Why did this happen?

Chromosome conditions are usually passed down in the sperm and egg cells. No environmental, dietary or lifestyle factors are known to cause them. So there is nothing you did before you were pregnant or during pregnancy that caused this to occur and there is nothing you could have done to prevent it. Both parents will be offered a chromosome and FISH test and if the results are normal, the 17p duplication has almost certainly occurred out of the blue (*de novo*). Occasionally a rearrangement of one parent's chromosomes is revealed but one that is balanced so that all the chromosome material is present, and the affected parents is then almost always entirely healthy.

Can this happen again?

Tests of the parents' chromosomes will show if you could have another baby with a 17p duplication. When both parents have normal results, the chances of having another baby with a 17p duplication or any other chromosome disorder are generally no greater than for anyone else in the population. When one parent has a rearrangement of their own chromosomes, the chances are higher and you will be able to discuss them with a genetic counsellor or geneticist.

Prenatal diagnosis

In subsequent pregnancies, 17p duplications can be diagnosed using interphase FISH and tests on chromosomes from a chorion villus sample or amniotic fluid.

Charcot-Marie-Tooth

Having an extra copy of a gene called *PMP22* that is found in band p12 of chromosome 17 causes a muscle wasting condition called Charcot-Marie-Tooth type 1A or hereditary motor sensory neuropathy. Your paediatrician or geneticist will tell you if your child has an extra copy of this gene.

How common are 17p duplications?

Large 17p duplications are very rare indeed. Smaller duplications such as dup17p11.2 may be surprisingly common, affecting as many as 1 child in 20,000.

Families say …

L never ceases to amaze me. She seems to acquire new skills every day and is a happy, sociable little girl who is becoming increasingly independent. L also possesses an impish sense of humour and enjoys playing friendly jokes. Her courage and determination to succeed is commendable.

L was 6 when her class teacher wrote this end-of-year comment.

Inform Network Support

Rare Chromosome Disorder Support Group,
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Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

Potocki-Lupski Syndrome Outreach Foundation
http://ptlsfoundation.org
http://www.potockilupskisyndrome.org
There is a Facebook page for Potocki-Lupski Syndrome (duplication 17p11.2) at www.facebook.com/groups/ptlsawareness

When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a full leaflet about the effects of 17p duplications.

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 Lorraine Potocki M.D., Assistant Professor, Department of Molecular and Human Genetics, Baylor College of Medicine and by Unique's Chief Medical Adviser 2004.

Rare Chromosome Disorder Support Group Charity Number 1110661
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Understanding chromosome disorders

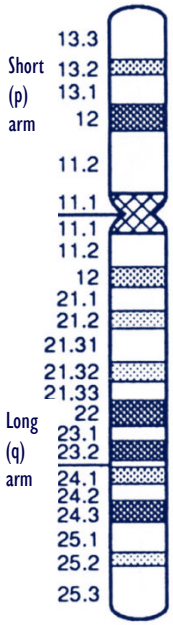
Unique

17p duplications

rarechromo.org

What are 17p duplications?

Chromosome 17



A 17p duplication is a very rare chromosome disorder where people have too much genetic material, usually in every cell in their body. The additional material is a copy of part of the short arm of chromosome 17. This condition has different names, such as **partial trisomy 17p** or **partial duplication 17p**. It is partial because only part of the short arm of chromosome 17 is duplicated, or copied. The most common duplicated segments are:

- A tiny stretch of 17p11.2. This causes dup17p11.2 syndrome.
- A much longer section covering most of the short arm of chromosome 17.

How can this affect my child's development?

The most important thing to know is what material has been duplicated. Children with tiny microduplications may be scarcely affected, particularly in early childhood. However, most babies with dup17p11.2 have difficulty feeding and putting on weight and are unusually floppy (hypotonic). Their development is also somewhat delayed. Children with larger duplications usually do face learning difficulties and developmental delay. Some have heart conditions that will be obvious from birth.

Dup 17p11.2 syndrome

Children may have no physical or learning difficulties or only slight ones. Although many understand speech, delay in talking is usual and all children should have access to speech therapy.

Babies tend to be very small at birth and put on weight very slowly with feeding difficulties that can be marked and continue into early childhood. Families may need support to prevent secondary eating behaviour problems.

Depending on the amount of extra DNA, children may have features typical of a larger duplication.

Larger duplications of 17p

Babies grow slowly during pregnancy and are born small for dates. They struggle to put on weight and tend to remain both short and slim. Typically, they have a small head but it may be in proportion to the rest of their body.

Doctors have noticed a typical look to the face that may be more obvious to parents when they see photographs of other children with a 17p duplication. The shape of the face is often triangular with a small chin, a sloping forehead and large, unusually formed ears. The eyes may slant downwards and are set wide apart. The area between the nose and the mouth (the philtrum) is unusually smooth and the baby's upper lip may be thin. You may also notice that your baby has more body hair than you would expect and extra folds of skin in the neck. A baby's fingers and perhaps toes may be clenched or bent.

Children with larger 17p duplications typically have learning difficulties that are in the moderate to severe range. It is hard to predict how your child will be affected, but a committed and attentive family and early access to learning support will give your child the best chance. Physical development is usually also mildly delayed. Your baby will be carefully checked for the medical problems that are known to be more common with a 17p duplication.

Medical concerns

- **Heart conditions**

Some babies may be born with a heart condition. This will be identified soon after birth and while some conditions resolve naturally surgery may be necessary.

- **Hypotonia**

Babies may have very floppy muscles. Physiotherapy and regular exercises improve the muscle tone and most babies achieve their milestones with only a moderate delay, walking on average at two and a half. Once mobile, children are typically very active.

- **Talipes**

Many babies have limited joint movements and some are born with the oddly angled foot positions known as club foot or talipes. These can be successfully corrected with plastic supports (splints), plaster casts or in some cases with surgery as well.

- **Hernias**

Hernias in the groin are fairly common and may need a small surgical operation. In boys with undescended testicles a surgical operation to bring one or both testicles down into the scrotum may also be needed.

- **Squint (strabismus)**

Treatment for children with strabismus (the eyes point in different directions) may involve patching, glasses and for some children an operation to realign the muscles controlling the direction of the eyeball.

- **Seizures**

These affect up to half of children with a large 17p duplication but are usually well controlled with medication.

- **Constipation**

Unique families say that this is common and often needs treatment with laxatives or stimulant medication when diet does not work.

- **Irregular teeth**

Many families report that their children have unusual or irregularly placed teeth.

***Unique* has 20-25 member families affected by a 17p duplication and many of them are happy to share their experiences with others.**