Different types of 8p duplication

The effects of 8p duplications depend mostly on what material has been duplicated and the exact points (breakpoints) where the chromosome has broken. Your geneticist or genetic counsellor can explain where the breakpoints are and this may help in predicting the outlook for your child.

Inv Dup Del 8p

Some people have a disorder called inverted duplication and deletion of 8p, inv dup del 8p for short. *Unique* publishes a separate leaflet on Inv Dup Del 8p.

Natural variants: 8p22 and 8p23.1

In some families, extra material is seen within band 8p23.1 which resembles a duplication but turns out on closer inspection to be a harmless variant. Most people have a total of four copies of a particular segment within 8p23.1, but the number of copies can vary between three and, rarely, 12. People with high copy numbers are healthy and develop normally and the unusual chromosome findings normally come to light by chance. The expanded material contains three genes that are important players in the immune system and these people may have an enhanced resistance to infection.

There is another normal variant within band 8p22 which was recently found in three generations of the same family.

Small, harmless duplications that run in families

Harmless duplications have been found in some families within the region 8p22 to 8p23.1 and 8p23.1 to 8p23.3. Harmless duplications of a particular stretch of the 8p23.2 band which contains few genes have also been found and may be another harmless variant.

Other small duplications in families

In some families a healthy parent can pass a duplication between bands 8p21.3 and 8p23.1 to a child who has some developmental delay or birth defects, particularly involving the heart. This can occur when parents have the duplication in all of their cells or only in some of them (a condition called mosaicism).

Most of these family duplications have been found in chromosome material at the upper end of the short arm (see diagram overleaf) but one report involved a relatively harmless duplication between 8p12 and 8p21.1.

Inform Network Support

Understanding chromosome disorders Unique

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When you are ready for more information, *Unique* may be able to help. We can answer individual queries and we also publish a more detailed leaflet about duplications of 8p.

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 and the content of the full leaflet on which this text was based was verified by Dr John Barber, Wessex Clinical Genetics Service and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

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Duplications of 8p

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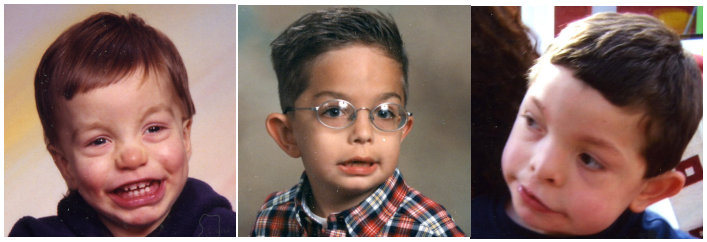


What is an 8p duplication?

An 8p duplication is a rare genetic condition in which there is an extra copy of some of the material from one of the body's 46 chromosomes.

Chromosomes are the structures in the nucleus of the body's cells that carry genetic information. There are 23 pairs of chromosomes, one of each pair having come from the mother and the other from the father. Chromosomes are numbered 1 to 22 according to size and the 23rd pair are the sex chromosomes. Each chromosome has a short (p) arm and a long (q) arm.

People with an 8p duplication have an extra copy of some material on the short arm of one of their chromosomes 8. Duplication of the entire arm can be called **trisomy 8p**.

Effects

The precise effects of gaining material from a chromosome depend on how large the duplication is, how many genes it contains and what they do. The effects may not be limited to the genes within the duplication because these genes may interact with others on the same or other chromosomes.

With 8p duplications the picture is very varied indeed, depending on which part of the chromosome is duplicated and whether the same duplication is present in one of the parents. Some people are healthy and develop normally, but others are mildly or more profoundly affected by their chromosome disorder.

