

Families say ...

" Where do we start? He is our son and the fact that he has some disabilities only makes us feel more protective and loving towards him. He leaps out of bed every morning unlike most teens. Apart from the odd blip in frustration he has a happy disposition and positive attitude. He loves being part of things, doing jobs and feeling a valued member of the proceedings. He has a great memory and is humble when using it, though pleased to be able to help. He is funny and often laughs in the face of adversity. He has a fun sense of humour, but if given an inch can take a mile, so this can cause difficulties with other children.

" As with any parent we share in his achievements and because some of these are such hard work for him we feel great joy with every step he takes. He has recently started a voluntary job helping at a local café and it is great to see him cycling off to work, so proud to be independent. Although some people can struggle to cope with disability, others have provided him and us with fantastic support and belief in him and his potential." - 16 years



Families say ...

" My son has a very good memory and is good at doing puzzles. I make him feel useful and included in my household chores: he throws out rubbish for me, and can Hoover when he is in a good mood. He isn't toilet trained and his fine motor skills are not good, so he needs help with all personal care. " But he is a natural charmer and likes to play tickles with his brothers and sister. Physically he has abnormal dentition, but it doesn't bother him. He looks very handsome. He is very, very special to me, and his big smile on his face every morning makes my day." - 10 years



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This guide 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. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. This guide was written by Dr Usha Kini, Consultant Clinical Geneticist, Oxford University Hospitals, UK.

2015 Version 1 (PM)

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SATB2 syndrome

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What is SATB2 syndrome?

SATB2 is a gene on chromosome 2q33.1, which was first found to be linked with cleft palate in 1999. Affected people were also noted to have learning difficulties, facial features such as a prominent nasal bridge and a small mouth and long, slender fingers. Further studies have shown that losing *SATB2* is a relatively common cause of learning difficulties with or without a cleft palate. Many of the features of the children with SATB2 syndrome are similar to those in people with a chromosome 2q33.1 microdeletion which includes the *SATB2* gene.

Can it be cured?

There is no cure for this disorder as the effects of the genetic change took place during your baby's formation and development. However, knowing this diagnosis means that appropriate monitoring and treatment can be put in place for your child.

What causes SATB2 syndrome?

SATB2 is a gene which is important for the development of the face and the brain. A disruption or the absence of this gene results in abnormalities in the structure of the face (such as cleft palate, teeth abnormalities, prominent nasal bridge) and the function of the brain (such as developmental delay, learning difficulties and behaviour problems).

Why did this happen?

When children are conceived your genetic material is copied in the egg and sperm that makes a new child. The biological copying method is not perfect and occasionally random rare changes occur in the genetic code of children that are not seen in the DNA of their parents. This happens naturally in plants and animals and is not due to your lifestyle or anything you did to cause a change in the *SATB2* gene. In most families the sequence change in *SATB2* occurs out of the blue (de novo). In a few families, one parent may have the same genetic change as their child but this is very rare.

Can it happen again?

The risk of having another child affected by a rare gene disorder depends on the genetic code of the parents. For SATB2 syndrome where parents do not carry the mutation the chance of having another child is very low. If the genetic analysis of the parents of a child with SATB2 syndrome shows they carry the same variant, the chance of it happening again is much higher. Each family situation is different and a clinical geneticist can give you specific advice for your family.

Most people have:

- Intellectual disability or slow learning or developmental delay
- Absent or severely delayed speech
- Cleft palate (a split in the roof of the mouth)
- Dental crowding or abnormality of teeth
- Prominent bridge and bulbous tip to the nose and a small chin
- Behaviour problems

Medical concerns

■ Cleft palate

In about half the children with SATB2 syndrome, a cleft palate is present. This cannot be identified on the scans that the mother has during pregnancy. It may however be found when your baby has a health check after birth. One sign can be regurgitation of milk from the nose. Your baby will need help with feeding and in due course surgery to close the palate.

■ Teeth abnormalities

These may be in the form of dental overcrowding, abnormal shape of the teeth or teeth being late coming through. It is therefore important that your child is under the care of a dentist.

■ Seizures

These are uncommon and are seen in a very small proportion of children and may sometimes occur with a temperature.

■ Healthy brain, heart and kidneys

No structural defects of the heart, kidneys or brain are seen.

Management

Community paediatricians should oversee care so that development and behaviour can be monitored and the best help in the form of physiotherapy, occupational, speech and behavioural therapies (including medications) can be given early if it is needed.

Children with SATB2 syndrome who have a cleft palate should be under the care of the multidisciplinary cleft team. Expert advice regarding feeding with a cleft is available from the cleft nurse specialists. The best time for the palate to be closed will be decided with the family by the cleft surgeon. Dental, audiology [hearing] and psychology assessment and support is also offered through the cleft team.

Development

■ Growth

Almost all babies are average in weight at birth and continue to grow along their centile lines. While some children grow well and are of average height, many children are short for their family. Some children may also have a relatively small head size.

■ Feeding

Some babies may have initial difficulty with feeding due to their cleft palate and low muscle tone. The low tone resolves with time and most children gain weight and grow well. Dribbling can be a problem for many children but this can be helped by medicines to reduce the saliva.

■ Sitting, moving and walking

Most babies will have low muscle tone and can appear floppy to begin with. Their joints may also appear excessively flexible. They are usually late in becoming mobile but most children start walking independently by the age of 2 years, although some can take much longer. Physiotherapy is useful.

■ Speech

Some children learn a few single words, but most do not develop speech. Instead they communicate by other means, such as sign language, gestures and vocal noises.

■ Learning difficulties

Most children need considerable support with their learning and may need to attend a special school where the right support can be given and non-academic and daily living skills focused on. Supervision may be needed even for adults.

■ Behaviour

On the whole, children usually have a happy disposition and are seen to be very sociable and even over-friendly. Autistic traits have only been noted infrequently. Hyperactivity may be seen, but aggressive behaviour is rare. Sleep disturbances have been frequently noticed by parents and can be severe enough to need medication.

