

Contact details

Clinical inquiries should be directed to:

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

Tel: (08) 8161 7375
Fax: (08) 8161 6088
Email: sacgs@mail.wch.sa.gov.au

Laboratory inquiries should be directed to:

Division of Molecular Pathology
Institute of Medical and Veterinary Science
Frome Road
Adelaide SA 5000

Tel: (08) 8222 3895
Fax: (08) 8222 3146
Email: jacqueline.carroll@imvs.sa.gov.au

Useful website:

The National Neurofibromatosis
Foundation
www.nf.org

Useful contact:

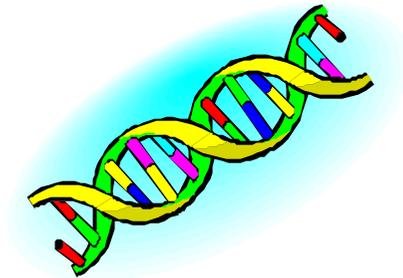
Association of Genetic Support of
Australasia (AGSA)
66 Albion Street,
SURRY HILLS NSW 2010
Email: agsa@ozemail.com.au
Website: www.agsageneticsupport.org.au

Checklist:

- ✓ Blood sample collected-10ml in EDTA
- ✓ Consent form completed and signed
- ✓ Service agreement form completed and signed
- ✓ All samples and paperwork sealed in IMVS Path-o-Pak

Samples should be kept at room temperature at all times and transported to the laboratory within 24 hours where practicable.

Genetic Testing for Neurofibromatosis Type 2 (NF2) for clinicians



**South
Australian
Familial
Cancer
Service**

Introduction

A person with an inherited mutation in the NF2 gene is at high risk of developing a number of tumours. The average age at diagnosis is 20 years with 90% of carriers being symptomatic by the age of 45. However some carriers do not present until the seventh decade.

Approximately 70% of patients have the typical "adult onset" form of NF2 which manifests as bilateral **vestibular schwannoma**. The patient may present with deafness or tinnitus. The early diagnosis of these tumours is associated with a better outcome. Vestibular tumours typically grow 2mm in diameter every year, with a faster rate of growth in children.

Up to 30% of patients may present in the first two decades of life with a non-vestibular tumour such as intracranial **meningioma**, **astrocytoma** or **spinal tumour**. **Ten percent (10%)** of carriers present under the age of 10 years. Early presentation with a non-vestibular tumour is an adverse prognostic indicator.

Spinal tumours of various types occur in the majority of patients but are often asymptomatic and remain benign.

NF2 Gene Testing

Germline mutations in the NF2 gene can be detected utilising current techniques in approximately 85% of people with the disease. Analysis of the NF2 gene involves screening the exons of the gene for any small insertions, deletions or single-base substitution mutations by direct DNA sequencing. If no mutation is found in this instance, then a technique for the investigation of whole exon deletion and/or insertions is performed. Once a mutation has been identified in a patient, genetic testing of relatives is essentially 100%.

Testing is conducted under the auspices of the South Australian Familial Cancer Service.

Turnaround time for testing

Most results are available within 2 months.

Detection of an identified mutation can be extended to presymptomatic testing in other family members. This testing takes about 4 weeks to complete.

Procedure For Testing

1. Collect 10ml fresh blood in EDTA. Please ensure that these samples are kept at room temperature at all times including transportation to the lab.
2. Ensure that a *Consent Form* from a Familial Cancer Clinic accompanies each of the samples to be tested. We require that informed consent be obtained and documented. (A suitable consent form is available from the laboratory if required).
3. Complete the *Service Agreement Form* indicating that payment can be guaranteed.
4. Place all paperwork and samples in the IMVS Path-O-Pak and complete the IMVS request form on the front.
5. Forward to the laboratory within 24 hours of collection where practicable.

Results

A full report will be issued on the outcome of the testing. Please indicate on the IMVS request form if additional reports are required to be issued to other specialists involved in the case.

Identification of a mutation in the NF2 gene serves to aid both in the genetic counselling process and in early clinical management.