CORONAL SYNOSTOSIS

Otherwise known as?

Coronal synostosis is a type of craniosynostosis in which the coronal suture (the fibrous joint that runs from ear to ear across the top of the skull) fuses prematurely, leading to a different skull and facial shape. While "coronal synostosis" is the most common and widely used term, it may be referred to by other names or descriptions, including:

1. Unilateral coronal synostosis – referring to the premature fusion of one side of the coronal suture.
2. Bilateral coronal synostosis – referring to the premature fusion of both sides of the coronal suture.
3. Anterior plagiocephaly – This term is often used in the case of unilateral coronal synostosis, describing the asymmetrical skull shape that results from one side of the coronal suture fusing prematurely.

Despite these variations, coronal synostosis remains the most commonly used and recognized term to describe this condition.

Signs & Symptoms

Coronal synostosis occurs when the coronal suture in the skull fuses prematurely, which can lead to abnormal head and facial development. The signs and symptoms can vary depending on whether the synostosis is unilateral (affecting one side of the coronal suture) or bilateral (affecting both sides).

1. Head Shape Changes

- Asymmetrical skull shape: one of the characteristic features, especially in unilateral coronal synostosis, is an asymmetrical head. The affected side of the skull may appear flattened, while the opposite side may bulge out, leading to a lopsided or irregular head shape.
  - This is often referred to as "plagiocephaly" (specifically anterior plagiocephaly when caused by coronal synostosis).
- Narrow or tall forehead: in bilateral coronal synostosis, both sides of the coronal suture are fused, leading to a broad, flat, or tall forehead with a noticeable lack of forehead prominence.

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2. Facial Differences

- Orbital irregularities: the eyes may appear uneven, with one eye appearing higher or more forward than the other. This is called ocular hypertelorism.
- Flattened cheeks: the cheekbones on the affected side may appear underdeveloped or flattened.
- Twisted or differences in positioning of the ears: in some cases, the ears may be slightly displaced or shaped differently due to skull differences.

3. Changes in Brain Development

- Increased pressure on the brain: the premature fusion of the coronal suture may limit skull growth, which could lead to increased intracranial pressure. This can cause symptoms like:
  - Headaches
  - Irritability
  - Vomiting
  - Delayed development (including motor and cognitive delays)

4. Delayed development

- Developmental delays: some children may experience delays in motor development (e.g., sitting, crawling, walking) or speech and language delays.
- Cognitive impairment: in more severe cases, the restriction of skull growth due to premature suture fusion can affect brain development, leading to potential intellectual disabilities, although this is not common.

5. Signs in infancy

- Noticeable skull differences: often, the first noticeable signs of coronal synostosis appear within the first few months of life when the infant's skull begins to harden. Parents or doctors may notice an different head shape, especially during routine check-ups.
- Difficulty feeding or sleeping: In some cases, babies may have trouble feeding or sleeping due to discomfort caused by skull differences or increased pressure inside the skull.

6. Possible Hearing Problems

- Hearing issues: in some cases, conductive hearing loss may occur if the different skull shape affects the middle ear or auditory pathways.

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7. Cosmetic Concerns

- Facial asymmetry: due to the impact on cranial and facial development, individuals with coronal synostosis may have noticeable facial asymmetry, leading to cosmetic concerns for the child and parents.

The signs and symptoms of coronal synostosis vary, but the most characteristic features include asymmetrical head shape, facial differences, and potential developmental delays. Early diagnosis and treatment, typically involving surgical intervention, can help alleviate pressure on the brain and improve the child's appearance and overall development.

Causes

Coronal synostosis occurs when the coronal sutures of the skull fuse prematurely, which can lead to abnormal skull and facial development. The exact cause of this condition is not always known, but there are several potential factors that can contribute to its development.

1. Genetic Factors

- Genetic mutations: in many cases, coronal synostosis is associated with genetic mutations which occur in the early stages of foetal development and affect the genes responsible for the growth and development of the skull.
- In some cases, coronal synostosis arises from de novo (new/random) mutations, meaning they occur for the first time in the affected individual and are not inherited from the parents.
- Alternatively, the condition may be inherited in an autosomal dominant pattern, meaning that a child has a 50% chance of inheriting the condition if one parent is affected.
- Coronal synostosis can also be associated with various craniofacial syndromes, including Crouzon, Apert, Pfeiffer and Saethre-Chotzen syndrome.

