

Supporting people with craniosynostosis

Craniosynostosis is a rare condition where a baby's skull does not grow properly and their head becomes an unusual shape. Having craniosynostosis can affect breathing, hearing, speech, language development and feeding. Working as part of a multidisciplinary team, speech and language therapists play an important role supporting children and young people with craniosynostosis and their families.



What is craniosynostosis?

A baby's skull has growing lines between the skull bones filled with flexible material called sutures. These sutures allow the skull to grow as the baby's brain grows. Craniosynostosis is when two or more of the bones in a baby's skull join together prematurely, before the baby's brain is fully grown.

This causes the baby's skull growth to be restricted perpendicular to the fused suture. In the other parts of the skull where the sutures remain open, the baby's skull will continue to grow. When that happens, the skull will develop an abnormal shape, although the brain inside has grown to its usual size. In some instances, the brain might not have enough room to grow to its usual size. This can lead to a build-up of pressure inside the skull in a small number of cases. Surgery is offered to expand and reshape the skull, both to allow normal brain development, and for aesthetic and psychosocial reasons. Craniosynostosis is estimated to affect between 1 in every 1,400–2,100 live births.^{1,2} Craniosynostosis is classified by NHS England as a rare disease.

Types of craniosynostosis

NON-SYNDROMIC CRANIOSYNOSTOSIS

Non-syndromic craniosynostosis is sometimes referred to as 'single suture' craniosynostosis, and accounts for about 65% of all cases of craniosynostosis.³ It usually involves one suture and is not normally accompanied by problems affecting other parts of the skull, face or body.

SYNDROMIC CRANIOSYNOSTOSIS

Craniosynostosis may also be a feature of a syndrome caused by a genetic change. We are still learning about new syndromes linked with craniosynostosis, and new genetic changes continue to be found.³ Some of the more common kinds of syndromic craniosynostosis are in the figure below.

Figure 1. Syndromic Craniosynostosis

Apert syndrome

Saethre-Chotzen syndrome

Pfeiffer syndrome

Craniofrontonasal syndrome

Crouzon syndrome

Muenke syndrome

TCF12-associated

ERF-associated

SMAD6-associated



Management of craniosynostosis

The primary treatment of craniosynostosis is surgical, but the management is multi-disciplinary. Specialist craniofacial speech and language therapists (SLTs) work together as part of a team with plastic and reconstructive surgeons, maxillofacial surgeons, neurosurgeons, audiologists, ENT surgeons, clinical psychologists, orthoptists, ophthalmologists, orthodontists, specialist nurses, geneticists and many others to provide specialist care for people with craniosynostosis. Specialist craniofacial units treat patients from birth to maturity.

The impact of craniosynostosis on communication and feeding

People with craniosynostosis fall into the category of low incidence, high need for speech and language therapy support.

HEARING: Most children with non-syndromic craniosynostosis are not at increased risk of hearing difficulties.⁴ In children with syndromic craniosynostosis, hearing may be affected due to changes in the bones for hearing, structural differences in the ear and face, or because the genetic change also affects the sensorineural component of hearing.^{5,6,7,8,9} Glue ear is common in all types of syndromic craniosynostosis.^{10,11}

SPEECH: In syndromic craniosynostosis, narrow airways in the nose and throat can obstruct the movement of air resulting in speech which sounds like the speaker has a cold (hyponasal resonance). Cleft palate is a feature of some syndromes (for example, Apert syndrome), and may have implications for speech sound production.⁹ Cleft palates are usually repaired in infancy. Even after repair of a cleft palate, some children might have difficulty closing off the nasal cavity when talking (hypernasal resonance). Structural differences such as dental crowding, a high narrow hard palate, and a small upper jaw are linked with syndromic craniosynostosis and can cause speech difficulties such as dental articulation (tongue further forward in the mouth), or backing (tongue pushed to the back of the mouth).^{8,12}

LANGUAGE DEVELOPMENT: Most children with craniosynostosis have normal language development.¹³ However, there is an elevated risk of children having language disorder associated with syndromic and non-syndromic craniosynostosis.^{5,8,13} There is a wide variability in the severity of language difficulties associated with craniosynostosis. Some children have difficulty with social communication skills.

ACADEMIC ATTAINMENT: Most children attend mainstream school, and do not require educational support. Some children with syndromic craniosynostosis require additional educational support.^{8,9}

FEEDING: Most children with craniosynostosis are able to feed normally. Children with syndromic craniosynostosis may have feeding difficulties associated with cleft palate, oral structural differences, cardiac difficulties, upper airway, and respiratory difficulties.^{9,14}

David's story

When David was first born, his parents and doctors noticed that he had a different head shape, and his fingers and toes were webbed and joined. He also had difficulty breathing and feeding. The doctors referred him to the NHS designated craniofacial unit when he was 1 day old where the team identified that he had clinical features of Apert syndrome.

