Mission statement:
The Muscular Dystrophy Foundation of South Africa is a non-profit organisation that supports people affected by muscular dystrophy and neuromuscular disorders.

1. **Is Spinal Muscular Atrophy also known by other names?**
There are different forms of this disorder, and the disorder is often referred to by its abbreviation "SMA". Infantile SMA is also known as Werdnig-Hoffman disease and Juvenile SMA is also known as Wohlfart-Kugelberg-Welander disorder, or just Kugelberg-Welander disorder.

2. **What is Spinal Muscular Atrophy?**
Spinal muscular atrophy is a term for a group of inherited neuromuscular disorders. All forms of the disorder attack specialised nerve cells, called motor neurons, which control the movement of voluntary muscles. The muscles weaken and waste away due to the degeneration of the motor neurons. Progressive spinal muscular atrophy characterizes a group of disorders with varying severity, manifesting mainly in childhood.

There are 3 main forms of this disorder:
- Infantile progressive spinal muscular atrophy (or Werdnig-Hoffman disorder)
- Intermediate spinal muscular atrophy
- Juvenile spinal muscular atrophy (or Wohlfart-Kugelberg-Welander disorder)

3. **What causes Spinal Muscular Atrophy?**
All of the Spinal Muscular Atrophies are caused by gene defects. The defective genes that cause Spinal Muscular Atrophy prevent the normal functioning of motor neurons, leading to their deterioration and muscle degeneration. Most forms of SMA are due to the absence of a gene called SMN (survival motor neuron) on chromosome 5.

4. **What are the symptoms?**
The different forms of SMA have differences in the age of onset as well as in the severity and in the progression of the condition.

<table>
<thead>
<tr>
<th>Form</th>
<th>Age of Onset</th>
<th>Symptoms</th>
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</thead>
<tbody>
<tr>
<td>Infantile form</td>
<td>before birth to 6     months</td>
<td>Rapidly progressive, by 3 months most affected individuals are unable to move due to paralysis in their arms, legs and lower and upper torso; they also have trouble swallowing and sucking; due to breathing distress and respiratory infection, life-span rarely exceeds the age of 2 years</td>
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<tr>
<td>Intermediate form</td>
<td>6 months to 3 years</td>
<td>Infants progress normally initially, then they develop moderate to rapid progressive muscle weakness in the arms, as well as in the legs and upper and lower torso, making standing difficult; most survive until early childhood and some live longer, depending on the extent to which respiratory muscles are affected.</td>
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<tr>
<td>Juvenile form</td>
<td>1 to 15 years</td>
<td>Moderately progressive weakness in the leg and hip muscles making it difficult to climb stairs and to stand up. The child waddles when walking; respiratory muscles may be weakened; can walk for at least 10 years after symptoms become apparent; generally unaffected life-span.</td>
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5. **Which muscles are affected?**
Infantile form: Generalised paralysis of the limbs; the legs are more severely affected than the arms; the muscles of the upper parts of the limbs are more affected than the muscles of the lower part of the limbs; there is marked weakness of all the axial muscles (central muscles); intercostal muscles (muscles of the rib-cage which help with breathing) are severely affected.

Intermediate form: As above except that the intercostal muscles are less severely affected.

Juvenile form: Weakness of the leg and hip muscles; may also have weakness of the muscles of the arms; respiratory muscles may be weakened.

6. **How is Spinal Muscular Atrophy inherited?**
All forms of Spinal Muscular Atrophy are inherited therefore, having the condition is beyond the control of the parents and children. The infantile, intermediate and juvenile forms of spinal muscular atrophy are all inherited in an autosomal recessive pattern, which means that both parents must pass on a faulty gene to an affected child. Males and females...
are equally affected. If a child receives only one faulty gene from one parent then they are not affected but are carriers of Spinal Muscular Atrophy. Children of parents who both carry the gene for spinal muscular atrophy have a 25% chance of inheriting the disorder, a 50% chance of being a carrier of the defective gene, and a 25% chance of being unaffected, a non-carrier and thus unable to transmit the condition. Inheritance in the adult form may follow a different pattern of inheritance. As this is a genetic condition, genetic counselling is strongly recommended. Genetic counselling provides information on the inheritance pattern, risks to other family members, prognosis, psycho-social support, as well as information about diagnostic testing, carrier testing, preclinical and prenatal testing (where available).

