Craniosynostosis - making the head fit the hat

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Aim

Craniosynostosis refers to the premature fusion of one or more of the cranial sutures. This occurs in around 1 in 2500 live births [1]. Fusion can involve part or all of a suture, or multiple sutures, and severity ranges from mild (not requiring surgical correction) to severe and potentially life threatening due to cerebral compression and raised intracranial pressure. Causes include mechanical (eg shunted hydrocephalus), metabolic (eg hyperthyroidism), and genetic (eg Crouzon syndrome). The syndromic forms of craniosynostosis are frequently associated with other anomalies, including deformities of the limbs. The aim after reading this presentation is to be able to recognize the radiological features of craniosynostosis and the common associated syndromes.
Methods and materials

Normal Closure of the Cranial Sutures.

Most of the sutures in the cranial vault remain patent until adulthood. The sagittal, coronal and lambdoid sutures all begin fusion in the early to mid twenties. The metopic suture is quite variable, and can begin fusing normally as early as 3 months of age [2]. This can make the diagnosis of metopic synostosis difficult, and makes the secondary signs such as trigonocephaly and "quizzical" orbits very important. The sutures of the skull base (except the spheno-occipital synchondrosis) are fused by 3 years of age, and contribute little to skull base growth postnatally.

Imaging in Craniosynostosis.

Initial evaluation of a clinically suspected craniosynostosis is with plain skull x-rays. Frontal, lateral, Townes and submentovertical (SMV) views should be performed to visualise all the major sutures. Operative planning is with low dose CT scanning and 3D reconstructions. Syndromic craniosynostosis is usually also imaged with MRI (including dynamic CSF studies), as there is an association with brain anomalies, Chiari I malformation and cord syrinx.

Isolated Sutural Synostoses.

Sagittal Synostosis.

Premature fusion of the sagittal suture is the most common craniosynostosis, accounting for 40 - 60% of cases [1]. Around 80% are isolated, and 75% occur in boys [3]. The skull becomes scaphocephalic in shape (Fig. 1 on page 5) due to lack of growth at the sagittal suture (Fig. 2 on page 5). Growth still occurs at the metopic and coronal sutures producing frontal bossing, mild hypertelorism and anterior inclination of the forehead (Fig. 3 on page 6). Normal growth at the lambdoid sutures produces occipital bulging. The skull base is normal.

Metopic Synostosis.

About 10% of craniosynostoses involves the metopic suture, and the majority (75 - 80%) are isolated, non-syndromal synostoses [1]. As with sagittal synostosis, males are more often affected than females. The fusion of the metopic suture produces a triangular shape...
to the cranium (Fig. 4 on page 7) (trigonocephaly) with hypotelorism (Fig. 5 on page 8), or "quizzical" orbits (Fig. 6 on page 9). The usual "M" shape at the anterior falx becomes "omega"-shaped [2] (Fig. 7 on page 10). The anterior cranial fossa is narrowed and the ethmoids are underdeveloped. There is an association with midline cerebral abnormalities, and the brain must be carefully evaluated. There is also an association with maternal valproate intake, as part of the Valproate Syndrome [4].

**Coronal Synostosis.**

Premature fusion of the coronal suture can be unilateral or bilateral. Isolated unilateral coronal synostosis represents 20 - 30% of craniosynostoses [1]. It differs from the midline synostoses in that females are over-represented, and the skull base is often involved. Bilateral coronal synostosis is typically syndromal and more likely to be associated with limb anomalies [3].

The coronal sutural fusion produces a "harlequin orbit" on the synostotic side due to elevation of the sphenoid wings (Fig. 8 on page 11). There is variable proptosis due to involvement of the frontosphenoid suture, and variable degrees of midface hypoplasia due to base of skull involvement. The cranial vault becomes plagioccephalic (Fig. 9 on page 12) in shape if only one suture is fused (Fig. 10 on page 13), or brachycephalic ("short cranium") if fusion is bilateral. Hypertelorism is also common in bilateral fusion.