He was later referred for testing by a clinical geneticist, which confirmed this diagnosis. He was referred to the craniofacial specialist SLT because he had some early problems with breathing related to his small mid-face, which were impacting his feeding. These difficulties were so significant that he was given a feeding tube. He had a tracheostomy tube inserted when he was four months old to help with his breathing.

The specialist craniofacial SLT continued to be involved with his early feeding and supporting his communication development because of his tracheostomy. The craniofacial SLT noticed that David was not responding to sounds, and referred him to the audiology team. Testing revealed that he had a hearing loss and he was given hearing aids.

As David grew, he needed help from an SLT to learn to eat food with lumps, drink from a cup and learn to use a speaking valve on his tracheostomy. The craniofacial SLT also worked with the community SLT to encourage early signing to help him communicate. David had several operations to expand and reshape his skull so his brain could grow (when he was a few months old, and again when he was 4 years old). He also had an operation to bring the bones of his nose, top jaw and cheeks forward to help with his breathing and eating when he was 6 years old.

The craniofacial SLT was involved in regular monitoring of his communication, liaising with local SLT services, and conducting an assessment of his palate function before and after his mid-face was moved forward to make sure he did not develop velopharyngeal dysfunction (disruption in the ability to close off the nasal cavity from the oral cavity). The craniofacial SLT was also involved in advising local SLTs about using an Augmentative and Alternative Communication (AAC) device to help with his communication. David received local speech and language therapy via school and he had an education, health and care plan.

As he continued to grow, David's parents felt communication was his biggest need and decided to move him to a special educational needs school to access local speech and language therapy. By age 6, he could manage a normal diet. David is now a teenager, and he attends a special school and uses a communication device.

At home his family can understand most of what he says. David continues to wear hearing aids and has a radio microphone to help him hear in the classroom. His speech, language and feeding will continue to be monitored by the speech and language therapists at the craniofacial unit until he is fully grown.



The role of specialist craniofacial speech and language therapy

- Assess the speech, language and feeding of children, young people and adults with craniosynostosis.
- Ensure that children and young people are under the care of local speech and language therapy services so that intervention is provided close to home.
- Provide specialist advice about speech, language and feeding difficulties related to craniosynostosis and craniofacial conditions to families and local SLTs.
- Provide specialist early intervention of feeding difficulties associated with craniosynostosis.
- Attend multidisciplinary clinics, contribute to audit and research, national benchmarking, collaborate with colleagues in other centres, present research at national and international meetings.

The impact of speech and language therapy

- Children with syndromic craniosynostosis have been shown to require long-term support from speech and language therapists.^{5,8,9,13}
- Early speech and language difficulties can improve with supportive interventions in some cases.¹⁵
- Early intervention with feeding difficulties associated with craniosynostosis can maximise safety and support positive feeding experiences for parents and children.

Knowing when to contact a specialist craniofacial speech and language therapist

All children receiving care from the four designated supra-regional Craniofacial Units will be under the care of speech and language therapists and will be seen at regular intervals during childhood. Day-to-day speech and language therapy support is undertaken by local speech and language therapists. You may wish to contact a craniofacial speech and language therapist if:

- There is a significant change to the child's attention and concentration.
- They are not making progress with therapy for a speech sound disorder and/or are not stimulable for target sounds.
- They start coughing or choking when eating or drinking and have changes to their swallowing or voice.
- They are having difficulties chewing food.
- A second opinion or advice is wanted.

The treatment of craniofacial conditions is commissioned by Highly Specialised Services from NHS England and carried out at four designated supra-regional craniofacial units. These are based at the following hospitals:

- **Alder Hey Children's Hospital** – Liverpool, Alder Hey Children's NHS Foundation Trust
- **Birmingham Children's Hospital** – Birmingham, Birmingham Children's Hospital NHS Foundation Trust
- **Great Ormond Street Hospital** – London, Great Ormond Street Hospital for Children NHS Foundation Trust
- **The John Radcliffe Hospital** – Oxford, Oxford University Hospitals NHS Foundation Trust



Children from Wales, Northern Ireland, and the Isle of Man are seen regularly by the English designated teams. If the child is accepted for treatment by the team then they are offered the same service from speech and language therapy. In Scotland, there is a Designated Specialised Craniofacial Unit at Glasgow Children's Hospital.

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- For more information about how speech and language therapists can support people with craniosynostosis please email info@rcslt.org
- Headlines is a national charity which supports people with craniosynostosis and rare craniofacial conditions. For more information about the support that Headlines can provide, please visit www.headlines.org.uk or email info@headlines.org.uk

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