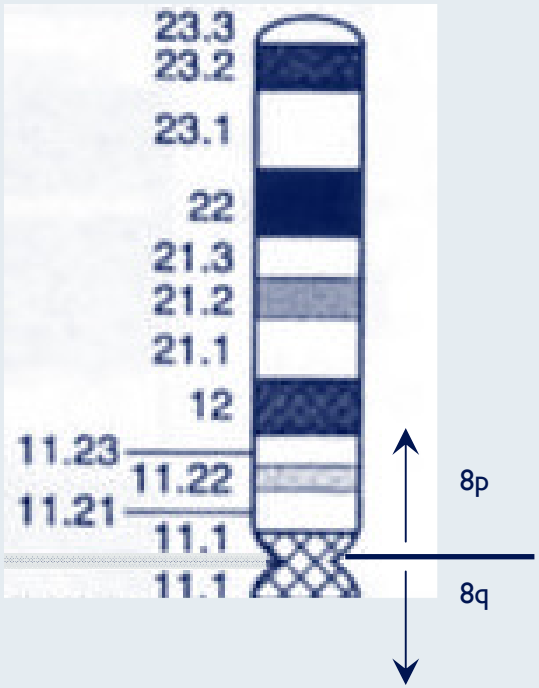
Why did it happen?

A test of the parents' chromosomes will show whether the 8p duplication is inherited or has occurred out of the blue. If it has occurred out of the blue, the actual cause is not known but should be regarded as an event that happened in cell division in the process of making sperm or egg cells. These events are not uncommon. They affect children from all parts of the world and from all types of background. They also occur naturally in plants and animals. So there is no reason to suggest that your lifestyle or anything that you did caused the duplication.

Can it happen again?

So long as tests show that the parents' chromosomes are normal, they are very unlikely to have another affected child. If tests show that one of the parents has rearranged chromosomes, it is best to discuss the inheritance pattern in your family with a geneticist or genetic counsellor.

Chromosome 8p



Overleaf (Different types of 8p duplication) you will find a list of specific 8p duplications, some harmless, others not. The features described on this page below and right are the most common ones associated with other general duplications and trisomies of 8p.

- Some level of developmental delay.
- A degree of learning difficulty.
- Floppiness of the skeletal muscles in babies (hypotonia).
- Internal organs are usually unaffected, but one child in five is born with an anomaly of the structure of the heart.
- Absence or thinning of the band of nerve fibres that joins the two hemispheres of the brain. This is called agenesis of the corpus callosum and is discovered when a child has a brain scan.

Development

Mobility and activity

As babies, children with 8p duplications may have hypotonia (low muscle tone) and can be late to achieve skills such as holding their head steady, sitting, crawling and walking. There is a wide range in the ages at which individual children acquire these skills. In *Unique*'s experience, children rolled over at 7 months to 3 years, sat without support at 8 months to 3 years and crawled or shuffled at 11 months to 7 years, although some children by-passed this stage. Generally children walked at 2 to 3 but a few did so earlier and some children had not yet walked at 6 to 8 years or walked and then lost the skill.

Learning

The chromosome disorder will affect the speed at which many children learn and set some limits on their eventual achievement. Just how slight or far reaching the effects are will become clear as your child develops, but the possible range is really very broad, from no effect at all in children who have or inherit a small, stable duplication to a more profound effect in others. It is not always possible to predict from the chromosomes what the effects on learning will be, but by your child's first birthday it will be easier to suggest the outlook from the chromosomes, the results of clinical investigations and observations of your child's development.

Speech and communication

Speech and language delay appears to be in line with children's ability to learn. In *Unique*'s experience of affected children, first words typically emerged at 2 to 5 years.

Behaviour

It is uncertain whether a particular behaviour pattern is typical but from parental report, children are usually happy and sociable. From around school age, occasional bouts of oppositional behaviour may develop. Children with chromosome disorders are subject to the same influences as other children, but the effects tend to be less modulated. *Unique* families report that challenging behaviour typically developed in response to stress or fatigue and was especially evident at puberty. Any behavioural difficulties should be followed by a psychologist, psychiatrist or developmental paediatrician who can help provide guidance, behavioural intervention strategies and medications if necessary.