SMA with Lower Extremity Predominance (SMA-LED)

You can also read this guide on our website at smauk.org.uk/mq80 where you can follow all the links we give to further information.

Who is this for

This information is for if you, if you / your child has been recently diagnosed with SMA-LED.

1. What is SMA-LED?

SMA-LED is a rarer form of SMA that begins in infancy or early childhood and progresses slowly. Like 5q SMA, SMA-LED affects the nerve cells called motor neurons. These cells transmit electrical signals from the spinal cord to muscles for voluntary muscle contraction.

SMA-LED causes motor neurons to malfunction, resulting in weakness in leg muscles, which is most severe in the thigh muscles (quadriceps). Children and adults often have a 'waddling' or unsteady walk and have difficulty climbing stairs and getting up from sitting.

There is only limited information about SMA-LED because it is so rare, but your or your child's medical team will be able to discuss how it affects you / your child and how to manage symptoms for maximum comfort and quality of life.

2. Which Gene is Affected?

SMA-LED has been linked to mutations in two different genes:

- For further information about the **DYNC1H1[1-2] gene ›**
- For further information about the **BICD2[3-6] gene ›**

Some of the genes that cause rarer forms of SMA are associated with more than one condition, so please be aware that the provided website links may give information about disorders additional to SMA.

3. Inheritance Pattern

SMA-LED has an Autosomal Dominant inheritance pattern, which means that only one faulty gene copy passed on from a parent with the condition is enough to cause the disease.

To find out more, please see : The Inheritance Patterns of Some Rarer Forms of SMA ›

4. Support and Resources

Though not a substitute for professional medical advice, MedlinePlus is a service of the US National Library of Medicine (NLM). It provides **more information about SMA-LED ›**

SMA UK › (smauk.org.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. Harms et al. (2012) Mutations in the tail domain of DYNC1H1 cause dominant spinal muscular atrophy. *Neurology* 78: 1714-1720.
2. https://www.omim.org/entry/158600 (last accessed 22nd January 2025)
3. Neveling et al. (2013) Mutations in BICD2, which encodes a golgin and important motor adaptor, cause congenital autosomal-dominant spinal muscular atrophy. *Am J Hum Genet 92*: 946-954.
4. Peeters et al. (2013) Molecular defects in the motor adaptor BICD2 cause proximal spinal muscular atrophy with autosomal-dominant inheritance. *Am J Hum Genet 92*: 955-964.
5. Oates et al. (2013) Mutations in BICD2 cause dominant congenital spinal muscular atrophy and hereditary spastic paraplegia. *Am J Hum Genet 92*: 965-973.
6. https://www.omim.org/entry/615290 (last accessed 22nd January 2025)

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