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.

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.
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

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.