**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.

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**Inform Network Support**

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**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.
**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 MYT1L 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 abnormalities**
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 her.”*