**Lambdoid Synostosis.**

Isolated fusion of the lambdoid suture is rare, occurring in < 5% of craniosynostoses [3]. Careful radiologic evaluation is needed to distinguish the condition from positional ("deformational") plagioccephaly (Fig. 11 on page 14), which is treated conservatively. True lambdoid synostosis produces unilateral occipital flattening with ipsilateral occipitomastoid bossing, producing a "mastoid bump" (Fig. 12 on page 15) due to growth at the patent posterior fontanelle. There is contralateral frontal and parietal bossing, and the skull vault is trapezoid in shape. The axis of the skull base swings towards the affected side due to growth arrest [5] (Fig. 13 on page 16).

Isolated bilateral lambdoid fusion is exceedingly rare, and produces a tall cranium (turricephaly). There is typically underlying rhombencephalosynapsis [1].
Fig. 1: Lateral skull x-ray in a 4 month old male. The skull is scaphocephalic in shape.

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**Fig. 2:** Townes view in a 4 month old male. The sagittal suture is fusing producing a prominent ridge (arrow) typical of sagittal synostosis.

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Fig. 3: 3D bony CT reconstruction of a 4 month old male. The sagittal suture is fused producing a prominent midline ridge, and the skull is scaphocephalic in shape. The remaining sutures are clearly patent.

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**Fig. 4:** Skull x-ray (SMV view) in a 3 month old male. There is trigonocephaly due to fusion of the metopoiic suture (arrow).

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**Fig. 5:** Frontal skull x-ray in a 3 month old male. There is hypotelorism, producing a "quizzical" appearance of the orbits.

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Fig. 6: 3D bony CT reconstruction in a 3 month old male. The ridge at the fused metopic suture site is clearly seen. The orbits have a typical "quizzical" appearance due to hypotelorism, and the skull is trigonocephalic in shape.

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Fig. 7: Axial CT image in a child with metopic synostosis. The inner table of the skull is omega-shaped anteriorly at the site of the metopic sutural fusion (arrow).

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Fig. 8: Frontal skull x-ray in an 8 month old male. There is elevation of the left sphenoid wing (arrow) producing a "harlequin" appearance to the orbit.

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**Fig. 9:** Skull x-ray, SMV view, in an 8 month old male. This view is very important to show the plagiocephalic shape of the skull.

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**Fig. 10:** 3D bony CT reconstruction of an 8 month old male with uni-coronal synostosis. The left coronal suture is fused while the right remains patent. Plagiocephaly is well demonstrated.

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Fig. 11: 3D bony CT reconstruction of a 5 month old male with plagiocephaly. All the major sutures are patent, the plagiocephaly is deformational or positional.

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**Fig. 12:** 3D bony CT reconstruction in a 6 month old male with plagiocephaly. The left lambdoid suture is fusing, the skull is plagiocephalic with a large mastoid bump.

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**Fig. 13:** Reconstructed CT base of skull "cut-down" view in a 6 month old male. The skull base has twisted, with the axis swinging towards the affected left side.

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Results

Multiple Sutural Synostoses.

Multiple premature sutural fusions are typically syndromal. Bilateral coronal synostosis is the most common; pansynostosis can occur following a severe hypoxic-ischaemic insult with resultant cerebral under-development. Fusion of both coronal sutures as well as the sagittal suture produces a "clover-leaf" skull (Fig. 14 on page 21), due to expansion of the membranous calvarial bones between the sutures. The combination of Crouzon syndrome, Pfeiffer syndrome, and Apert syndrome account for most syndromal craniosynostoses [6].

Crouzon Syndrome.

Crouzon syndrome (craniofacial dysostosis) represents ~4% of craniosynostosis cases [7] (Fig. 15 on page 21 Fig. 16 on page 22). Inheritance is autosomal dominant, although a quarter of cases are sporadic. The clinical picture includes:

• brachycephaly more often than scaphocephaly or trigonocephaly
• parrot beaked nose
• bilateral exophthalmos and hypertelorism
• relative mandibular prognathism (to short midface)
• variable degrees of mental retardation
• poor vision

Radiologically, the craniofacial deformities include:

• premature fusion of any or all of the cranial sutures
• hydrocephalus
• Chiari 1 malformation in 70%
• syringomyelia in 20%

Pfeiffer Syndrome.

