LAMBDOID SYNOSTOSIS

Otherwise known as?

Lambdoid synostosis is a type of craniosynostosis where the lambdoid suture (located at the back of the skull) fuses prematurely. It is also known by the following names:

- Lambdoid craniosynostosis
- Posterior craniosynostosis (because the lambdoid suture is at the back of the skull)
- Lambdoid suture synostosis

The term "lambdoid synostosis" is the most commonly used.

Signs & Symptoms

Lambdoid synostosis occurs when the lambdoid suture, located at the back of the skull, fuses prematurely. This condition can lead to skull and facial differences. The signs and symptoms of lambdoid synostosis can vary depending on the severity and whether the condition affects one side (unilateral) or both sides (bilateral) of the skull. Here are the common signs and symptoms:

1. Skull Differences

- Flattening of the back of the head (also known as posterior plagiocephaly): The most noticeable sign of lambdoid synostosis is flattening on one side of the back of the head. In unilateral lambdoid synostosis, one side of the head is flatter than the other, while bilateral synostosis can cause an overall flattening of both sides.
- Asymmetry of the skull: The skull may appear asymmetrical, particularly at the back, with a noticeable tilting or twisting of the head.
- Raised area on the fused side: In some cases, there may be a slight bulging or raised ridge on the side of the fused suture due to differences in skull growth.

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Craniofacial Australia
204 Melbourne Street
North Adelaide
South Australia, 5006

W: craniofacial.com.au
E: familysupport@acmff.org.au
P: (08) 8267 4128

2. Facial Asymmetry

- Ear displacement: In cases of unilateral lambdoid synostosis, the ear on the affected side may appear slightly displaced or pushed forward, as the skull is unable to expand normally.
- Uneven facial features: Although less common than in other types of craniosynostosis, facial asymmetry may occur, with one side of the face appearing slightly more prominent than the other due to the differences in skull shape.

3. Developmental Delays

- In some cases, especially if there is increased intracranial pressure (ICP) or the synostosis affects brain development, there may be:
  - Delayed motor skills or developmental milestones.
  - Learning or cognitive delays (though this is less common in lambdoid synostosis compared to other forms of craniosynostosis).

4. Increased Intracranial Pressure (ICP)

- If lambdoid synostosis leads to restricted brain growth or causes pressure inside the skull, signs of increased intracranial pressure may develop, including:
  - Headaches.
  - Vomiting.
  - Irritability or fussiness.
  - Bulging fontanelle (the soft spot on a baby's head).
  - Poor feeding or difficulty sleeping (especially in infants).

5. Neck and Head Posture

- Torticollis (twisted neck): Some infants with lambdoid synostosis may develop torticollis, a condition where the head tilts to one side due to the shortening or tightening of neck muscles. This is often caused by the skull differences affecting the positioning of the neck.

6. Visual or Hearing Problems

- In some cases, visual or hearing issues may arise, but these are more common when lambdoid synostosis is associated with other syndromes (e.g., Crouzon syndrome or Apert syndrome). However, if there is significant facial asymmetry, the position of the ears and eyes may be altered, which can affect hearing and vision in rare cases.

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Causes

The exact cause of lambdoid synostosis (the premature fusion of the lambdoid suture at the back of the skull) is not always clear, but several factors can contribute to its development. It may occur as an isolated condition or as part of a genetic syndrome.

1. Genetic Factors

- Inherited mutations: In some cases, lambdoid synostosis may be caused by genetic mutations that affect the growth and development of the skull. These mutations can lead to the premature fusion of one or both lambdoid sutures.
- Genetic syndromes: Lambdoid synostosis can sometimes be associated with certain genetic syndromes that affect cranial development such as Crouzon, Apert, Pfeiffer and Saethre-Chotzen syndrome. In these syndromes, the cranial differences result from mutations in specific genes that affect bone development, such as the FGFR2 gene in Crouzon and Apert syndromes.

2. Environmental Factors

- Environmental factors are less commonly associated with lambdoid synostosis, but some external influences during pregnancy might contribute to the condition.

