What is DSMA type V?

Distal hereditary motor neuropathy type V is a progressive condition that affects nerve cells in the spinal cord. It results in muscle weakness and affects movement of the hands and feet.

What are the symptoms and effects?

The first symptom is often cramps in the hand brought on by exposure to cold temperatures. Symptoms usually begin during adolescence, but onset varies from infancy to the mid-thirties.

Over time this first symptom is followed by weakness and wasting (atrophy) of hand muscles, specifically on the thumb side of the index finger and in the palm at the base of the thumb. It’s also common to have a high foot arch, and some people eventually develop difficulties with way they walk.

People with this condition have a normal life expectancy.

Which gene is affected?

Some of the genes that cause rarer forms of SMA are associated with more than one condition, so please be aware that the website links suggested might provide information that isn’t just about SMA.

The affected gene is:

- **BSCL2**
  - Further information can be found at: [www.ghr.nlm.nih.gov/gene/BSCL2](http://www.ghr.nlm.nih.gov/gene/BSCL2)

- **GARS**
  - Further information can be found at: [www.ghr.nlm.nih.gov/gene/GARS](http://www.ghr.nlm.nih.gov/gene/GARS)

Inheritance pattern

DSMA – V has an **autosomal dominant** inheritance pattern.

To find out more about this please see our information sheet, ‘**The inheritance patterns of some rarer forms of SMA**’.
Support and resources

Though not a substitute for professional medical advice, the US National Library of Medicine, Genetics Home Reference provides more information:


SMA UK
Provides information and support for anyone in the UK affected by any form of SMA.
Phone: 01789 267520
Website: www.smauk.org.uk

Contact
Provides information and support for families with children with a disability.
Phone: 0808 808 3555
Website: www.contact.org.uk

References


We are grateful to the writers and reviewers who assist us in our information production. A list of who this includes may be viewed on our website: www.smauk.org.uk/our-writers-and-reviewers-panel or requested from supportservices@smauk.org.uk

Whilst every effort is made to ensure that the information in this publication is complete, correct and up to date, this cannot be guaranteed and Spinal Muscular Atrophy UK shall not be liable whatsoever for any damages incurred as a result of its use. SMA UK does not necessarily endorse the services provided by the organisations listed in our information sheets.

If you have any feedback about this information, please do let us know at supportservices@smauk.org.uk