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A Young Boy Presenting with Headache: Crouzon Syndrome

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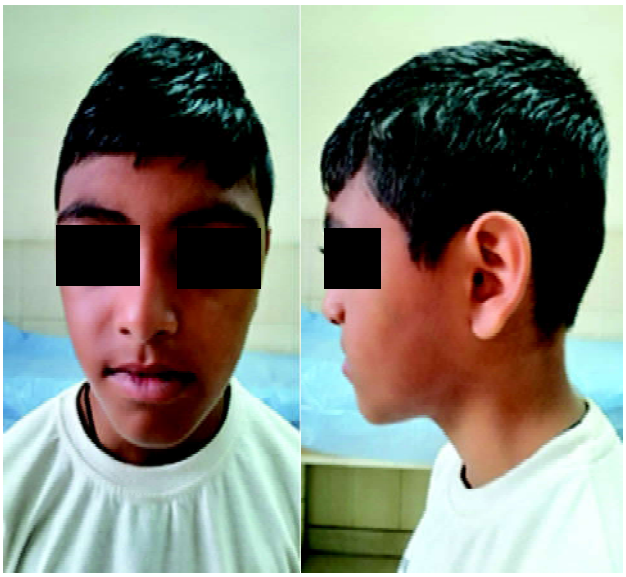
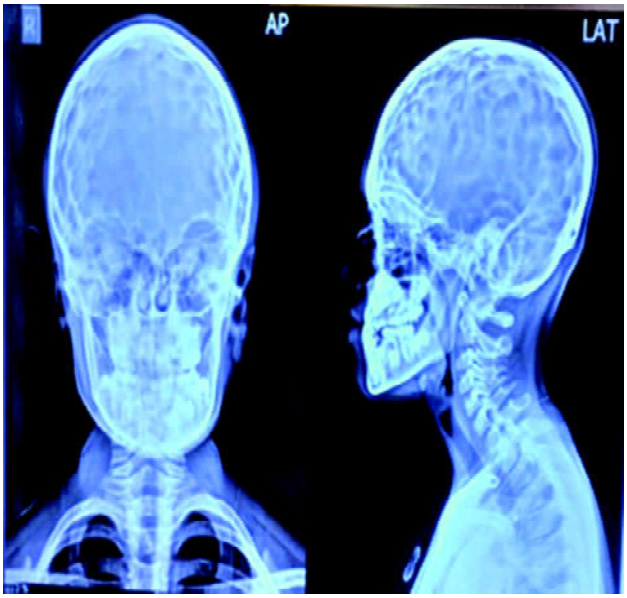
A 12-year-old boy presented with recurrent headaches for last three years. The headache was frontal in location and responded to analgesics. For the last 6 months he complained of persistent headache with blurring of vision and poor response to pain relievers. There was history of increasing proptosis for last few months. The antenatal and neuro-developmental history was normal with the child attending regular school. Examination revealed a conscious boy with normal intelligence. The child had an abnormal skull shape with proptosis as shown in the figure. Fundus examination revealed bilateral papilloedema. There were no focal neurological deficits and other systems were normal on examination. A Plain X-ray skull revealed a characteristic silver beaten appearance – Figure. A diagnosis of Crouzon syndrome was made.

Crouzon syndrome, also known as craniofacial dysostosis or acrocephalosyndactyly, is a complex genetic syndrome that occurs in about 1 in 61,000 newborns and ranks among the most prevalent types of craniofacial dysostosis[1]. It is caused by a mutation (inherited or sporadic) in the Fibroblast Growth Factor Receptor 2 (FGFR2) or the FGFR3 gene located on chromosome 10. Crouzon syndrome is characterised by a classic trio of an atypical skull structure, distinctive facial features, and protruding eyes. Crouzon syndrome causes the skull's sutures to fuse together too early (craniosynostosis) resulting in an abnormal head and face





shape. It can be syndromic or non-syndromic, the latter being more common. Non-syndromic craniosynostoses are not associated with other body dysmorphisms and usually affect only one suture of the skull, while syndromic craniosynostoses are known to affect multiple skull sutures and are associated with craniofacial dysmorphisms, abnormalities of extremities, and other bony anomalies[2].

Commonly, fusion of bilateral coronal sutures causes brachycephaly (short, wide skull) as seen in this case. The main mechanism behind papilloedema is postulated to be high intracranial pressure (ICP), secondary to anomalous skull development. Radiology reveals a copper beaten appearance of skull, lacunae in the skull and shallow orbits. A multidisciplinary team (typically including a pediatrician with dysmorphology training, a craniofacial surgeon, a neurosurgeon, an oculo-facial plastic surgeon, and an oral maxillofacial surgeon) is needed for optimum management such that patients can achieve maximum functional status and relief of chronic headache.

References

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