

### Why did this happen?

A chromosome 4p duplication usually happens out of the blue, and the child with the chromosome disorder is the only affected person in the family.

The duplication can also occur as a result of a rearrangement in one parent's own chromosomes. 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.

In either case, the cause of the original chromosome rearrangement is not known and it should be regarded as an accident that happened in the process of making sperm or egg cells. These accidents are not uncommon and can 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 duplication.

### Can it happen again?

Where both parents have normal chromosomes, it is unlikely that another child will be born with a 4p duplication. Where a parent has a rearrangement of their chromosomes, the risk of having another affected child is higher. You should be able to discuss this with your genetics service.

### How common are 4p duplications?

As isolated chromosome rearrangements, 4p duplications are very rare. They occur more often as part of an unbalanced chromosome arrangement, often involving loss of material from another chromosome.

At the time of writing this leaflet, *Unique* had a small community of 13 member families affected by a pure 4p duplication and very many more in whom a 4p duplication forms part of a more complex chromosome rearrangement. For details of specific 4p duplications, look on the *Unique* website at [www.rarechromo.org](http://www.rarechromo.org). Members who wish to can have contact with other affected families.

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For support,  
contact with other families and information  
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**[www.rarechromo.org](http://www.rarechromo.org)**

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 duplications of 4p.

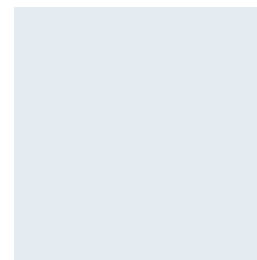
This leaflet is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication and the genetic content has been verified by Dr Laila Zahed, American University of Beirut, Lebanon and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, *Unique*'s chief medical advisor, 2005.

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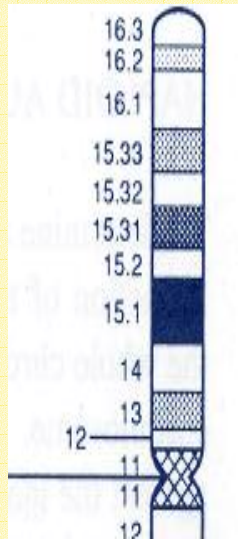
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# Duplications of 4p



## What are 4p duplications?



Chromosome 4p

A chromosome 4p duplication is a rare chromosome disorder in which there is an extra copy of part of the genetic material that makes up 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. People with a 4p duplication have an extra copy of part of the material in the short arm of one of their chromosomes 4. The other chromosome 4 is the usual size. Chromosome 4p duplications are sometimes called **trisomy 4p**.

### Main features

People with apparently the same chromosome rearrangement can vary widely, even within the same family. Each individual is unique, but with a 4p duplication, the likelihood of one or more of these features increases.

- Developmental delay
- Variable degree of difficulties with learning
- Small head
- Slow growth as a baby, usually with a normal birth weight
- Some joints may be contracted
- Unusual genital features in boys

### Other features

These features have also been noticed in some babies..

- Unusual muscle tone. In some babies tone is low and a baby feels floppy to hold (hypotonia). Other babies feel taut (hypertonia).
- Recurrent respiratory infections
- Unusual foot development
- Feeding difficulties in the newborn period

## Development

### ■ Growth

Babies are usually born a normal weight but grow slowly as children. Some catch up at puberty and grow into average to tall adults, sometimes with a large build. This is not universal, though, and some adults are under 150cm (five feet) tall.

### ■ Mobility and activity

Some delay in mobility is to be expected, although the range is broad. In *Unique's* experience, babies have mastered rolling over between two and 11 months; sitting upright has developed between nine and 19 months; becoming mobile by crawling or bottom shuffling between eight and 20 months and walking between 18 months and five years.

### ■ Learning

Children can be expected to need some support with their learning. For a few children learning difficulties will be mild, even when the duplication is large. Most children, however, have moderate to severe learning difficulties but can still grow into adults who will function in society with support.

### ■ Speech and communication

Most children are likely to experience a delay in speech and language in line with their learning difficulties. An individual child's ability to communicate depends on many factors – their level of understanding, their ability to co-ordinate the actions of speech, gesture or signing and their need to communicate. Speech therapy is helpful and should be available.

### ■ Behaviour

No consistent pattern of behaviour has been found, although most families have described young children as well-mannered, happy, alert and responsive, if timid. During childhood, challenging behaviour and mood swings may develop and some youngsters have displayed obsessive behaviours. Behavioural difficulties should be followed by a psychologist, psychiatrist or developmental paediatrician, who can help provide guidance to the family, behavioural intervention strategies and medications if necessary.

Generally speaking, the risk of major birth defects and illness does not appear to be significantly raised. The most consistent problem is with joints. In *Unique's* experience, other problems affect only a minority of people with a 4p duplication.

### ■ Joints

Joints including the fingers, knees, hips, elbows and ankles can be stiff and contracted. Hip dislocation has also been seen, as has radio-ulnar synostosis, where the two bones of the forearm are fused.

The experience of *Unique* members is that effects were usually mild or moderate and were significantly helped by regular physiotherapy to keep joints mobile and supple.

### ■ Feet

At birth the feet may be seen to be unusually positioned or formed. The most common conditions appear to be club foot (talipes equinovarus, in which the foot points downwards and inwards) or rocker bottom feet (the sole of the foot is curved like a rocker). If physiotherapy is not appropriate, surgery can improve function to achieve the best position possible for walking.

### ■ Genital features

For boys, although not apparently for girls, there is an increased risk of being born with one or more unusual genital features. These are typically minor and can be corrected with surgery or may need no correction. The hole at the end of the penis may be on the underside instead (hypospadias), the penis may be unusually small, and the testes that usually descend into the scrotum before birth remain in the abdomen.

### ■ Respiratory infections

Respiratory infections in young children are more common than in children without a chromosome disorder. Babies with gastro oesophageal reflux are vulnerable to a type of chest infection called aspiration pneumonia caused by inhaling part of feeds that return into the gullet. Your child's doctors will suggest sensible protective measures against respiratory infections, including routine immunisations.

### ■ Spinal curvature (scoliosis)

The spine may develop a degree of curvature but degree generally appears to be mild.

### ■ Eyesight

Strabismus (squint) is common and ptosis (hooded upper eyelid) also occurs, but most people have normal vision. Both ptosis and strabismus can be corrected surgically if necessary.