virtus DIAGNOSTICS

Virtus Diagnostics offers a complete range of general pathology services as well as specialist fertility and genetic testing.

Virtus Diagnostics conducts over a million tests each year across the following disciplines: general pathology, fertility pathology and advanced genetic testing.

We continually innovate and expand this range and with over 200 scientists, many who are internationally renowned in their field, you can be assured you are accessing world class diagnostic services.

Virtus Diagnostics is committed to supporting patients' health throughout their lifetime, and in particular, during their reproductive life. For women this includes general gynaecology and for both men and women it includes general health, fertility planning and conception, through to infertility treatment.

Our service offers:

- Specialist pathologists
- A comprehensive range of tests
- Fast turnaround of results
- Highly skilled and experienced scientists, with access to the latest technology
- Convenient collection centres, and
- Personalised service

Virtus Diagnostics Collection Centres

The network of Virtus Diagnostics collection centres are across NSW, QLD, VIC & TAS. Please refer to virtusdiagnostics.com.au for the complete list of our collection centres.

Virtus Diagnostics General enquiries
1800 837 284

Virtus Diagnostics Accounts enquiries
1800 090 325

virtusdiagnostics.com.au

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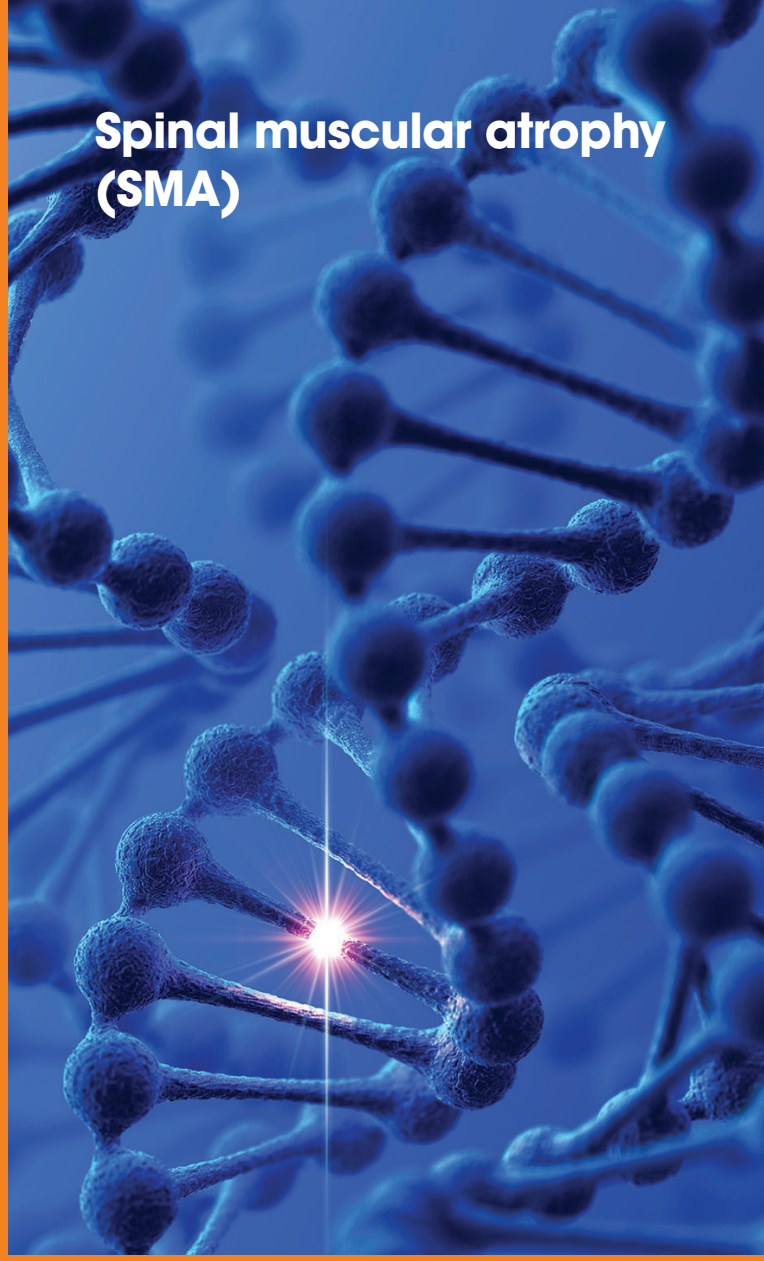
A member of virtushealth

Spinal muscular atrophy (SMA)



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Spinal muscular atrophy (SMA)

The facts:

- SMA is a disease affecting nerves and the spinal cord resulting in muscles becoming progressively weaker
- Carriers of SMA are normal
- If both parents are SMA carriers the chance of having a child with SMA is 1 in 4
- There are 4 distinct types of SMA
- SMA type 1 is the most common and most severe
- Survival for SMA type 1 is usually not greater than two years of age
- Babies with SMA appear normal but are very weak
- Children with SMA have normal intelligence
- There is currently no cure for SMA

Spinal muscular atrophy (SMA)

Spinal muscular atrophy (SMA) is a neuromuscular disorder of the nerves. The nerves in the spinal cord are affected and the link between the brain and the muscles in the body is lost causing the muscles to become wasted. The progressive muscle wasting often leads to early death. The gene (SMN1) involved in this disorder has part of its genetic information deleted.

There are four types of SMA, with type 1 being the most common as well as the most severe. Affected individuals usually do not survive much beyond two years of age. A drug, Spinraza, has recently been released and can be prescribed to help ameliorate the symptoms of the disease.

Usually there is no family history of the disorder as carriers are unaffected. Pre-conception screening offers couples the ability to determine their carrier status prior to pregnancy.

SMA is inherited in an autosomal recessive manner. For a child to have SMA both parents must be carriers. When two SMA carriers have a child there is:

- a 1 in 4 chance their child will be affected by SMA
- a 1 in 2 chance their child will be a carrier of SMA but unaffected like the parents
- a 1 in 4 chance the child will not inherit the SMA gene variant and will not be at risk of passing SMA to future generations
- every pregnancy carries the same risk of conceiving an affected child as the previous one

The gene for the disorder, SMN1 (survival of motor neuron protein) is found in all ethnic groups. The prevalence of affected individuals is estimated at 1 per 10,000 with one in 40-60 persons being carriers.

The majority of cases (approximately 95%) are caused by a deletion of genetic information in the SMN1 gene; however rare cases are caused by point mutations. In a small number of cases, neither parent is shown to be a carrier.

A SMA screening blood test is available at Virtus Diagnostics. If one member of the couple is tested initially and shown to be a carrier your doctor will recommend that your partner be tested. If both members of a couple are shown to be carriers, referral for genetic counselling can be arranged.

Cascade Carrier screening

If you are shown to be a carrier, and you have siblings or other close relatives, they may also be at risk of being carriers. Testing for these individuals is also available should they wish to determine their carrier status.

Accuracy of testing:

This screening test detects the most common cause of SMA - 95% of all cases are due to a deletion in the gene.

How much does it cost?

The test does not have a Medicare rebate – please contact Virtus Diagnostics for costings. This test can be ordered separately or can be bundled as part of the Virtus Genetic Screen (Cystic Fibrosis, Fragile X and Spinal Muscular Atrophy).