Clinical Manifestations of Cruzon Syndrome.

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Abstract: Cruzon syndrome present similarly to the one we presented in this image, and genetic and radiological studies are used to confirm the diagnosis. This image shows the typical clinical and radiological features of Cruzon syndrome, so that if these are present in a patient, the diagnosis of Cruzon syndrome should be presumed.

Key words: Cruzon syndrome, Craniosynostosis

Key clinical message: Cruzon syndrome is the most common syndromic craniosynostosis characterized by craniofacial dystosis. The syndrome shares some clinical features with other conditions such as Apert syndrome.

Case presentation: A 17 years old female for facial deformities since birth. On examination, she had dysmorphic cranial and facial features curtailing enlarged cranial vault with frontal bossing, maxillary hypoplasia and relative mandibular proganthism. Ocular manifestations such as shallow orbits, hypertelorism, bilateral proptosis, exophthalmos and strabismus were present. Other facial features included depressed nasal bridge, wide spaced teeth, highly arched palate and low set ears. Her hands were normal but feet showed syndactyly bilaterally. X-ray skull lateral view showed copper beaten appearance and fused 2^{nd} and 3^{rd}cervical and 4^{th} and 5^{th} cervical vertebrae. (Figure.1) The patient was born as a result of consanguineous marriage at 28 week of gestation through normal vaginal delivery. Family history was positive for similar condition and seizures in one brother who died at the age pf 15 years. Genetic studies revealed mutation of fibroblast growth factor receptor 2. Based on the clinical features, x ray findings and genetic what is the most likely diagnosis?

Discussion/ conclusion: The clinical features of Cruzon syndrome includes obliterated coronal and sagittal sutures, flattening of the acrocranium, high and wide forehead, hypoplastic maxilla with pseudo prognathism, hypertelorism, divergent squint, beaked nose and malocclusion of the teeth. Skull, spine and hand radiography is usually needed to confirm the diagnosis. Skull radiograph can show oxycephaly, deep digitate impression, short anterior cranial fossa, and underdeveloped lateral sinuses, while x-ray spin usually reveals lumbarization and the presence of bifid spinous process.[1]

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writing the manuscript. All the co-authors approved the manuscript before submission.

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