How many people have a 1q23.3 microdeletion?

A 1q23.3 microdeletion is very rare. Very few patients are reported in the scientific literature. However, with increasing use of 'next generation' genetic diagnostic technology, we will identify more people affected by this condition. This will help provide more useful information to families.

Why did this happen?

There is no known cause for 1q23.3 deletions. Nothing you did or did not do has caused this.

Can it happen again?

Provided that neither parent carries the same genetic change as their child, the chance of having another affected child is extremely low – less than one percent. The residual risk is due to a phenomenon called 'gonadal mosaicism', where a genetic change is present in the egg or sperm but not in any other cells of the body. None of the children reported with 1q23.3 microdeletions inherited it from a parent. Specific advice should be sought from your clinical genetics team, such as a genetic counsellor.

Can this be cured?

There is no cure as the effects of the deletion took place during development of the baby in the womb. However, knowing the diagnosis means that appropriate monitoring and treatment can be put in place.

Inform Network Support

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

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Understanding Chromosome & Gene Disorders

1q23.3 microdeletion

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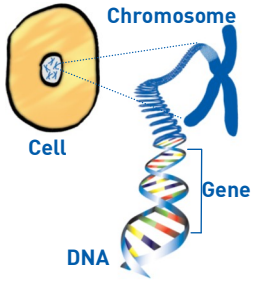
What is a 1q23.3 microdeletion?

A 1q23.3 microdeletion is a genetic condition where a small segment of chromosome 1 is lost. There are 46 chromosomes in the body in 23 pairs. One of each pair comes from the mother's egg, the other from the father's sperm. Each chromosome has a long arm (q) and a short arm (p). A 1q23.3 microdeletion occurs on the long arm of chromosome 1 at band 23.3 (see diagram).



Genes are made of DNA and packaged into chromosomes, found in almost every cell of our body. They are the instructions telling our bodies how to develop and function. A deletion in a chromosome can disrupt many surrounding genes. Although there are only a few reported cases of a microdeletion at 1q23.3, it is thought that the main associated gene is *PBX1*. This gene is thought to play an important role in the development of the kidney, urinary tract and brain in babies. The *LMX1A* gene is also part of the deleted region of the chromosome and is thought to play an important role in brain development.

- There are no specific features that enable a precise diagnosis of a 1q23.3 deletion to be made.
- Diagnosis must be made with a specific genetic test.

Medical concerns

Alterations to kidney and/or urinary tract structures

Many children with 1q23.3 deletions have alterations to the structure of their kidneys or urinary tract. These include both kidneys being small or one kidney being absent. In some people there are alterations to the tube leading from the kidney to the bladder. Undescended testes (cryptochidism) are common in boys with the deletion.

Alterations to ear structure

In many children with the deletion the external ear has been of altered appearance. Some children may have impaired hearing.

Congenital Heart Disease

About half of children with this deletion will have an alteration to the structure of their heart. This can be relatively minor such as an atrial septal defect (ASD) that closes by itself or more complicated such as a ventricular septal defect (VSD), which may require surgery.

Development

Physical development

Children with 1q23.3 microdeletions showed signs of developmental delay and some showed general motor difficulties. Most children were able to walk at around 18-24 months but all people with this deletion so far described have been able to walk independently.

Learning

All reported children with the condition had significant difficulties with learning and showed a below average IQ for their age. Most attended a special education needs school or needed extra help while in a mainstream school.

Speech

Children typically experience speech delay and difficulties to a varying degree.

Using their hands

Some children may experience difficulties with holding and using everyday items such as cutlery.

Growth

Children with 1q23.3 microdeletions tend to be smaller than average and have a smaller than average head circumference.

Behaviour

Some children with this condition have been identified as showing autistic or challenging behaviour.

Management:

- A scan to examine the structure of the kidneys/ urinary tract (ultrasound or MRI).
- A scan of the heart (echocardiogram).
- Physical examination for undescended testes.
- Follow up by a developmental paediatrician or community paediatrician.
- Physiotherapy and speech and language therapy as needed
- Hearing assessment, perhaps with a special scan (MRI) to examine the development of the inner ear structures for hearing and balance.
- Genetic counselling as needed to provide support and advice about the genetic condition.