Why did this happen?

There is no known cause for the alterations to the *MYT1L* gene that cause this syndrome. There is nothing that you, as parents, did or did not do which caused the condition.

Can it happen again?

In all of the people with *MYT1L* gene alterations or 2p25.3 deletions so far reported, the genetic change happened for the first time in them and was not inherited from a parent (the change was *de novo* (dn)).

This means that the chance of having another child affected by the same condition is very low (<1%). This very low chance of having a second affected child is due to something called "gonadal mosaicism". This means that the gene alteration or 2p25.3 deletion would only be found in the sperm or egg cells, but not the blood, of the parent. This could theoretically result in a parent who had a normal blood test for the *MYT1L* gene alteration or 2p25.3 deletion having a second affected child.

Management:

- Children with MYT1L syndrome should be under the care of a developmental paediatrician or community paediatrician so that developmental milestones can be monitored.
- Speech therapy or physiotherapy, as appropriate, may be required depending on developmental progress.
- An awareness of the potential for overeating and excessive weight gain, and strategies to manage these.
- An EEG (tracing of electrical activity in the brain) may be required if epilepsy is suspected.
- Tests for eye problems such as squint should be performed by paediatric eye doctors.

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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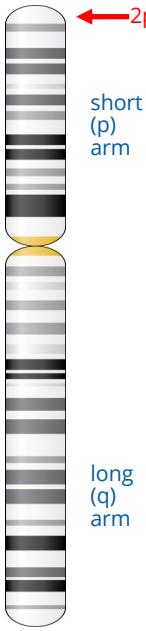
Understanding chromosome disorders
Unique

MYT1L Syndrome/2p25.3 deletions

rarechromo.org

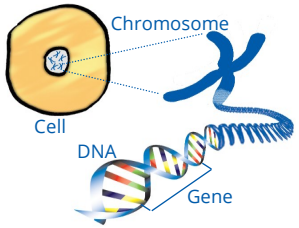
What is MYT1L Syndrome?

MYT1L syndrome is a very rare medical condition due to alteration of the function of a gene called *MYT1L*. Genes are instructions, which have vital roles in our growth and development. They are made of DNA and are incorporated along with many other genes into organised structures called chromosomes. The *MYT1L* gene is on chromosome 2. The *MYT1L* gene gives an important instruction for brain development. MYT1L syndrome can occur because of complete deletion of one of the two copies of the *MYT1L* gene which we all carry. This is known as chromosome 2p25.3 deletion. MYT1L syndrome can also occur because of mutations (changes to the code) of the *MYT1L* gene, which stop the gene from working. People with a deletion or mutation of the *MYT1L* gene have very similar symptoms; all have learning problems and the majority also are overweight.



What does the MYT1L gene do?

The *MYT1L* gene gives an important instruction for brain development. It plays a crucial role in turning on and off other genes during the development and growth of the brain, before the baby is born. The *MYT1L* gene also seems to play a vital role in the development of the chemical pathways in the brain which control appetite and weight gain.



Most Common Features:

- Developmental delay
- Intellectual disability, often including a speech delay
- Excessive weight gain

Other possible features:

- Behavioural concerns, including autistic traits and challenging behaviours
- Minor skeletal abnormalities

People with mutations (changes) to the *MYT1*L gene and deletions at 2p25.3 (the part of the chromosome where the *MYT1L* gene is found) have very similar symptoms.

Medical concerns

Excessive weight gain

The majority of people with mutations of *MYT1L* or deletions at 2p25.3 have problems with over eating and excessive weight gain.

Minor skeletal abnormalies

Minor skeletal anomalies, such as inward curving of the 5th (little) finger, have been reported.

Seizures/Epilepsy

Some children experience epilepsy, although this is not a consistent feature.

Other concerns

One person with a mutation of the *MYT1L* gene had spina bifida and was unable to walk. It is unclear if this is a specific feature of the condition or a coincidence.

Families say ...

“ As her only parent I have at times reached my limits, but her spark and my own sense of humour has helped a lot! It has been hard to experience all that my daughter has had to deal with with her. ”

Development

Physical development

Most of those so far identified as having MYT1L syndrome have learned to walk, on average around two years of age. Two people with MYT1L syndrome have never walked (one adult who had spina bifida and one teenager).

Learning

All reported children and adults with MYT1L syndrome have had significant difficulties with learning and showed a below average IQ for their age. As a result they have attended a special educational needs school or have extra help at school.

Speech

Children typically experience speech and language delay to a varying degree, with the level of attainment less than that of the general population.

Growth

Most people affected by MYT1L syndrome have a normal head size but have a heavier than average weight.

Behaviour

Some people with this condition display autistic traits, attention hyperactivity disorder (ADHD) or challenging behaviour.

Families say ...

“ My daughter is a very complex person with very complex needs and she faces challenges every moment of her day. Her autism and challenging behaviour are the hardest challenges for her. Thankfully she has personality and a sense of humour. ”