

If your child has recently been diagnosed with:

# X-linked Spinal Muscular Atrophy

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## 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)<sup>1</sup>**

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

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