Why did this happen?
Four times out of five, a 2q37 deletion occurs as a new event, not caused by any abnormality in the parents’ chromosomes. This is part of a natural process and as a parent there is nothing you can do to control it. Children from all parts of the world and from all types of background have 2q37 deletions. No environmental, dietary or lifestyle factors are known to cause them. So nothing you did before you were pregnant or during pregnancy caused this to occur and there is nothing you could have done to prevent it.

Can it happen again?
The chances of having another child affected by a 2q37 deletion depend on the results of chromosome tests on the parents. If the parents’ chromosomes are normal, your chances of having another affected child are generally no higher than for anyone else in the population. In a minority of families with a child with a 2q37 deletion, one parent has an unusual chromosome rearrangement (called a balanced translocation). This does not generally affect their own health or development but can cause problems when they create eggs or sperm. Because of this possibility, both parents should have their chromosomes tested and have a personal interview to discuss the findings with a geneticist or genetic counsellor.

How is a 2q37 deletion diagnosed?
A blood sample is taken. Cells from this blood sample are treated so that the chromosomes can be examined under a high resolution microscope. The missing portion of chromosome 2 can usually be seen but sometimes additional molecular tests such as FISH tests are needed. These check for the presence of specific lengths of chromosome material normally found at or very near the end of chromosome 2.

Families say …
… Pure joy. She doesn’t miss a beat and loves life - age 5, 2q37.3.
… A very loving child who also has a very caring side to her. Good sense of humour - age 8, 2q37.3.
… Full of fun with a wicked giggle - age 19, 2q37.1.
… Ann is Ann. What you see is what you get - age 20, 2q37.2.

Inform Network Support

Unique
Understanding chromosome disorders

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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 2q37 deletions. 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 Dr Micheala Aldred, Division of Medical Genetics, University of Leicester, UK and by Unique’s Chief Medical Adviser 2004
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What is a 2q37 deletion?

2q37 deletion syndrome is a very rare chromosome condition where people have lost a small amount of genetic material from the end of one of their chromosomes. As well as two sex chromosomes, everyone has 22 pairs of chromosomes in the cells in their body. People with a 2q37 deletion have lost the end of one of their chromosome 2s and the missing material tends to affect similar areas of development. All the same, people with 2q37 deletions vary hugely – much more than people with other chromosome conditions. This means that the full impact of a 2q37 deletion on development, needs and achievements can be hard to predict at first.

Not every child will have all or even most of them but most will have some.

- **Developmental and learning delay**
- **Baby hypotonia (floppiness)**
- **Short stature**
- **Short hands and feet, particularly the 4th (ring) finger and sometimes the 3rd (middle) and 5th (little) fingers**
- **Possibly increased vulnerability to emotional, social & behavioural stress**
- **Tendency to put on weight**
- **Unusual behaviour, including autistic features.**

How common are 2q37 deletions?

This is not known. Around 60 people have been described in research reports. Unique has around 40 member families with this chromosome condition and can put families in touch with each other.

Medical conditions

Most children do not have serious medical issues. However, the following features are fairly common.

- **Feeding problems** Weak sucking means that breast and bottle feeds are slow. Many babies bring milk back and need to be carefully positioned to lessen the risk of inhaling milk. Some babies need antacid medicines to soothe the inflammation inside the food pipe. Once on bottles, babies may need thickeners to help keep liquids down. A few babies need surgery to control reflux.
- **Lax joints** Some babies are born with dislocated hips or hip dysplasia that is corrected by splinting and occasionally surgery. Some children have highly flexible fingers.
- **Kidneys and urinary tract** A small number of children have a kidney that is unusually shaped or situated. However, this usually causes no problems. It tends to be discovered while screening children for a rare type of kidney cancer called Wilms’ tumour. Although Wilms’ tumours are not common in babies with a 2q37 deletion, they have been known to occur, so children are usually offered regular ultrasound checks in early childhood.
- **Hernias** at the navel (umbilical) or in the groin (inguinal) can occur.
- **Heart conditions** are a little more common than in other children but may resolve without surgery. Most often there is a hole between two chambers of the heart that takes longer to close than expected and needs monitoring. Occasionally the problem is more complex and surgery is needed.
- **Eczema** may affect young babies and is frequently linked to a cow’s milk allergy. Children usually outgrow this but then may develop asthma.
- **Digestive problems** can occur. Occasionally babies develop a blockage in the intestine and need surgery.