Distal Spinal Muscular Atrophy Type V (DSMA-V)

You can also read this guide on our website at smauk.org.uk/fneu where you can follow all the links we give to further information.

Who is this for

This information is for you, if you or your child have been recently diagnosed with DSMA-V.

1. What are the Symptoms and Effects?

The first symptom is often cramps in the hands and / or feet 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 foot and / or hand muscles, specifically on the thumb-side of the index finger and in the palm at the base of the thumb. It is also common to have a high foot arch, and some people develop difficulties with walking.

The condition is slowly progressive and with time will start to impact muscles in the legs and arms. People with this condition have a normal life expectancy.

DSMA-V is a form of genetic peripheral neuropathy that affects the motor neurons, so is also often referred to as distal hereditary motor neuropathy, type 5. Symptoms

and effects of genetic peripheral neuropathy are described in more detail in the information published by the charity Charcot-Marie-Tooth UK (see below under Support and Resources).

2. What is the Genetic Cause of DSMA-V?

DSMA-V has been linked to mutations in several different genes:

- For further information about the **BSCL2 gene**[1-2] **>**
- For further information about the **GARS1 gene**[3-4] **>**
- For further information about the **REEP1 gene** [5-6] **>**

Based on the gene that causes the condition, DSMA-V is sometimes subdivided into DSMA-VA (GARS1), DSMA-VB (REEP1) and DSMA-VC (BSCL2).

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 provided may also give information about other conditions.

3. Inheritance Pattern

DSMA-V has an autosomal dominant inheritance pattern, which means that only one faulty gene copy passed on from a parent with the condition will cause the condition.

For more about this form of inheritance, please see: The inheritance patterns of some rarer forms of SMA >

4. Support and Resources

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

Charcot-Marie Tooth UK >

enquiries@cmt.org.uk

Phone: 0300 323 6316

- **CMT and You >**
- **CMT Symptoms >**

SMA UK>

- Phone: 01789 267520
- Email: **office@smauk.org.uk**

Provides information and support for anyone in the UK affected by any form of SMA.

Contact >

- Phone: 0808 808 3555

Provides information and support for families with children with a disability.

5. References

1. Windpassinger et al. (2004) Heterozygous missense mutations in BSCL2 are associated with distal hereditary motor neuropathy and Silver syndrome. *Nat Genetics* 36: 271-276.
2. https://www.omim.org/entry/619112 (last accessed 22nd January 2025)

3. Antonellis et al. (2003) Glycyl tRNA synthetase mutations in Charcot-Marie-Tooth disease type 2D and distal spinal muscular atrophy type V. *Am J Hum Genet* 72: 1293-1299.
4. https://www.omim.org/entry/600794 (last accessed 22nd January 2025)
5. Beetz et al. (2012) Exome sequencing identifies a REEP1 mutation involved in distal hereditary motor neuropathy type V. *Am J Hum Genet* 91: 139-145.
6. https://www.omim.org/entry/614751 (last accessed 22nd January 2025)

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