Why did this happen?

Most 22q13 deletions arise out of the blue from an event that occurred when the parents' sperm and egg cells were forming. 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 22q13 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 and you could have done nothing to prevent it.

Will we have another baby with a 22q13 deletion?

The chances of having another affected baby depend on the results of chromosome tests on the parents. If the tests show that your chromosomes are normal, your chances of having another affected baby are no higher than for anyone else in the population. No family has yet been found to have two children with a 22q13 deletion when the parents have normal chromosomes.

In around three families out of 20, the chromosome tests show that one parent has a balanced translocation of their own chromosomes. This means that fragments of the parent's chromosomes have swapped places but because no chromosome material has been lost or gained, the vast majority of people are themselves unaffected. However, for these parents the chances of having another affected child are much higher.

As each family is unique, you should have a personal interview to discuss the findings of your chromosome study with a geneticist or genetic counsellor.

Prenatal diagnosis

A chromosome study of cells taken from an amniocentesis can reveal a 22q13 deletion. However, the deletion is often difficult to find and if it is suspected, an additional FISH test should be performed.

Inform Network Support

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

Phelan-McDermid Syndrome and 22q13 Deletion Foundation
Greenwood Genetic Center, 6 Doctors Drive, Greenville, SC 29605, USA
www.22q13.org

Unique mentions other organisations' message boards and websites to help families looking for information. This does not imply that we endorse their content or have any responsibility for it.

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 Dr Katy Phelan, Greenwood Genetic Center, USA and by Professor Maj Hulten BSc, PhD, MD, FRCPath, Professor of Medical Genetics, University of Warwick, UK 2008.

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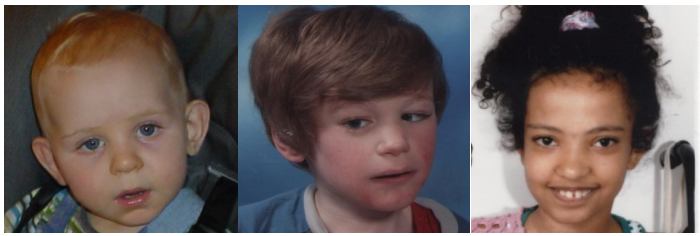
Phelan-McDermid syndrome (22q13 deletions)



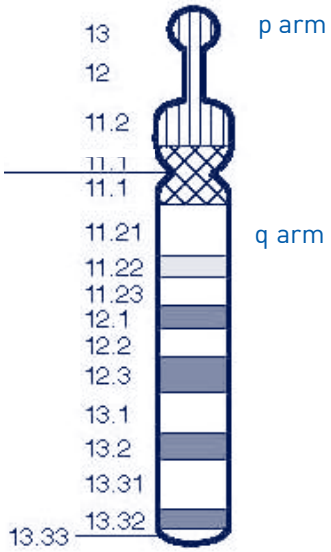
rarechromo.org

What are 22q13 deletions?

People with 22q13 deletion syndrome (also called Phelan-McDermid syndrome) have lost genes from the end of the long (q) arm of one of their two chromosome 22s.
22q13 deletions are rare, but no one knows yet exactly how often they occur because they can be very difficult to detect. At present, more than 200 people are known to have a 22q13 deletion, but the total is certainly much higher.

The sex ratio is equal – as many boys as girls are affected.

Almost everyone with a 22q13 deletion shows the features described below.



Chromosomes contain the genes that instruct the body to develop and work properly. The typical breakpoint is at 22q13.31 in the long (q) arm

- Delay in reaching baby 'milestones'
- Muscles that feel 'floppy' in babies
- Learning difficulties or disability
- Delay in starting to speak or no speech
- Normal or rapid growth
- Unusual look to the face. This is usually subtle, more obvious to a doctor than to a parent.

Apart from normal growth, these signs are common in other chromosome disorders. Unusually formed toenails and a tendency not to sweat normally are more distinctive features. However, children vary and your child's whole genetic make-up and environment also matter.

Development

Children vary widely. The broad range seen in the age at which children become mobile is mirrored in learning, social and speech skills.

Motor development

Babies reach their milestones late, crawling on average at 16 months (range 7 months to 36 months) and walking at 33 months (range 13 months to 8 years). Since relative weakness in upper body muscles and poor balance contribute to this delay, physiotherapy and specific exercises to strengthen the upper body are helpful.

Learning

Most children have considerable developmental delay and benefit from early intervention strategies, intensive occupational therapy and any approach to increase their concentration, lengthen their attention span and improve their communication skills.

Behaviour

Children typically show 'autistic-like' behaviour and are unusually tolerant to pain. Most behave well but around a quarter can be aggressive when they feel frustrated. Behaviour modification programmes that use positive reinforcement have been successful for some children. Children who become hyperactive can be helped with prescribed medicines.

Speech

Many babies babble at the appropriate age and may learn a few words. Early in childhood, at about 4 years, many children seem to lose their verbal skills. Active speech therapy and communication training help but verbal communication remains impaired. Children often understand more than they can express. Some can understand spoken words and follow simple instructions, although this is not true for all. Sign language, computer touch screens, voice based systems and picture exchange systems all improve communication skills.

Feeding Issues

Newborn babies typically have some difficulty with sucking and swallowing but most learn to drink milk successfully. About 30 per cent of babies have difficulty keeping their feeds down (gastro oesophageal reflux - GO or GE reflux). In young babies giving small feeds, adding thickener to formula milk and careful upright positioning may be all that's needed. Reflux may be more difficult to spot in older babies and young children because of their increased tolerance of pain and discomfort. Eating smaller meals, avoiding foods that cause irritation, raising the head of your child's cot or bed, sleeping on the left side and eating no food within two to three hours of bedtime all help.

Medical concerns

Children with 22q13 deletions are usually healthy. A small number of children have developed kidney problems, and arachnoid cysts are 10 times as common as in other children. These can be linked to uncontrolled crying, vomiting and severe headaches but some children have no symptoms from them. Older children may be prone to develop swollen legs or ankles. Most people also overheat easily.