Why did this happen?

Most chromosome disorders arise out of the blue from an event that occurred when the sperm and egg cells were forming. This is part of a natural process and as a parent there is nothing you can do to control it. No environmental, dietary or lifestyle factors are known to cause deletions of the short arm of chromosome 6. So there is nothing that could have been done before or during pregnancy to cause this and nothing that could have been done to prevent it.

Can this happen again?

Where both parents have normal chromosomes, it is unlikely that another child will be born with a 6p deletion or any other chromosome disorder. Very rarely (less than 1%), both parents have normal chromosomes by a blood test, but a few of their egg or sperm cells carry a 6p deletion. This is called germline mosaicism and it means that parents whose chromosomes appear normal when their blood is tested can have more than one child with the deletion.

In families where the 6p deletion has been inherited from a parent, the possibility of having another child with the deletion, either a girl or a boy, rises to 50% in each pregnancy. However, the effect of the deletion on the child's development, health and behaviour cannot be reliably predicted. Your genetics centre should be able to offer counselling before you have another pregnancy.

How common are 6p deletions?

6p deletions are very are, however, with the increased availability and use of genetic testing, more families with 6p deletions are being identified. When this information was compiled in 2004, only 43 people had been described in the published medical literature and *Unique* had 25 members. *Unique* now has 129 members with a 6p deletion (2018) and many more families have been, and continue to be identified worldwide.

Families say ...

" L is a generous person who delights in giving and puts a tremendous amount of time and thought into selecting the right gift. " - Age 20.

" C is generally very happy and loving but there is always a sense of sadness and frustration. " - Age 14.

" H likes to be popular and to entertain people. " - Age 9.

Inform Network Support

Unique
Understanding Chromosome & Gene Disorders

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Chromosome 6 research project

The C6 project works with families to collect detailed information with the aim of linking specific disease characteristics with specific regions of chromosome 6
https://www.chromosome6.org/

When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish another leaflet about the effects of 6p deletions from 6p25 to the end of the chromosome.
This guide 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. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. *Unique* does its best to keep abreast of changing information and to review its published guides as needed. This guide was verified by Dr Ordan Lehmann, Associate Professor, Departments of Ophthalmology & Medical Genetics, University of Alberta and by Unique's Chief Medical Adviser in 2004.

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6p deletions

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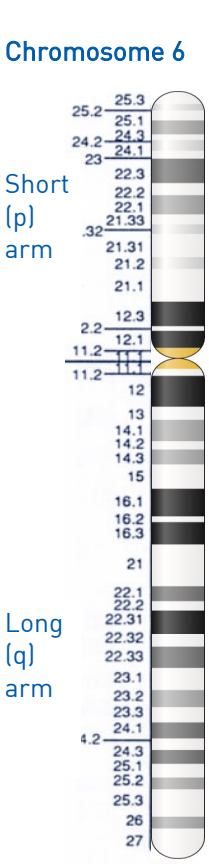
What are 6p deletions?



People with a 6p deletion have lost a small but variable amount of chromosomal material from the short (p) arm of one of their chromosome 6s. Different deletions contain different genetic information (genes). Some people with a 6p deletion may not be obviously affected while others may have developmental and medical concerns; this can be dependant on the missing genetic information.

The short arm of chromosome 6 contains hundreds of different genes. The loss of specific genes can affect people in relatively predictable ways. However, other genes and individual personalities help to determine each individuals development, needs and achievements.

Loss of genetic material from within the chromosome is called an interstitial deletion. Loss of material from the end of the chromosome is called a terminal deletion. Terminal means *from the end*, it does not mean that it's lethal.

Your geneticist will tell you about your child's deletion, where the chromosomal breakpoints are, and if any important genes are included in the deletion.

Deletions within the short (p) arm of chromosome 6 (interstitial deletions)

Possible features:

- Developmental delay
- Short neck with excess skin folds
- Eye anomalies
- Hand anomalies
- Heart defects

Deletions from the end of the short (p) arm of chromosome 6 (terminal deletions)

Possible features:

- Developmental delay
- Defects of development of the front portion (anterior chamber) of the eye
- Hearing loss
- Heart defects
- Wide set eyes (hypertelorism)

Development

Motor development

Children can be late to sit and walk but then go on to become fully active, cycling, swimming and dancing for fun. A minority do not learn to walk smoothly and tire easily, so may rely on a wheelchair for outdoor mobility.

Learning

Children face a very variable degree of learning difficulty ranging from no learning difficulty to mildly to severely affected. Those with moderate to severe learning difficulties may still learn to read enough to help with daily living.

Speech

Speech and language can be delayed and children who also have hearing loss may be more severely affected. Typically children start to talk at two or three years and the use of sign systems may help them communicate.

Behaviour

Children can be sociable and very friendly but also become anxious and lack self confidence. When they are frustrated and unable to express their needs some children may have aggressive outbursts. There is no evidence that 6p deletions make people more vulnerable to mental illness.

Medical conditions

Eye problems

Some children with a 6p deletion have subtle changes to the front portion of the eye. This can result in glaucoma, a condition in which the pressure inside the eye becomes raised. Monitoring by an ophthalmologist, a doctor who specialises in eye disorders, will ensure appropriate treatment is considered.

Hearing loss

Glue ear and conductive (nerve) deafness have been identified in some children with a 6p deletion. Treatment by insertion of T-tubes may be enough to rectify hearing difficulties but some children may require hearing aids.

Heart conditions

Heart conditions have been associated with some 6p deletions and may reduce activity levels and slow down a child's growth. Among *Unique* members, all heart problems resolved naturally or were successfully treated with surgery.

Lax or dislocated joints

Some children with a 6p deletion have lax joints that may be prone to dislocation. Joints may be supported with orthoses (braces/supports), adapted footwear or splints. Occasionally surgery is needed.

Kidney problems

Kidney anomalies are known to occur in some people with interstitial 6p deletions so it may be recommended that babies and children undergo a kidney examination.

Umbilical and inguinal hernias

Small hernias near the belly button (umbilical) and in the groin (inguinal) have been identified in some children with a 6p deletion, and may require surgery.

Dry skin and eczema

Some babies and children with a 6p deletion are known to have dry skin and eczema. If regular moisturising does not control this, restrained use of steroid creams prescribed by your child's doctor may help.

Genital anomalies

Occasionally, genital anomalies are identified in children with a 6p deletion. They appear to be more common in boys than girls but are usually minor. Occasionally surgery may be needed. Undescended testicles are common.