Why did this happen?

A blood test to check both parents' chromosomes is needed to find out why the 1q4 deletion occurred. In the majority of cases the 1q4 deletion occurred when both parents have normal chromosomes. The term that geneticists use for this is *de novo* (dn) which means 'new'. *De novo* 1q4 deletions are caused by a change that occurred when the parents' sperm or egg cells formed or possibly during formation and copying of the early cells after the egg and sperm joined. Some 1q4 deletions are accompanied by a gain of material from another chromosome and are often the result of a rearrangement in one parent's chromosomes. This is usually a rearrangement known as a balanced translocation in which material has swapped places between chromosomes. As no genetically important material has been lost or gained, the parent usually has no clinical or developmental problems, although they may have difficulties with fertility or childbearing. Balanced translocations involving one or more chromosomes are not rare: one person in 500 has one, making a total world population of over 13 million balanced translocation carriers. Whether the deletion is inherited or *de novo*, what is certain is that as a parent there is nothing you did to cause the 1q4 deletion and nothing you could have done would have prevented it from occurring in your baby. No environmental, dietary or lifestyle factors are known to cause these chromosome changes. No one is to blame when this occurs and nobody is at fault.

Can it happen again?

The possibility of having another pregnancy with a 1q4 deletion depends on the parents' chromosomes. If both parents have normal chromosomes when their blood cells are tested, the deletion is very unlikely to happen again. However, there is a very small theoretical possibility that the deletion occurred during the formation of the egg or sperm cells in a parent. When this occurs there is a tiny chance that parents with apparently normal chromosomes could have another affected pregnancy. On the other hand, if either parent has a chromosome rearrangement or deletion involving 1q4, the possibility is greatly increased of having other affected pregnancies. Parents should have the opportunity to meet a genetic counsellor to discuss the specific recurrence risks and options for prenatal and preimplantation genetic diagnosis (PGD). PGD requires the use of in vitro fertilisation and embryo biopsy, and only healthy embryos are transferred to the mother's uterus. If the parents choose to conceive naturally, prenatal diagnosis options include chorionic villus sampling (CVS) and amniocentesis to test the baby's chromosomes. Testing is generally very accurate, although not all of these tests are available in all parts of the world.

Families say ...

" She may sound like she has lots of health problems but she lives life to the full and brings so much happiness, joy and love to everyone that meets her " – *3 years*

" People love him because he is so adorable and loving. Because he has a limited understanding of social 'niceties' he says what he is thinking without consequence (both good and bad!) – you definitely need a sense of humour living with him! " – *9 years*

" She likes music and going out shopping and for meals. She is very people-friendly. She is always happy and smiley and easy to look after " *25 years*

Inform Network Support

Understanding chromosome disorders Unique

Rare Chromosome Disorder Support Group,
G1, The Stables, Station Rd West, Oxted, Surrey. RH8 9EE
Tel: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

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 1q4 deletions.
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. It was compiled by Unique and reviewed by Brenda Barry, Genetic Counsellor and Research Co-ordinator, Walsh Laboratory, Children's Hospital, Boston, USA and by Professor Maj Hulten BSc, PhD, MD, FRCPath, Professor of Medical Genetics, University of Warwick, UK.
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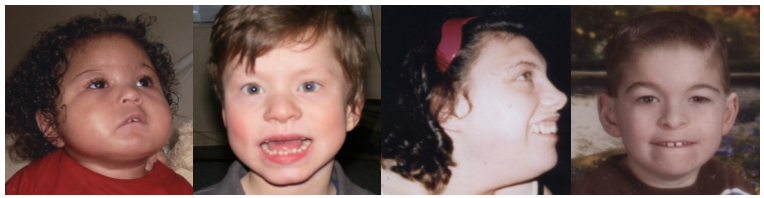
Understanding chromosome disorders Unique