2. Other Factors

- Although much rarer, environmental factors during pregnancy can potentially contribute to craniosynostosis.
- Multiple births, such as twins or triplets, could contribute to the development of coronal synostosis.

In many cases, the exact cause remains unclear, especially when the condition occurs by itself (not associated with other syndromes).

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Testing & Diagnosis

The diagnosis of coronal synostosis involves a combination of clinical evaluation and imaging studies to confirm the condition and assess its severity.

1. Clinical Examination

- Physical Examination by a doctor:
  - Inspection of the head: looking for characteristic skull differences, such as an asymmetrical head shape, flattened forehead (especially on one side if unilateral), or a broad, flat forehead (if bilateral).
  - Facial features: checking for facial asymmetry, including uneven eye positioning (ocular hypertelorism) or flattened cheekbones.
  - Palpation of the skull: feeling the skull to assess whether the coronal sutures are prematurely fused. In normal skull development, sutures should be flexible and separated, but in coronal synostosis, they may feel rigid or abnormally joined.
- Family history and genetic evaluation: Since coronal synostosis can sometimes be linked to genetic syndromes (like Crouzon syndrome or Apert syndrome), the doctor may ask about the family's medical history of craniofacial conditions. A family history of craniosynostosis or related syndromes may provide clues for further investigation.

2. Imaging Studies

- X-ray: An X-ray of the skull can help identify the early closure of the coronal sutures and visualize any differences in skull shape or flattening. It can also help confirm that the sutures have fused prematurely.
- Computed Tomography (CT) Scan: A CT scan is typically the most detailed imaging test used for diagnosing coronal synostosis. It provides a three-dimensional image of the skull, which allows doctors to:
  - Confirm the fusion of the coronal suture.
  - Assess the extent of the fusion and how it has affected the skull and brain development.
  - Evaluate for increased intracranial pressure (ICP) or any brain changes that might result from the premature fusion.

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Magnetic Resonance Imaging (MRI): An MRI may be used to assess the brain in more detail, particularly if there's a concern about increased intracranial pressure or developmental delays. MRI is less commonly used for diagnosing coronal synostosis directly but may be helpful in assessing the brain's condition in severe cases.

3. Genetic Testing and Counselling

- Genetic Testing: If coronal synostosis is suspected to be part of a genetic syndrome (such as Crouzon syndrome, Apert syndrome, or Pfeiffer syndrome), genetic testing may be recommended. This can help identify specific mutations in genes that are associated with craniosynostosis.
- Genetic Counselling: If a genetic syndrome is suspected, genetic counselling can help parents understand the inheritance pattern, the likelihood of recurrence in future pregnancies, and the potential for other family members to be affected.

4. Developmental and Cognitive Assessment

- In cases where coronal synostosis is more severe and there are concerns about brain development or increased intracranial pressure, doctors may monitor developmental milestones to assess for:
  - Cognitive delays.
  - Motor development delays.
  - Speech or language delays.
  - Vision or hearing impairments.

5. Referral to a Craniofacial Specialist

- Craniofacial team consultation: If coronal synostosis is confirmed, a referral to a craniofacial surgeon or a paediatric neurosurgeon is usually necessary. This team will assess the severity of the condition, discuss treatment options (including surgery), and monitor the child's growth and development.

Early diagnosis of coronal synostosis is critical for determining the appropriate course of treatment. It usually involves a combination of physical examination, imaging studies (especially CT scans), and sometimes genetic testing to identify any underlying syndromes. The diagnosis helps guide treatment decisions, including the possibility of surgical intervention to address skull and facial differences.

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Treatment

The primary treatment for coronal 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.

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 8 babies per 100,000 births are diagnosed with coronal synostosis.*

Unlike other types of craniosynostosis, coronal synostosis is usually more common in females than males.

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
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