SAGITTAL SYNOSTOSIS

Otherwise known as?

Sagittal synostosis is also known as scaphocephaly or dolichocephaly. These terms are used interchangeably to describe the condition where the sagittal suture, which runs along the top of the skull, fuses prematurely, affecting the growth of the head.

Signs & Symptoms

Sagittal synostosis is a condition where the sagittal suture in a baby's skull closes too early, before the brain has fully developed. The sagittal suture runs from the front to the back of the skull, and its premature closure prevents normal side-to-side skull growth.

Key characteristics of sagittal synostosis:

- Elongated, narrow head shape (scaphocephaly)
- Prominent forehead (frontal bossing)
- Bulging at the back of the head (occipital bossing)
- No soft spot (fontanelle) or an unusually hard ridge along the sagittal suture
- Increased head circumference in some cases

Causes

Sagittal synostosis occurs when the sagittal suture in a baby's skull closes too early, preventing normal skull growth. The exact cause is not always known, but it can result from a combination of genetic and environmental factors.

1. Genetic Causes

- Sporadic (random) mutations – most cases occur randomly, with no clear hereditary link.
- Familial cases (rare) – in some families, sagittal synostosis is inherited in an autosomal dominant pattern, meaning a parent with the condition has a 50% chance of passing it on.
- Syndromic craniosynostosis – sagittal synostosis can be part of genetic syndromes, such as:
  - Carpenter syndrome
  - Crouzon syndrome
  - Apert syndrome
  - Pfeiffer syndrome

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2. Environmental Factors

- Foetal positioning in the womb – restricted space in the uterus may put pressure on the skull, affecting suture closure.
- Multiple pregnancies (twins, triplets, etc.) – less space in the womb may contribute to differences in skull development.
- Maternal health factors – some studies suggest links between sagittal synostosis and certain prenatal exposures, such as:
  - medications taken during pregnancy.
  - smoking or other environmental toxins.
  - poor nutrition or vitamin imbalances (such as low folic acid levels).

Because the exact cause is often unknown, there is no guaranteed way to prevent it. However, prenatal care, avoiding harmful exposures, and monitoring skull growth in early infancy can help with early detection and treatment.

Testing & Diagnosis

Diagnosis of sagittal synostosis is based on physical examination, imaging studies and, in some cases, genetic testing.

1. Physical Examination

A doctor (often a paediatrician, neurosurgeon, or craniofacial surgeon) will look for a:

- long, narrow head shape (scaphocephaly)
- ridge along the sagittal suture (indicating early fusion)
- prominent forehead and back of the head (frontal and occipital bossing)
- small or absent soft spot (fontanelle)
- lack of normal side-to-side skull widening

2. Imaging Tests

If sagittal synostosis is suspected, imaging can be used to confirm the diagnosis and assess severity:

a. X-rays

- can show early closure of the sagittal suture.
- are less detailed than CT scans but sometimes used for initial evaluation.

b. CT Scan (Computed Tomography)

- is the gold standard for diagnosis.
- shows suture fusion and skull shape differences in detail.
- helps differentiate sagittal synostosis from other skull shape conditions (like positional plagiocephaly).

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3. Genetic Testing (if needed)

- may be done if syndromic craniosynostosis (e.g., Carpenter, Crouzon, Apert syndromes) is suspected.
- involves DNA analysis to check for mutations in craniosynostosis-related genes (e.g., RAB23, FGFR genes).

Treatment

The primary treatment for sagittal synostosis is surgery to address skull shape and allow normal brain growth.

The specific procedure undertaken depends on the child's age and severity of the condition, but may include cranial vault remodelling (i.e., surgery to reshape the skull) or endoscopic surgery in conjunction with springs or helmet therapy. Internationally, including Australia, there is no single preferred type of surgery, with surgical options depending on the craniofacial team and the craniofacial surgeon's expertise and assessment of the individual child.

Individual craniofacial units and craniofacial surgeons in Australia can advise parents about their preferred surgical technique.

Did you know?

In Australia, approximately 2 babies per 10,000 births are diagnosed with sagittal synostosis.*

Some scientists speculate that King Tutankhamun (King Tut) may have had sagittal synostosis, based on the elongated shape of his skull seen in forensic reconstructions. However, this remains debated among historians and medical experts.

References:

*Junaid, M., Slack-Smith, L., Wong, K., Bourke, J., Baynam, G., Calache, H., & Leonard, H. (2022). Epidemiology of Rare Craniofacial Anomalies: Retrospective Western Australian Population Data Linkage Study. The Journal of Pediatrics, 241, 162-172.e169 https://doi.org/10.1016/j.jpeds.2021.09.060

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How we can support you:

- Care packs
- Financial assistance
- Family support coordinator
- Connection to other families