7. **How is Spinal Muscular Atrophy diagnosed?**
A clinical diagnosis is made by an experienced physician or neurologist who performs a thorough physical examination as well as evaluating the affected person's medical history. The clinical diagnosis is then confirmed by a series of laboratory tests. DNA analysis can also be done to confirm the diagnosis. In all types of SMA a gene called SMN is generally missing on both copies of the chromosome. DNA testing should be done before a muscle biopsy as it is non-invasive. If the DNA test is positive, the diagnosis is confirmed and further testing may be unnecessary. Other tests include a muscle biopsy where a small piece of muscle tissue from an affected individual is analysed by a pathologist to determine whether the person has SMA; an electromyogram (EMG) which is done by placing small electrodes in muscle, this then creates a graph that indicates the health of the body's muscles and nerves. Blood tests are also done to evaluate the levels of certain enzymes and help to distinguish Spinal Muscular Atrophies from other neuromuscular disorders.

8. **Is there a cure?**
Unfortunately, there is no known cure yet that will stop or reverse any form of SMA.

9. **Is there any treatment?**
Physical therapy and orthopaedic devices can alleviate the symptoms of the person affected with intermediate, juvenile and adult SMA. They can also help the affected person walk until a later stage. Orthopaedic devices or surgery may also help to counteract scoliosis, or curvature of the spine.

10. **Is carrier detection available?**
Not at present, but this may become available shortly.

11. **Is there a prenatal diagnosis?**
Prenatal testing is possible for SMA in South Africa. It can be done as early as 10 weeks of pregnancy through a specialist genetic centre.

12. **Is there a risk during anaesthesia?**
SMA is associated with a higher risk of complications during anaesthesia than that of the general population because of the muscle weakness as well as sensitivity to some of the drugs. It is therefore of the utmost importance that anaesthesia only be given by a specialist anaesthetist who is well aware of these possible complications. It is always recommended that the presence of a muscular disorder be mentioned to your doctor.

13. **Has research been conducted on Spinal Muscular Atrophy in South Africa?**
Yes, researchers in the Department of Human Genetics at the SAIMR and University of the Witwatersrand have shown that virtually all Caucasian affected persons with SMA have the same genetic fault, that is the same SMN gene is missing. This is extremely helpful as it simplifies diagnostic testing. In black patients, only 65% are missing the SMN gene. In the other 35% the cause is unknown. Research is ongoing to determine the cause of SMA in black patients.

14. **The role of the Muscular Dystrophy Foundation in South Africa**
The MDF supports individuals affected by muscular dystrophy and their families by offering emotional support, information - including a series of fact sheets, referrals to genetic counselling and other clinics, formation of support groups, assistance with special equipment, when possible, as well as financial support for research projects in muscular dystrophy in South Africa. Creating public awareness for muscular dystrophy is also an important aspect of our work, since the MDF relies solely on contributions from its members and other donors to provide an on-going support service. Through our newsletter members are kept informed of all the activities and receive national and international research updates. Please contact any office of the MDF if you require information about any of our activities or programmes.

15. **Support group or contact person**
Zeta Starograd (children)  (011) 640-1531
Lucie Swanepoel  (017) 683- 0287
16. **Where can we find assistance?**

Please contact your local MDF office for further information.

### National Office

**Cape Branch**
- P.O. Box 13449, Mowbray, 7705
- Tel: (021) 448-8766, Fax: (021) 448-8766
- email: cape@mdsa.org.za

**Gauteng Branch**
- P.O. Box 1535, Pinegowrie, 2123
- Tel: (011) 789-7635, Fax: (011) 781-3935
- email: gauteng@mdsa.org.za

**Kwazulu-Natal Branch**
- P.O. Box 290, New Germany, 3620
- Tel: (031) 701-3801, Fax: (031) 701-3801
- email: kzn@mdsa.org.za

### Local Clinics:

Please contact your local MDF branch for further information.

### General Information:

- Independent Living Centre (ILC): (011) 482-5476
- Disability Info and Care (DIC): (011) 917-3284

### Parking Concessions:

Application to the Traffic Department of your Local Authority.

Criteria: (a) if you need the extra width as provided by the special parking bays, or
(b) if you have a problem with walking long distances.

### Special Equipment:

Phone either ILC or DIC for information on where to get special equipment.

### MDF Website:

Please visit our MDF website (www.mdsa.org.za) for muscular dystrophy news updates.

15. **Please note**

The treatments and drugs mentioned in this fact sheet are for information purposes ONLY. Please consult your physician or other health care specialist for information regarding the use of any of the above. The MDF encourages duplication of this fact sheet, under the following condition: that it is duplicated in its entirety - including the MDF logo and full text. Only individuals authorised by the MDF may make changes to this fact sheet (the information "updated by" and "last update" should be completed). Alterations to this fact sheet by any other party are strictly prohibited.

This fact sheet was adapted from the following source(s): Fact sheet(s) of the Muscular Dystrophy Group of Great Britain and Northern Ireland.

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