3. Positional Factors

- Positional plagiocephaly (non-synostotic positional flattening) is sometimes confused with lambdoid synostosis, but it is caused by external pressures on the skull during infancy. This condition is not due to premature fusion of the sutures, but it can lead to similar flattening of the skull. It typically resolves with changes in head positioning and should improve gradually as the baby grows. However, in true lambdoid synostosis, the suture itself is prematurely fused, and skull shape differences persist.

In many cases of lambdoid synostosis, no underlying genetic syndrome or environmental factor is identified. Although the cause is unknown it may involve complex genetic or developmental factors that are not yet fully understood.

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Testing & Diagnosis

Testing and diagnosis of lambdoid synostosis typically involve a combination of physical examination, imaging studies, and in some cases, genetic testing to rule out associated syndromes. Early diagnosis is important for determining the severity of the condition and deciding on the appropriate treatment, such as surgery or other interventions.

1. Physical Examination

The first step in diagnosing lambdoid synostosis is a thorough physical exam conducted by a doctor, usually a paediatrician or a craniofacial specialist. The doctor will assess:

- Skull shape and symmetry: Flattening on one side of the back of the head, asymmetry, or abnormal head position (tilting or twisting).
- Facial features: Ear displacement or facial asymmetry that may indicate cranial differences.
- Head posture: Any signs of torticollis (twisted neck), which can sometimes occur with lambdoid synostosis.
- Palpation of sutures: The doctor will gently feel the skull for any ridges or areas of hardness that could indicate early suture fusion.

2. Imaging Studies

Imaging is crucial for confirming the diagnosis and assessing the severity of the synostosis. Commonly used imaging methods include:

- X-rays: X-ray imaging can show the premature fusion of the lambdoid suture. It may help visualize the skull shape and identify areas of fusion, although X-rays are less detailed compared to other imaging techniques.
- CT (Computed Tomography) Scan: A CT scan is the most commonly used imaging tool for diagnosing lambdoid synostosis. It provides detailed cross-sectional images of the skull and can clearly show the premature fusion of the lambdoid suture. It helps identify:
  - The extent of the suture fusion.
  - Any associated differences in the skull and brain.
  - Potential intracranial pressure (ICP) concerns.
  - The need for surgical intervention.
- MRI (Magnetic Resonance Imaging): An MRI is sometimes used to assess the brain's development and structure, especially if there are concerns about increased intracranial pressure (ICP) or developmental delays. MRI can show how the skull shape differences might be affecting brain growth.

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3. Genetic Testing

If lambdoid synostosis is suspected to be part of a genetic syndrome (e.g., Crouzon syndrome, Apert syndrome, or Saethre-Chotzen syndrome), genetic testing may be recommended to confirm the diagnosis. Genetic testing can help identify mutations in genes like FGFR2 or FGFR3, which are associated with these conditions.

- Genetic counselling may also be offered to families to understand the inheritance patterns, potential risks for future pregnancies, and the possibility of associated conditions.

4. Additional Evaluations

In cases where lambdoid synostosis is suspected to be part of a broader craniofacial syndrome, additional assessments may include:

- Ophthalmological evaluation: To check for any eye-related issues, such as strabismus (crossed eyes) or optic nerve problems.
- Audiological testing: Hearing tests to evaluate any hearing loss, which may occur with craniofacial conditions.
- Neurodevelopmental evaluation: To assess any delays in motor skills, cognitive development, or speech.

Differential Diagnosis

- Positional plagiocephaly: A common condition where the skull flattens from external pressure, such as lying on one side too much. Unlike lambdoid synostosis, positional plagiocephaly does not involve suture fusion and often improves gradually as the baby grows, becomes more mobile, and pressure on the flattened area is reduced. It can usually be diagnosed through physical examination, with imaging used when needed to rule out suture fusion.

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Treatment

The primary treatment for lambdoid synostosis is surgery to address skull shape and allow 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 6 babies per 100,000 births are diagnosed with lambdoid synostosis.*

Lambdoid synostosis is often confused with positional plagiocephaly, a much more common and benign condition caused by prolonged pressure on one side of a baby's head. However, unlike positional flattening, lambdoid synostosis causes a true difference in skull shape.

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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