

How does one access genetic counselling and testing for NF2?

Genetic counselling should precede genetic testing. In South Australia this is available through the Familial Cancer Unit. Similar services exist in other states or territories. We recommend that you discuss this with your doctor.

Familial Cancer Unit
SA Clinical Genetics Service
Women's and Children's Hospital
North Adelaide
SA 5006
AUSTRALIA

Tel: (08) 8161 6995
Fax: (08) 8161 7984
Email: sacgs@mail.wch.sa.gov.au

Useful website:

The National Neurofibromatosis
Foundation
www.nf.org

Useful contacts:

Association of Genetic Support of
Australasia (AGSA)
66 Albion Street
SURRY HILLS NSW 2010
Email: agsa@ozemail.com.au
Website: www.agsageneticsupport.org.au

The Neurofibromatosis Association of
South Australia Inc.
c/- Disability Information and
Resource Centre
195 Gilles Street
Adelaide SA 5000
Phone: (08) 8223 7522
Fax: (08) 8223 5082

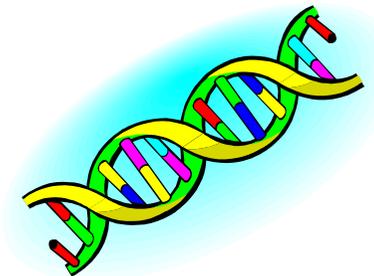
Useful contact:

Cancer Help Line
13 11 20

Information, advocacy, counselling
and support services can all be
accessed through the
Cancer Help Line

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Genetic testing for Neurofibromatosis type 2 (NF2) for families



**South
Australian
Familial
Cancer
Service**

What is Neurofibromatosis type 2?

Neurofibromatosis type 2 is a rare genetic disorder which causes tumours to grow on various types of nerves in the skull and spine. It is also known as Central Bilateral Acoustic NF or hereditary schwannomatosis.

Signs of NF2 usually appear in the 20s but may occur earlier or later in life.

Features of NF2 include:

- ❖ Tumours on both of the auditory nerves which may cause deafness, ringing in the ears or problems with balance.
- ❖ Tumours on the brain, spinal cord or meninges.
- ❖ A family history of NF2.
- ❖ Cataracts at a young age.

How do people get NF2?

Genes are working units of DNA, a complex set of genetic information within the body's cells that instructs them to work normally. NF2 is a disease that results from an error, or a mutation in a gene called the NF2 gene.

In half of the cases NF2 is inherited from a parent. Genes come in pairs and a child inherits one gene from each parent. The mutation may be transmitted from a parent to a child. If one parent has a mutation of the NF2 gene the child has a 50% chance of inheriting that mutation.

The remaining half of NF2 cases result from a new or spontaneous mutation in the sperm or egg cell from which the person developed. In this situation the siblings and parents of an affected person are at low risk of having NF2, though their offspring have a 50% chance of inheriting NF2.

What is Genetic Testing for NF2?

It is possible to test a person's blood in order to find out whether they have a mutation in the NF2 gene. The current laboratory

techniques that are used can detect over 60% of NF2 mutations.

Once a mutation has been identified in an affected family member, genetic testing for NF2 for other family members is 100% accurate. It is important to have genetic counselling before undergoing genetic testing.

What are the advantages of genetic counselling and testing for NF2?

- ❖ The presence of a mutation in the NF2 gene identifies those people who may be at risk of developing tumours and would derive the greatest benefit from regular screening.
- ❖ Relatives of those with NF2 can also be tested for the same mutation to determine whether they also would benefit from regular screening.
- ❖ If a relative is found not to have inherited the mutation then no further screening is necessary. The emotional and financial costs of such screening can therefore be avoided.