

Why did this happen?

A blood test to check both parents' chromosomes is needed to find out why the 8p23 deletion occurred. In the majority of cases the 8p23 deletion occurred when both parents have normal chromosomes. The term that geneticists use for this is *de novo* (dn) which means 'new'. *De novo* 8p23 deletions are caused by a change that occurred when the parents' sperm or egg cells formed or possibly during formation and copying of the early cells after the egg and sperm joined. Some 8p23 deletions are accompanied by a gain of material from another chromosome and are often the result of a rearrangement in one parent's chromosomes. This is usually a rearrangement known as a balanced translocation in which material has swapped places between chromosomes. As no genetically important material has been lost or gained, the parent usually has no clinical or developmental problems, although they may have difficulties with fertility or childbearing. Balanced translocations involving one or more chromosomes are not rare: one person in 500 has one, making a total world population of over 13 million balanced translocation carriers. Whether the deletion is inherited or *de novo*, what is certain is that as a parent there is nothing you did to cause the 8p23 deletion and nothing you could have done would have prevented it from occurring in your baby. No environmental, dietary or lifestyle factors are known to cause these chromosome changes. No one is to blame when this occurs and nobody is at fault.

Can it happen again?

The possibility of having another pregnancy with an 8p23 deletion depends on the parents' chromosomes. If both parents have normal chromosomes when their blood cells are tested, the deletion is very unlikely to happen again. However, there is a very small theoretical possibility that the deletion occurred during the formation of the egg or sperm cells in a parent. If this occurs there is a tiny chance that parents with apparently normal chromosomes could have another affected pregnancy. On the other hand, if either parent has a chromosome rearrangement or deletion involving 8p23, the possibility is greatly increased of having other affected pregnancies. Parents should have the opportunity to meet a genetic counsellor to discuss the specific recurrence risks and options for prenatal and preimplantation genetic diagnosis (PGD). PGD requires the use of in vitro fertilisation and embryo biopsy, and only healthy embryos are transferred to the mother's uterus. If the parents choose to conceive naturally, prenatal diagnosis options include chorionic villus sampling (CVS) and amniocentesis to test the baby's chromosomes. Testing is generally very accurate, although not all of these tests are available in all parts of the world.

An informal survey

has shown that as a group children are sociable and affectionate. They seem to be good at music, especially singing. Some children have a remarkable memory.

Inform Network Support

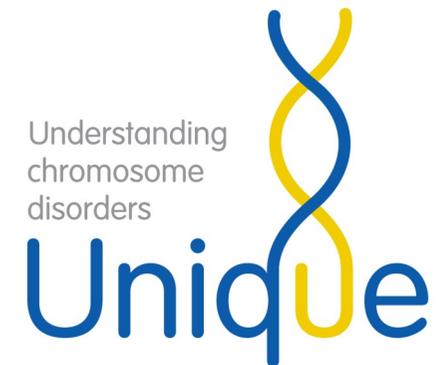


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This leaflet is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication. It was compiled by Unique and reviewed by Professor Koenraad Devriendt, University of Leuven, Belgium, Dr John Barber, Deputy Director, Wessex Regional Genetics Laboratory, Salisbury, UK and by Professor Maj Hultén BSc, PhD, MD, FRCPath, Professor of Medical Genetics, University of Warwick, UK.

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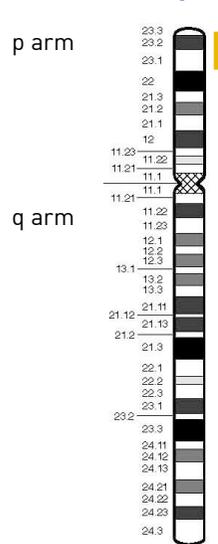


8p23 deletions



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What are 8p23 deletions?



An 8p23 deletion means that the cells of the body have a small but variable amount of genetic material missing from one of their 46 chromosomes - chromosome 8. Chromosomes come in different sizes, each with a short (p) and a long (q) arm. They are numbered from largest to smallest according to their size, from number 1 to number 22, in addition to the sex chromosomes, X and Y. People with an 8p23 deletion have lost genetic material from the end of the short arm of one of their two chromosome 8s, leaving the other one intact. The majority of deletions of 8p23 are terminal. This means that the tip of the long arm is included in the deletion. However, just under half of reported 8p23 deletions are

Chromosome 8

Chromosomes contain the genes that instruct the body to develop and work properly

interstitial, in which a piece of the short arm of chromosome 8 is missing, but the end of the chromosome is still present. Some people show no obvious effects from this loss, while others face a range of difficulties. These features are common. Not every child will have all of them but nearly all will have some.

- Slight delay in physical (motor) development
- Learning difficulty. Some children are scarcely affected while others have mild to moderate difficulty
- Unusual behaviour (often including hyperactivity and impulsiveness)
- Heart conditions
- Some children may have seizures.

Development

■ Motor development

Motor development is typically slightly delayed but most children can walk by the age of 2 and acquire other skills like swimming, despite underlying muscle weakness in some. Problems with balance hold back skills like cycling and co-ordination challenges make ball games and certain sports difficult. Practice and expert physiotherapy help children to overcome most of their problems by their teen years.

■ Learning

Most children will need some support with learning but will learn to read and write and to cope with simple mathematics. Abilities are scattered but many children cope in a mainstream school at least for the early years of primary education and some children may have average intelligence. Children who have attention problems are likely to need more learning support.

■ Speech

Children usually speak a little late and take longer than others to talk fluently. Some children speak unclearly but are helped by speech therapy. By the age of 8 to 10 most children speak well but a few continue to need help.

■ Behaviour

Children may have a strikingly similar pattern of behaviour. This is not universal and it eventually fades. Typically children have a warm and affectionate temperament but show sudden and extreme mood changes, with outbursts of aggression and destruction. Their frustration tolerance is low, they are determined and tend to oppose both adults and other children. They may additionally be sensitive to stress and hyperactive.

How common are 8p23 deletions?

No one knows, but they are more common than has been thought because some people with no real problems are never identified. Unique has more than 50 affected members with a 'pure' 8p23 deletion (no other chromosome is involved) and more than 50 have a loss or gain of material from another chromosome arm as well as an 8p23 deletion.

Medical concerns

■ **Heart conditions** affect around half of children, so all need a thorough cardiac examination. Both simple problems and complex faults in the structure and functioning of the heart can occur. Problems such as open ducts and small holes may heal naturally with age. Those children who do need surgery are typically fit and healthy afterwards. Heart conditions are more likely, although not inevitable if the deletion includes the GATA-4 gene located in 8p23.1 (see chromosome diagram opposite).

■ **Seizures** appear to be more common than among other children but are usually well controlled with anti-epileptic medicines.

■ **Unusual genital features.** Boys may be born with undescended testicles and hypospadias - where the hole is situated on the underside of the penis - is common. A surgical operation can correct these problems if necessary.

■ **Vision.** A squint (strabismus) affects around a quarter of Unique children. Long sight and short sight have also been reported.

■ **A congenital diaphragmatic hernia (CDH),** affects around a fifth of Unique children, where there is a hole in the muscular wall (the diaphragm) which separates the heart and lungs from the contents of the abdomen, which may allow some of the contents of the abdomen (including the stomach, intestines, spleen and liver) to move up through the hole and into the chest cavity potentially depriving the lungs of space to develop properly. This means that the lungs may be smaller than they should be.

