

## Why?

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 you did before you were pregnant or during pregnancy that caused this and nothing you could have done to prevent it.

## Can this happen again?

6p deletions usually happen out of the blue (de novo). You are then no more likely to have another child with a chromosome disorder than anyone else. Occasionally one parent is a carrier of a chromosome rearrangement that only becomes apparent in the child. A blood test to check the parent's chromosomes will tell you if either parent carries a chromosome rearrangement.

## Prenatal diagnosis

6p deletions can be detected during pregnancy by examining prenatal chromosomes obtained by chorion villus sampling (CVS) or by amniocentesis, followed by a FISH test for confirmation. Some physical anomalies associated with a 6p deletion such as heart or kidney defects can be seen on an ultrasound scan by 18 to 20 weeks of pregnancy.

## How common are 6p deletions?

There is no official estimate, but they are rare. When this information was compiled, only 43 people had been described in the published medical literature and *Unique* has 20-25 members.

## 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*

- L, age 20.

*C is generally very happy and loving but there is always a sense of sadness and frustration*

- C, age 14.

*H likes to be popular and to entertain people*

- H, age 9.

## Inform Network Support



## Rare Chromosome Disorder Support Group,

G1 The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK

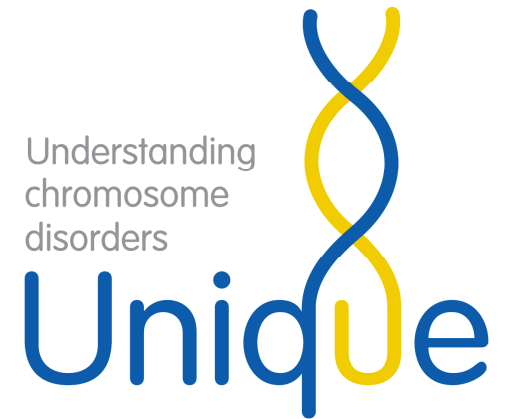
Tel/Fax: +44(0)1883 723356

[info@rarechromo.org](mailto:info@rarechromo.org) | [www.rarechromo.org](http://www.rarechromo.org)

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 Dr Ordan Lehmann, Associate Professor, Departments of Ophthalmology & Medical Genetics, University of Alberta and by Unique's Chief Medical Adviser 2004.v2 (PM)

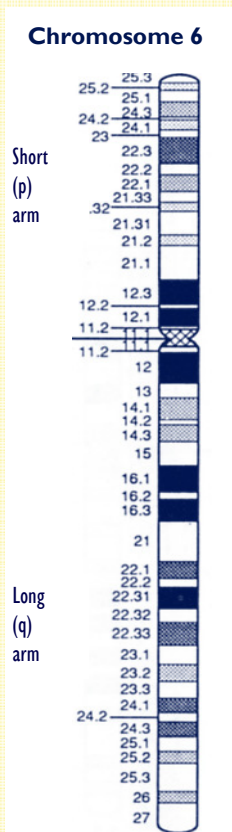
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# 6p deletions

[rarechromo.org](http://rarechromo.org)

## What are 6p deletions?



People with 6p deletions have lost a small but variable amount of chromosome material and genes from the short arm of one of their chromosome 6s. The loss of specific genes affects people in relatively predictable ways. However, other genes and personality help to determine a child's future development, needs and achievements.

People who lose a segment from within the chromosome have an interstitial deletion. People who lose material from the end of the chromosome have a terminal deletion. Terminal means *from the end*. It does not mean that it's lethal. There are many reports of adults with 6p deletions and *Unique* has a number of adult members.

Your geneticist will tell you about your child's deletion and where the breakpoints are in the chromosome.

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

#### Common features:

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

## Development

### ■ Motor development

Children are usually late to sit and walk but then go on to become fully active, cycling, swimming and dancing for fun. A minority never walk smoothly and tire easily, so they rely on a wheelchair for outdoor mobility.

### ■ Learning

Children face a very variable degree of learning difficulty. One child with a very tiny deletion has no learning difficulty but others are 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 are particularly delayed as many children also have a hearing loss. Typically children start to talk at two or three years and most use sign systems to help them communicate. A few children speak fluently and clearly enough to be understood outside the family.

### ■ Behaviour

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

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

#### Common features:

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

## Medical conditions

### ■ Eye problems

A proportion of children with these chromosomal rearrangements 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, in perhaps up to half of all children. Monitoring by an ophthalmologist, a doctor who specialises in eye disorders, will ensure appropriate treatment is begun.

### ■ Hearing loss

Glue ear and nerve deafness are common. Inserting T-tubes to treat conductive deafness may be enough but most children learn to communicate very much better with hearing aids.

### ■ Heart conditions

Heart conditions are common and may reduce activity levels and slow down growth. Among *Unique* members all heart problems resolved naturally or were successfully treated with surgery.

### ■ Lax or dislocated joints

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 and all babies and children can expect to be examined for these.

### ■ Umbilical and inguinal hernias

Hernias near the belly button (umbilical) and in the groin (inguinal) are usually small but may need surgery.

### ■ Dry skin and eczema

This appears to affect a significant number of babies, from as early as three weeks. If regular moisturising does not control this, restrained use of steroid creams prescribed by your child's doctor will help.

### ■ Genital anomalies

Genital anomalies are more common in boys than girls but are usually minor. Occasionally surgery may be needed. Undescended testicles are also common.