As there are a number of variations in the gene, there are also variations in how CF affects the person with the condition, some have more serious complications than others, some keep very healthy most of the time.



How common is CF?

CF is the most common genetic condition in New Zealand, affecting one in every 3500 births. It occurs mostly in Caucasian people, and rarely in Africans, Asians and Polynesians. Currently there are more than 500 children and adults living with CF in New Zealand.

Where can you get help or further information?

- Your local GP and health care professionals
- Regional Genetic Services, who provide genetic counseling and genetic testing
- The Cystic Fibrosis Association of New Zealand