Acrocephalosyndactyly or Pfeiffer syndrome occur in around 1 in 100 000 births [8] (Fig. 17 on page 23 Fig. 18 on page 24). Inheritance is autosomal dominant, although commonly sporadic. The clinical picture includes:

• acrocephaly, brachcephaly
• hypertelorism
• proptosis
• beaked nose
• high, arched palate
• soft tissue syndactyly of the fingers and toes
• variable degrees of mental retardation

Radiologically, the deformities include:

• sagittal and bilateral coronal synostosis
• hypertelorism
• shallow anterior cranial fossa
• brachydactyly, clinodactyly
• broad thumbs with ulnar deviation, broad great toes with tibial deviation
• tarsal and/or carpal bone fusion

Apert Syndrome.

Apert syndrome occurs in approximately 1 in 250 000 births and has an autosomal dominant inheritance [7] (Fig. 19 on page 25 Fig. 20 on page 26). The clinical manifestations include:

• macrocrania, turribrachycephaly
• hypertelorism
• proptosis
• depressed nasal bridge
• high arched or cleft palate, open mouth
• dental anomalies
• "mitten hand" and "sock foot"
• mental retardation

Radiological abnormalities include:

• premature sutural fusion, especially coronals
• midface hypoplasia
• ventriculomegaly
• callosal agenesis
• syndactyly of the 2nd to 5th digits, more common in the hand than foot
• progressive cervical vertebral fusions

Post-Surgical Complications.

The extensive vascular supply of the skull and scalp predicts significant blood loss during corrective surgery for craniosynostosis, and most infants require transfusion during or
after surgery. Infection is another serious complication, due to exposure of the brain and meninges (Fig. 21 on page 27). A small percentage of patients will require further surgery for "restenosis", and hydrocephalus requiring shunting, and shunt blockage will also affect a number of children.
Fig. 14: Skull x-ray in a 3 year old child. There is global craniosynostosis producing a skull shape resembling a clover-leaf. The copper-beaten appearance indicates raised intra-cranial pressure due to the global synostosis.

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Fig. 15: 3D CT reconstruction in a 12 year old male with Crouzon syndrome. The skull is turribrachycephalic in shape, and there is global craniosynostosis. There is midface hypoplasia with relative prognathism, and mild hypertelorism.

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Fig. 16: Sagittal T2 weighted imaged in a 12 year old male with Crouzon syndrome. There is a Chiari 1 malformation (tonsillar herniation, solid arrow) and a large cervial cord syrinx (dashed arrow). Note also the dilated cerebral ventricles.

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**Fig. 17:** 3D reconstructed CT in a 7 year old male with Pfeiffer syndrome. There is marked turribrachycephaly with proptosis and hypertelorism. The ears are low set and there is global craniosynostosis.

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Fig. 18: X-ray of the right hand in a child with Pfeiffer syndrome. Note the very broad thumb, and carpal fusion.

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Fig. 19: 3D reconstructed CT of a 3 year old child with Apert syndrome. The head is large, and turribrachycephalic with global craniosynostosis. There is hypertelorism and proptosis, and midface hypoplasia with a depressed nasal bridge.

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**Fig. 20:** X-ray of the left hand of an older child with Apert syndrome. There is soft tissue syndactyly producing a "mitten-hand", and the middle 3 digits show bony fusion classical of the syndrome.

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**Fig. 21:** Post-operative 3D bony CT reconstruction in a child with sagittal synostosis. The skull reshaping is extensive, with extended barrel-staving osteotomies, and often fronto-orbital advancement. Simple excision of the fused suture does not correct the skull shape and prevent re-stenosis.

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Conclusion

Identification and management of craniosynostosis is only partly for cosmetic reasons. The early fusion of the cranial sutures can cause brain damage if not treated at an early age. Recognition of the radiological features of these synostoses is crucial for treatment to be successful.
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