

Why did this happen?

Rearrangements occur in chromosomes as part of evolution. 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 material from chromosome 3.

In most families, the 3p25 deletion occurs out of the blue (*de novo*). In a minority of families, a chromosome study shows that one of the parents has a rearrangement of their own chromosomes that gave rise to the deletion when egg or sperm cells were formed.

Can this happen again?

The risk of having another child affected by a rare chromosome disorder depends on the parents' chromosomes. When a 3p25 deletion occurs out of the blue (*de novo*), your chances of having another affected child are almost certainly no higher than for anyone else in the population. If a chromosome study shows that one parent has a rearrangement of their own chromosomes or, as is very occasionally the case, one parent has the same 3p25 deletion as the child, the chances of it happening again are much higher. Each family's situation is different and a clinical geneticist can give you specific advice for your family.

How common are 3p25 deletions?

They are a rare chromosome disorder. Only around 25 people have been described in the medical literature. At the time of writing, *Unique* had 34 members with a 3p25 deletion and can put families in touch with each other if they want contact.

Families say ...

- "Every day she achieves something new makes it all worth while. Also, she has a great sense of humour. When we look at each other, we just connect and that is something that only I, her mother, have with her - age 5.*
- "She has become a very loving child and is generally very happy, enthusiastic and likes school - age 7.*
- "He is very loving, always greets us with a smile - age 14.*

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For support,
contact with other families and information
**Rare Chromosome Disorder
Support Group**
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When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a more detailed leaflet about the effects of 3p25 deletions.

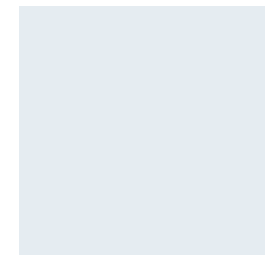
This information sheet is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication and the medical content has been verified by Professor Eamonn Maher, Professor of Medical Genetics, University of Birmingham, UK, 2004 and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, UK, 2005.

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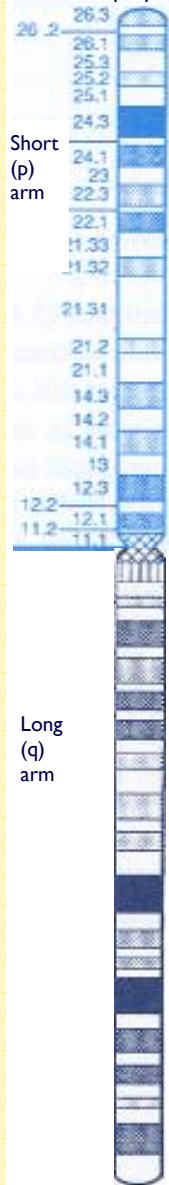
3p25 deletions



What is a 3p25 deletion?

Chromosome 3

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



People with a 3p25 deletion have lost chromosome material from the end of the short arm of one of their two chromosome 3s. The other chromosome 3 is intact but the missing genetic material causes the symptoms of this rare chromosome disorder.

Some people have one break in the chromosome, while others have lost a segment out of it.

Frequent features

The most common features in children with a 3p25 deletion are also found in many other chromosome disorders. Almost all children have the first three features. The other features are also typical of this chromosome disorder but an individual child may or may not have them.

- Low birth weight and, in most children, slow growth and eventual short height
- Microcephaly (small head)
- Delay in reaching baby 'milestones'
- Speech delay or absence of speech
- Hypotonia - floppiness
- Vision problems
- Ptosis – an inability to fully raise the upper eyelid
- Hearing impairment, in some children temporary
- Extra fingers and/ or toes
- Heart conditions
- Triangular face with a small chin
- Almost all children also have a degree of learning difficulty.

Development

■ Growth

Almost all babies are small for dates and some are also born one or two months early. While many babies remain small and are short as children, some babies grow well and are of average height.

■ Feeding

Parents are likely to need support as feeding difficulties can be considerable at first. Typically, babies suck weakly and some need high energy milks to encourage weight gain. Many babies readily bring feeds back (gastro oesophageal reflux) and need careful positioning for feeding and while sleeping. Some babies are helped by medicines for reflux. Occasionally surgery is helpful to improve the effectiveness of the valve between the stomach and the food pipe (a fundoplication). Some babies need to be fed temporarily through a tube direct into the stomach (gastrostomy). Older children typically have chewing difficulties.

■ Sitting, moving, walking

Babies are usually quite late to become mobile. With the help of standing and walking aids, some children learn to walk and eventually to swim, run and dance but for others this is not possible.

■ Speech

Children typically experience delay in communicating and learning to use words. The eventual range of achievement is very broad, from a few children who have a large vocabulary to others who use signing, gestures and vocal noises to express their needs.

■ Learning difficulties

Most children need very considerable support with learning. A few learn to write and read but non-academic skills will be more important in their lives.

■ Behaviour

As a group, children appear to have a happy disposition. Some families have noticed autistic tendencies in young children and hyperactivity at school age, but this has been well controlled with medication.



“ He has brought us a great deal of love and happiness - age 18 months.

Medical concerns

These disorders have been found in a group of babies and children with a 3p25 deletion. They are not found in all babies and your child may not be affected.

■ Heart conditions

Around one baby in three is born with a heart condition, typically an atrioventricular septal defect (AVSD). This is a group of defects (also known as endocardial cushion defects) affecting the development of the walls between the two upper and the two lower chambers of the heart (the atria and ventricles) and the valves that control the blood flow between them. Many babies will need open heart surgery but *Unique's* experience is that they thrive afterwards.

■ Seizures

Quite a few babies will have seizures, although these may be rare or occasional.

■ Ptosis

Some babies will not be able to open their upper eyelids fully on one side or both. Very mild ptosis does not need treatment but if vision is obscured, surgery can be performed to hold the eyelid up.

■ Eyesight

In *Unique's* experience, many babies have eyesight problems. However, the great majority can be corrected with surgery (such as strabismus, a squint) or with glasses (such as short sight).

■ Hearing

Hearing impairment, either temporary or permanent, affects many children.

■ Extra fingers and/ or toes

These are fairly common but can be removed and do not usually cause any long-term problems.

■ Genitals

Some boys have hypospadias, where the hole usually at the end of the penis is on the underside, and some are born with undescended testicles. Both conditions may need no treatment but if they do, they can usually be corrected with straightforward surgery.