1q4 deletions: from 1q42 and beyond

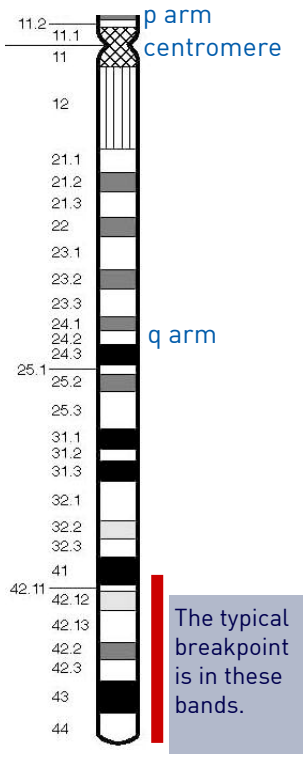
rarechromo.org



What are 1q4 deletions?

A 1q4 deletion means that the cells of the body have a small but variable amount of genetic material missing from one of their 46 chromosomes – chromosome 1. For healthy development, chromosomes should contain just the right amount of genetic material (DNA) – not too much and not too little. Like most other chromosome disorders, having parts of chromosome 1 missing may increase the risk of birth defects, developmental delay and learning difficulties. However, the problems vary and depend very much on what and how much genetic material is missing.

The end of chromosome 1 can break at different points. Breaks occur at the very end, at band 44, or further along in bands 43 or 42. Most children lose material from the end of the long (q) arm of chromosome 1, but some lose a segment of material from within it. In general when the chromosome breaks off further from the end, the clinical difficulties are more obvious. However, many features are similar, however much genetic material is missing.



The first published description of a person with a 1q4 deletion was in 1976. There have since been over 50 cases reported in the medical literature and Unique has 47 members. The deletion occurs with equal frequency in males and females.

Development

Growth

Many babies are small at birth and have feeding difficulties at first. They suck weakly or have difficulty co-ordinating sucking, swallowing and breathing and may need to be tube fed until they can take a bottle or the breast. Some babies bring their feeds back and even careful positioning, feed thickeners and prescribed antacids may not control the symptoms of reflux. If reflux is persistent, a surgical operation known as a fundoplication will create a one-way valve. Children can also be fed through a tube direct into the stomach.

Motor development

There is typically some delay in physical development. Children can usually sit between the ages of 18 months and 3 years and walk by middle childhood, but some children do not walk. However, many swim with support, ride horses with assistance and some dance, trampoline and play football.

Learning

Children need very considerable support with learning. Visual skills are usually better developed than verbal skills. Some children write their name, draw a simple picture, read a few words and become computer literate but this is not possible for all.

Speech

Some children learn to use words and short sentences, but most communicate with signs, gestures and expression. In many children receptive language seems to be better than expressive language skills - many children understand far more than they are able to express. This is shown by their ability to understand words and follow instructions and respond when told to do tasks.

Behaviour

Many children have an easy disposition. They smile around 6 months and enjoy being with people. As babies, they can be undemanding and passive although they may become more assertive as they mature. Parents say that older babies and children often have a good sense of humour and fun.

Medical conditions

Seizures

Experience so far suggests that many children will have seizures. Some children outgrow their seizures, while others need ongoing medication.

Heart conditions

Over half of babies with a 1q4 deletion are born with a heart condition. Many defects are minor and resolve naturally but a few children have complex problems that require surgery.

Brain structure

One baby in three has a structural brain anomaly, most often the absence of the band of nerve fibres (corpus callosum) that links the two hemispheres of the brain. On its own this may not affect functioning greatly but the outlook depends on any other unusual findings.

Unusual genital features

This is very common in boys. However, most problems are minor and need no treatment or can be corrected with straightforward surgery.

Cleft palate affects around one child in five

Microcephaly (an unusually small head)

is very common, however, the baby's head may appear to be in proportion to the rest of the body.

Vision

Problems occur in over half of children although they are usually relatively minor. Long sight, strabismus (squint) and visual delay are common.

Hearing

Around a quarter of children have a hearing loss although this is often transient.