If your child has recently been diagnosed with:

X-linked Spinal Muscular Atrophy

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's limited information about X-linked SMA because it's 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.

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 is not just about SMA.

The affected gene is:

- **UBA1 (X sex chromosome)**[1]

Further information can be found at: www.http://ghr.nlm.nih.gov/gene/UBA1

Inheritance pattern

X-linked SMA has an **X-linked Recessive** 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

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

Together for Short Lives
Provides information, support and services for families of children who are expected to have short lives
Phone: 0808 8088 100
Website: www.togetherforshortlives.org.uk

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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.

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

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