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A Case report of Craniosynostosis

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Abstract : Craniosynostosis is a disorder of skull development occurs in approximately 1 in 2000 live births with 31 male predominance8. When an abnormal calvarial configuration detected, a thorough radiological evaluation necessary to characterize the deformity and to guide the corrective surgical procedure. Here we report a case of Craniosynostosis in a 1 year old male child.

Keyword : Crouzon syndrome, Craniosynostosis, Proptosis, copper beaten appearance.

Introduction :

Craniosynostosis is defined as premature closure of one or more of the cranial sutures and is classified as primary and secondary types. It is associated with varying types of abnormal skull shape. Lack of growth at the fused suture in combination with compensatory overgrowth of normal sutures results in characteristic skull shape anomalies. Primary craniosynostosis refers to closure of one or more sutures owing to abnormality of skull development, whereas secondary craniosynostosis results from failure of brain growth and expansion. Isolated premature fusion of a single suture causes a predictable cranial deformity that can typically be recognized without the need for radiologic imaging by an experienced practitioner. However, complex craniosynostosis, consisting of multiple suture fusions, can be difficult to diagnose without radiologic imaging2.

Case report:

The subject is a third born male child to parents of second degree consanguinity. The child was apparently normal at birth, and then the mother noticed skull and eye deformity one month later. History of delayed milestones present. The child was brought with complaints of increasing proptosis and recurrent Lower respiratory infections. On examination the child had Turricephaly, bilateral proptosis and umbilical hernia. widely open anterior fontanelle, fundus examination was normal. Plain radiograph showed significantly exaggerated convolution marking noted on the skull vault [copper beaten appearance]. No sutural line could be demonstrated. Bilateral bony orbits poorly developed. Turricephaly noted. Spine screening showed no abnormality. Findings in the CT are fused coronal, lambdoid, sagittal sutures with open anterior fontanelle seen. Altered shape of cranium seen with multiple focal defects in outer and inner table of calvaria. Poorly formed both orbits seen with bilateral proptosis, dilated supratentorial ventricular system seen. Effacement of basal cisterns and bilateral Cerebello-Pontine angle cistern were seen. Cerebral parenchyma showed normal attenuation, grey white differentiation was well preserved with no midline shift. Brain stem and cerebellum were normal in morphology and attenuation. The clinical and radiological pictures are suggestive of Crouzon syndrome.

Figure : 1 | Xray Skull Ap view showing Turricephaly

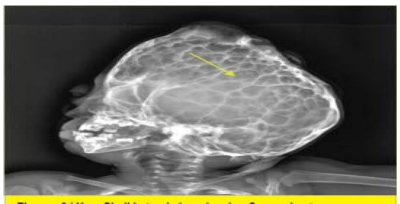
Figure : 2 | Xray Skull Lateral view showing Copper beaten apperance

Discussion:

The skull develops from the viscerocranium (responsible for development of the facial bones) and the neurocranium (the portion of the skull that surrounds the brain). The neurocranium has cartilaginous (chondrocranium that develops from endochondral ossification and gives rise to the skull base) and membranous (dermatocranium that develops from membranous ossification and gives rise to the calvarial vault) components. The brain grows rapidly in the early years of life, with growth of the neurocranium essentially ceasing at about 7 years of age6. The infant skull undergoes a period of rapid expansion during the first year of life, which is driven by brain growth. Bone growth at patent cranial sutures causes calvarial expansion in a distinct morphologic pattern. Typically, suture fusion occurs from anterior to posterior and lateral to medial. The metopic suture, which normally closes by 8 to 9 months of

age, is the only suture to close completely during infancy; the remaining sutures do not completely fuse until adulthood. Most patients presenting with craniosynostosis have prenatal onset of suture fusion. However, in severe cases of syndromic craniosynostosis, progressive, multiple suture fusion can occur over the first 3 to 4 years of life. In this situation, the calvarial deformity is typically not detected at birth except when quite severe. After a few months of rapid growth, the deformity typically becomes apparent1. Multiple suture synostosis is more frequently syndromic with Crouzon and Apert being most common. Children with syndromic Craniosynostosis demonstrate multiple cranial, facial abnormalities. Growth progresses forward toward forehead with Parieto-occipital areas having least growth. Orbits have a typical roof leading to exophthalmos and proptosis. Maxilla fails to progress downward and forward and this type of growth can lead to airway compromise and nasopharyngeal constriction10. A number of dural-related cytokines, such as heparin-binding factor, fibroblast growth factor (FGFs), bone morphogenic proteins (BMPs), transforming growth factor (TGF-B) and transcription factors Msx2 and TWIST, have a role in the regulation and coordination of suture patency. FGF receptor mutations causing constitutive activation of the receptor occur in many of the human craniosynostosis syndromes including Apert, Crouzon, Muenke, and Pfeiffer syndromes. one mechanism of bony fusion across the suture occurring with the FGF-R mutation is the loss of Noggin expression in the involved suture mesenchyme. when Noggin is not 7p,r1e1sent, bone forms across the mesenchyme and the suture fuses .

Crouzon syndrome:

It is the most common syndromic craniofacial dysostosis, with an estimated incidence of 1 in 25,000 live births. Crouzon syndrome is caused by a mutation in the FGFR2 gene, causing increased receptor activation. The syndrome is characterized by the triad of bicoronal craniosynostosis, exorbitism, and midface retrusion. Exorbitism is the feature of this syndrome, which results in prominent globes. As such, ocular protection is a key feature the clinician must keep in mind when assessing these children. Exposure keratitis and conjunctivitis may prompt earlier surgical intervention to avoid permanent visual impairment 5,9.

Conclusion:

A thorough history and physical examination is the cornerstone of diagnosing synostosis of the calvarial sutures3. Early Surgical Correction of craniosynostosis is performed in the hope of limiting or preventing neuro developmental sequelae4.

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