Craniosynostosis diagnosis

Some cases of “synostosis” are really deformities caused by positional flattening (e.g. “lazy lambdoid”, see below). If this is suspected, instruct parents to keep head off of the flattened area and recheck patient in 6–8 weeks: if it was positional, it should be improved, if it was CSO then it usually declares itself. The diagnosis of CSO may be aided by:

1. palpation of a bony prominence over the suspected synostotic suture (exception: lambdoidal synostosis may produce a trough)

2. gentle firm pressure with the thumbs fails to cause relative movement of the bones on either side of the suture

3. plain skull X-rays:

a) lack of normal lucency in center of suture.Some cases with normal X-ray appearance of the suture (even on CT) may be due to focal bony spicule formation

b) beaten copper calvaria, sutural diastasis and erosion of the sella may be seen in cases of increased ICP

4. CT scan:

a) helps demonstrate cranial contour

b) may show thickening and/or ridging at the site of synostosis

c) will demonstrate hydrocephalus if present

d) may show expansion of the frontal subarachnoid space

e) three-dimensional CT may help better visualize abnormalities be demonstrated

5. in questionable cases a technetium bone scan can be performed

a) there is little isotope uptake by any of the cranial sutures in the first weeks of life

b) in prematurely closing sutures,increased activity compared to the other(normal)sutures will be demonstrated

c) in completely closed sutures, no uptake will be demonstrated

6. MRI: usually reserved for cases with associated intracranial abnormalities. Often not as helpful as CT

7. measurements, such as occipito-frontal-circumference may not be abnormal even in the face of a deformed skull shape

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Commonly, craniosynostosis is present at birth, but it is not always diagnosed when mild. Usually it is diagnosed as a cranial deformity in the first few months of life. The diagnosis relies on physical examination and radiographic studies, including plain radiography and computed tomography (CT). Clinical history should include complications of pregnancy, duration of gestation, and birth weight [1)].

Premature fusion of the cranial sutures restricts cranial growth perpendicular to the affected suture with compensatory overgrowth along the other patent sutures. This results in the characteristic skull shape deformities noted in craniosynostosis. Diagnostic imaging is necessary to confirm the fused suture and to assess the accompanying skull deformities, intracranial pathology and other complications. A prematurely fused suture shows perisutural sclerosis, linearity, reduced serration, bony bridging or the absence of the suture on a plain skull radiography or CT image. Secondary signs of increased ICP, such as a “copper-beaten” appearance, are also observed in severe cases [2)].

Soboleski et al. [3)] reported the ultrasonographic findings of craniosynostosis as follows : 1) the loss of the hypoechoic fibrous gap between hyperechoic body plates; 2) an irregular, thickened inner sutural margin; 3) the loss of a beveled edge; and 4) asymmetric fontanels. On “Black Bone” MRI, the affected fused sutures are demonstrated as absence of suture [4)].

A normal patent suture is demonstrated as a radiolucency, serrated and nonlinear line on plain skull radiography and 3D-CT images [5) 6) 7) 8)].

Ultrasonography shows a normal patent suture as an uninterrupted hypoechoic fibrous gap between hyperechoic cranial bones with an end-to-end appearance on a transverse scan of the sagittal sinus and a beveled appearance on a transverse scan of the coronal and lambdoid sutures [9) 10) 11)]

Conventional MRI has typically been unreliable in identifying sutures individually. However, Eley et al. described a novel gradient echo MRI sequence (“Black Bone”) that minimizes soft tissue contrast to enhance the bone-soft tissue boundaries and can demonstrate normal patent cranial sutures as hyperintensity distinguished from the signal void of the cranial bones [12)].

Proisy et al. from Rennes first described a high-resolution sonography technique and its limitations. They then analyzed the reliability, effectiveness and role of ultrasonography in routine practice using a PubMed literature review.

Ten studies reported excellent correlations between ultrasonography and 3D-CT. Cranial US for the diagnosis of a closed suture had 100% sensitivity in 8 studies and 86-100% specificity before the age

of 12 months. Negative findings mean imaging investigation can be stopped. If ultrasonography confirms diagnosis, neurosurgical consultation is required. Thus, 3D-CT can be postponed until appropriate before surgery.

Cranial suture ultrasound is an effective and reliable technique for the diagnosis of craniosynostosis. It has many advantages: it is fast and non-irradiating, and no sedation is required. It should be used as first-line imaging in infants below the age of 8-12 months when craniosynostosis is clinically suspected. [13].

Head CT

The majority of surgeons obtain preoperative head CTs, whereas only 25% obtain CTs postoperatively, often to evaluate outcomes. Because outcomes may be evaluated clinically, this is a poor use of resources and exposes children to radiation. Consensus guidelines are needed to create best practices and limit unnecessary studies [14].

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