

Why did this happen?

A 9p deletion can occur as a result of rearrangements in one parent's own chromosomes or it can happen out of the blue, so the child with the chromosome disorder is the only affected person in the family. If an analysis of the parents' chromosomes reveals a structural rearrangement, this is usually balanced so that all the chromosome material is present, and the parent is then almost always healthy. Occasionally the rearrangement will be the same as in the child, and again the parent may be healthy.

How did this happen?

Changes to the structure of chromosomes such as 9p deletions occur most often during the cell divisions that lead to the creation of eggs or sperm. They affect children from all parts of the world and from all types of background. They also happen naturally in plants and animals. So there is no reason to suggest that your lifestyle or anything that you did caused the loss of chromosome material.

Can it happen again?

Where both parents have normal chromosomes, it is unlikely that another child will be born with a 9p deletion. Where a parent has a rearrangement of their chromosomes, the risk of having another affected child is higher. Where one parent has the same 9p deletion as the child, the risk of passing it on can be as high as 50 per cent.

How rare are 9p deletions?

Chromosome 9p deletions are reported to occur in 1:50,000 newborn babies, of whom two thirds are girls. Studies of more than 100 people have been published but there are undoubtedly many more.

The oldest patient described was 61 years old. At the time of writing, *Unique* had more than 40 member families with Alfi's syndrome or a similar deletion from 9p. Members who wish to can have contact with other affected families.

Inform Network Support



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9p- Syndrome Network

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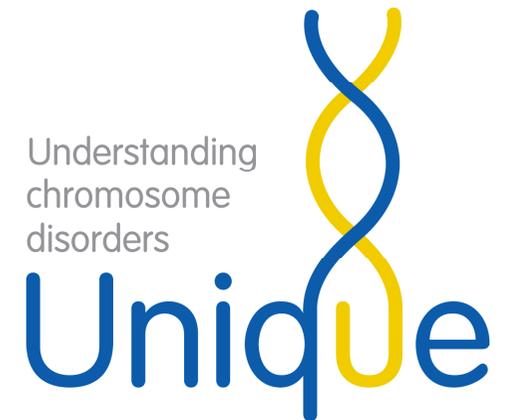
Unique mentions other organisations' message boards and websites to help families looking for information. This does not imply that we endorse their content or have any responsibility for it. When you are ready for more information, Unique may be able to help. We can answer individual queries and we also publish a more detailed leaflet about 9p deletions.

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 Conny van Ravenswaaij, clinical geneticist and cytogeneticist, Dr Marielle Swinkels, University Medical Center, Nijmegen, Netherlands and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

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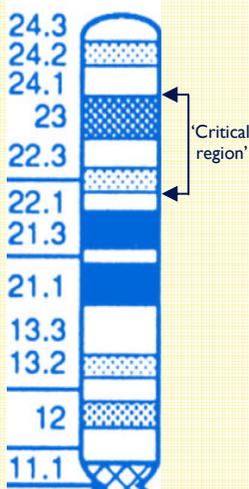


9p deletions

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What are 9p deletions?



Chromosome 9p

A chromosome 9p deletion is a rare genetic condition in which there is a missing copy of part of the genetic material that makes up one of the body's 46 chromosomes.

Chromosomes are the microscopically small structures in the nucleus of the body's cells that carry genetic information. They come in different sizes, each with a short (p) and a long (q) arm. The likelihood of problems developing depends very much on which genetic material is missing. A geneticist or genetic counsellor can tell you exactly how much material has been lost and where the breakpoints are in the chromosome.

Chromosome 9 can break almost anywhere along the short arm, but in many people the breakpoint is in the region called 9p22. People with a break in this region that deletes a so-called 'critical region' at 9p22-3 are said to have 9p- or Alfi's syndrome.

Main features

People with apparently exactly the same chromosome rearrangement can vary widely. Each child is unique – but with a 9p deletion, the likelihood of showing one or more of these features increases.

- Developmental delay
- Unusual head shape with the forehead pointing forwards like a keel. The bone plates that form the forehead joins early (premature fusion of metopic suture)
- Low muscle tone, making the body floppy

Other features

- Structural heart abnormalities
- Hernias (an organ protrudes out of its normal position), especially near the navel or in the groin
- Spinal curve
- Respiratory problems and ear infections in early childhood
- Unusual genital features

Development

Sitting, moving, walking ...

Babies are typically floppy (hypotonic) at birth and are late to develop muscle control. Rolling over may be achieved at 3 months to 3 years, supported sitting at 8 months to two years and crawling or bottom shuffling at 1-3 years.

Supported walking is generally achieved by school age but some children need a further year or two to walk alone.

Mobility levels are varied, with some children swimming and cycling with ease. For almost all children, hypotonia improves with therapy, exercise and maturity.

Learning

Most children need some support with learning but the range of ability is wide and generally the outcomes have been good with some adults in supported work.

The spectrum of learning difficulty ranges from mild to severe but within *Unique* the great majority of families have described their child's special learning needs as moderate. A small study has suggested specific difficulties with certain visual skills and relative verbal fluency.

Speech and communication

Typically, people with 9p deletions speak well and children may develop an adult style of speaking. Communication is a relative strength and most children talk, sometimes fluently, by school age. First words are generally expected to appear between a child's second and third birthdays, with understanding usually more advanced than expression. Researchers who recently surveyed children with 9p deletions found that they understood very much better than they could express themselves and this caused a frustration that could be vented as temper tantrums. As speech improved, tantrums subsided. They stress that it is important to encourage communication using means such as sign language until speech emerges and alongside it.

Behaviour

Children are generally regarded as affectionate and sociable. When denied their own way, frustrated or required to do something they do not want, they may react with fierce temper tantrums. These eased with age and increasing speech fluency, but many families have sought professional help. Behavioural difficulties should be followed by a psychologist, psychiatrist or developmental paediatrician, who can help provide guidance, behavioural intervention strategies and medications if necessary.

Medical concerns

Head

Some babies are born with a narrow, pointed forehead, shaped like the keel of a boat when viewed from on top. The seam between the bone plates of the forehead (the metopic suture) fuses early, making the skull triangular (trigonocephaly). A minor degree of early fusion may leave the forehead a normal shape but with a ridge down the middle. Affected babies will be assessed by a craniofacial team and if necessary the bones of the forehead can be separated in a surgical procedure.

Nasal passages and palate

The passages at the back of the nose and throat may be unusually narrow (choanal atresia) and many children have a high-arched palate (roof of the mouth). If necessary, the nasal passages can be surgically widened.

Ear infections

Ear infections are common and children may develop glue ear, with a transient hearing loss caused by fluid in the middle ear. Long standing glue ear can be treated by inserting grommets (tubes) into the ear drum.

Chest/ respiratory infections

Some children are prone to chest and throat infections and persistent wheezing, especially as babies and very young children. When they catch an infection, children may be more unwell than others without a chromosome disorder.

Heart

Babies will have a careful heart exam and more detailed investigations if abnormal heart sounds are heard. Abnormalities of the structure of the heart have been found in some babies, but in most they resolved naturally. Examples include holes between the lower or upper chambers of the heart (ventricular septal defects, VSDs or atrial septal defects, ASDs).

Hernias

Hernias of the umbilicus or in the groin (inguinal) are more common in children with 9p deletions than in other children. If necessary the hernia can be corrected with surgery.

Genital area

Genes believed to play a role in sex differentiation are found near the end of 9p. Losing these genes is believed to disrupt genital development. A wide spectrum of effects has been observed and within *Unique* most children show no effects.