X-Linked Spinal Muscular Atrophy

You can also read this guide on our website at smauk.org.uk/cip4 where you can follow all the links we give to further information.

Who is this for

This information is for you if your child has been recently diagnosed with X-linked SMA.

1. What is X-linked SMA?

This very rare form of SMA appears in infancy and causes severe muscle weakness and difficulty breathing. Children often have misshapen joints that make movement difficult. Babies who are severely affected may be born with broken bones.

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

2. Which Gene is Affected?

The affected gene is:

- **UBA1 (X sex chromosome) >** [1–2]

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 is not just about SMA.

3. Inheritance Pattern

X-linked SMA has an X-linked Recessive inheritance pattern.

To find out more about this, please see our information guide The Inheritance Patterns of Some Rarer Forms of SMA >.

4. Research into Treatment

Little is known about the biology of X-Linked SMA. However, in 2023 Muscular Dystrophy UK awarded a grant to Professor Thomas Gillingwater and colleagues to characterise a newly generated mouse model. This is much needed to test new treatments for the condition.

Read more on MDUK's website >.

5. Support and Resources

Though not a substitute for professional medical advice, the US National Library of Medicine, Genetics Home Reference provides more information. Click here to read more.

SMA UK >

- Phone: 01789 267520

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.

Children's hospices

Located throughout the UK, these offer a wide range of services and support to eligible children and families; some also offer short breaks. Details of hospice services and more information are available from **Together for Short Lives ›**

6. References

1. Ramser et al. (2008) 'Rare missense and synonymous variants in UBE1 are associated with X-linked infantile spinal muscular atrophy', *American Journal of Human Genetics*, 82, pp. 188-193.

2. **https://www.omim.org/entry/301830** (accessed 22nd January 2025).

Version: 3

Author: SMA UK Information Production Team

Last Reviewed: January 2025

Next Review: January 2028

Links last checked: January 2025

This publication, and its links, provides information. It is meant to support, not replace, clinical and professional care. Find out more about ***how we produce our information****.*

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