What causes 1p36 deletion syndrome?

Children inherit one copy of each of the 23 chromosomes from each parent, giving 46 chromosomes in each cell. In 1p36 deletion syndrome, the end of the short arm of one of the two chromosome 1s has been lost. Breakpoints differ, so people have different sized pieces of chromosome missing. The deletion in some people is 10 times larger than in others. In general children with larger deletions with more genes missing are believed to be more severely affected, but there is no straightforward link between genes and effects. Indeed, one study found no correlation between the size of the deletion and the number of features that children had. Even children with small deletions can have most of the features of 1p36 deletion syndrome.

Why did this happen?

Both parents of a child with a 1p36 deletion should have their own chromosomes tested, although 90 - 95 per cent will have normal chromosomes. The chromosome break is then said to have occurred out of the blue (*de novo*). In the remaining families, one parent usually has a balanced rearrangement of their own chromosomes which has become unbalanced when eggs or sperm were created. In both situations, there is nothing you can do to stop this, just as there are no environmental, dietary or lifestyle factors known to cause it. So there is nothing that either parent did before or during pregnancy that caused the deletion to occur and equally nothing could have been done to prevent it.

Can it happen again?

Where both parents have normal chromosomes, it is unlikely that another child will be born with a 1p36 deletion or any other chromosome disorder. Where a parent has a rearrangement of their chromosomes with a break at 1p36, the risk of having another affected child is higher.

Families say ...

“ Our son continues to prove the doctors wrong. We are humbled by his sheer determination and guts ” - age 2.

“ Ferrin giggles at the slightest thing and gets very excited when she sees you. She has become very loving lately ” - age 5.

“ Laura is delightful - cheerful, happy and very amusing. She has the ability to change the mood in the house very quickly ” - age 12.

Inform Network Support

Understanding chromosome disorders Unique

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1p36 deletion syndrome

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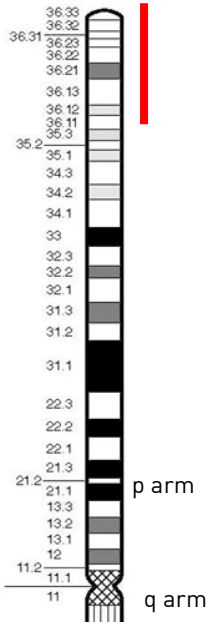
What is 1p36 deletion syndrome?

1p36 deletion syndrome is a chromosome disorder. A chromosome disorder is a change in chromosome number or structure which results in a set of features or symptoms. People with 1p36 deletion syndrome have lost a small but variable amount of genetic material from one of their two chromosome 1s.

Chromosome 1 is the largest chromosome and represents about eight per cent of the total DNA in cells. For healthy development, chromosomes should contain just the right amount of material – not too much and not too little. People with 1p36 deletion syndrome have one intact chromosome 1, but the other is missing a tiny piece which affects their learning and physical development in relatively predictable ways. Most of the clinical difficulties are probably caused by the presence of only one copy (instead of the usual two) of a number of genes. However, a child's other genes and personality also help to determine future development, needs and achievements.



Chromosome 1

Most people have, as babies:

- Delay in development
- Feeding difficulties
- Muscles that feel floppy (hypotonia)
- Very large fontanelle (soft spot)
- Eyesight and hearing problems
- Heart conditions, usually mild.

Medical Concerns

Feeding At least two babies out of three need feeding support as they suck too weakly or have difficulty co-ordinating sucking, swallowing and breathing. Many babies also have difficulty keeping their feeds down. This condition is called gastro-oesophageal reflux - GO or GE reflux. In young babies, giving small feeds, adding prescribed thickeners to formula milk and careful positioning may be all that's needed. Babies with severe reflux can have surgery to make a valve that allows food into the stomach but stops the stomach contents returning up the food pipe. Another approach is to insert a feeding tube (G-tube, gastrostomy) direct into the stomach.

Seizures Between half and three quarters of children have seizures. Some children outgrow them or the seizures become less frequent, but many need to take anti-epileptic medicines.

Heart conditions More than 40 per cent of children are born with a heart condition. However, this is often minor and heals naturally. A small number of children need surgery. One child in three has a condition where the activity of the heart muscle is abnormal, called dilated cardiomyopathy. In some children this too improves with time.

Eyesight problems affect four children in five. Most commonly, children are long-sighted. Other problems (such as strabismus/squints) are also treatable but a sizeable number of children have long -lasting problems which mean that they are registered partially sighted or blind.

Hearing Most children have at least a mild sensori-neural (permanent) hearing loss and glue ear (usually temporary) may increase their difficulties.

Infections Vulnerability to infection has not been formally reported as a feature of 1p36 deletion syndrome, but in Unique's experience it affects 2 in 3.

Thyroid One child in five has a low thyroid level. As this can be corrected, thyroid function should be checked at birth, at 6 months and then every year.

Constipation affects more than half of children and occasionally is severe.

Development

- Physical development

Children are slow to sit and walk. Hypotonia (floppiness) makes it hard for them to stay upright and as babies they prefer to move by rolling or wriggling. Typically children sit between the ages of 2 and 3 and walk between three and eight, but some children never walk. All children benefit from physiotherapy and swimming is a popular and valuable form of exercise.

- Learning

Children need very considerable support with their learning. A few learn to use some words and understand clear spoken directions, but most rely on a signing system. Some children do not communicate at this level. Controlling seizures is believed to strongly influence children's ability.

- Behaviour

Children can be a delight.

They are often finely emotionally attuned and show and receive affection. The Unique experience suggests that they are sensitive to music. As they mature, children may become more demanding.

- Growth

Some babies are born very tiny and remain short. However, a good number catch up and grow to average height. Some children even put on too much weight from middle childhood.