Early Diagnosis of Harlequin Eye and Other Craniosynostoses Is Key

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Working as a team with referring clinicians, pediatric radiologists can facilitate the early detection of this important pediatric diagnosis.

Clinicians who care for children are often faced with the task of evaluating an infant for abnormal skull shape. Most cases of localized skull flattening are caused by deformational or positional plagiocephaly, particularly given the successful implementation of the “back-to-sleep” campaign. Although abnormal skull shape is due to positioning in the majority of infants, it is sometimes due to craniosynostosis, or premature fusion of a cranial suture or sutures. Craniosynostosis is a serious clinical problem that can require surgical treatment, so differentiation between craniosynostosis and positional plagiocephaly is important.

In general, true craniosynostosis is ten-fold less common than positional plagiocephaly. Of the craniosynostoses, most involve just one suture and are isolated events, rather than being part of a genetic syndrome. The suture most frequently involved by craniosynostosis is the sagittal suture (40 to 60 percent of cases), followed by the coronal suture (20 to 30 percent of cases). When coronal suture synostosis is unilateral, the result is one of the more recognizable clinical and radiographic presentations of craniosynostosis, that of the “harlequin eye” deformity of the orbit. The “harlequin eye” is meant to describe elevation of the ipsilateral lesser wing of the sphenoid, with posterior displacement or retraction of the superior and lateral rims of that orbit.

A more common, and perhaps more challenging, clinical scenario is that of distinguishing between positional plagiocephaly and the unusual but clinically important lambdoid craniosynostosis. The evaluation begins with the physical examination findings, and there are features here that can help differentiate the two. Both can cause occipitoparietal flattening, but in positional plagiocephaly, when examining the infant from above, one sees frontal bossing and anterior displacement of the ear on the same side as the posterior (occipitoparietal) skull flattening. To the contrary, in the rare cases of lambdoid synostosis, one may see frontal and parietal bossing on the opposite side of the posterior (occipitoparietal) flattening, and the ear on the side of the occipitoparietal flattening may be displaced posteriorly, toward the fusing suture. With lambdoid synostosis, one may also see a “mastoid bump” on the same side as the posterior flattening, as growth can still occur at the patent posterolateral fontanel.

When a clinician encounters an infant with abnormal skull shape or facial features that cannot be attributed to positional plagiocephaly, referral to a pediatric radiologist is an important next step. A radiographic skull series serves as the appropriate screening study in evaluation of such infants. The pediatric radiologist evaluates the radiographs for any signs of craniosynostosis such as increased sclerosis at the sutures, narrowing of a suture or loss of suture clarity, or frank bony bridging across a suture. In addition, if performed at a dedicated pediatric imaging center, the pediatric radiologist has the opportunity to examine the patient when indicated. If an abnormality is detected, a low-dose head CT that is tailored to the evaluation of craniosynostosis can be performed. With this head CT, the infant is assessed for additional abnormalities that could not be appreciated on skull radiographs, recognizing that more than one suture may be affected in a minority of patients. In addition, using the pediatric craniosynostosis protocol, three-dimensional reconstructed images can be rendered to aid the surgeon in his or her treatment planning. Working as a team with referring clinicians, pediatric radiologists can facilitate the early detection of this important pediatric diagnosis.

A pediatric radiologist is available for consultation weekdays from 8am to 5pm at the Wake Radiology Pediatric Imaging Center, 4301 Lake Boone Trail in Raleigh, (919) 782-4830.

References