**Abstract:** Crouzon syndrome and Apert syndrome share clinical similarities to those depicted in the illustrated case. Diagnostic confirmation typically involves genetic testing, radiological assessment, and thorough physical examination. This image effectively showcases the key clinical trait distinguishing Apert syndrome from Crouzon syndrome- complex syndactyly of the extremities. While these two types of syndromic craniosynostosis share very similar characteristics, a correct diagnosis is vital to address the patient’s distinct needs.

**Keywords:** Apert syndrome, Crouzon syndrome, Craniosynostosis

**Key clinical message:** Apert syndrome, a syndromic craniosynostosis, is typified by craniofacial dysostosis and syndactyly of the extremities. Given its many overlapping clinical features with Crouzon syndrome, a comprehensive understanding is essential for medical practitioners.

**Case presentation:** A 17-year-old female patient presents with congenital craniofacial anomalies characterized by dysmorphic cranial and facial features. These included an enlarged cranial vault, frontal bossing with prominent supraorbital ridges, and apparent pseudo-prognathism. The patient exhibits bilateral proptosis, strabismus, a depressed nasal bridge, widely spaced dentition, a highly arched palate, and low-set ears. While the upper extremities display no discernible anomalies, syndactyly is evident bilaterally in the lower extremities. Radiographic examination via lateral skull X-ray reveals a distinctive "copper beaten" appearance and fusion of the second and third cervical and fourth and fifth cervical vertebrae (Figure 1). The patient was born prematurely at 28 weeks via uncomplicated vaginal delivery to consanguineous parents. Family history is notable for similar phenotypic presentations and a history of seizures in a brother who passed away at 15. Genetic analysis identified a mutation in the fibroblast growth factor receptor 2 gene. The clinical manifestations, radiological imaging, and genetic testing led to the presumptive diagnosis of Apert syndrome.

**Discussion/Conclusion:** Syndromic craniosynostosis, such as Crouzon and Apert syndrome, is often the result of de novo autosomal dominant mutations of the fibroblast growth factor receptors. Skull-base abnormalities and midface hypoplasia are common due to premature fusion of cranial sutures. Crouzon syndrome is predominantly caused by a mutation in fibroblast growth factor 2 (FGFR), although mutations in FGFR3 have also been reported [1]. Apert syndrome’s most common genetic mutation is also fibroblast growth factor 2. Crouzon syndrome specifically demonstrates very similar features to Apert syndrome. The clinical manifestations of Crouzon syndrome include a flattened forehead due to bicoronal synostosis, proptosis, and midface hypoplasia [1]. Apert syndrome can present identically but typically consists of the distinguishing complex syndactyly of the extremities.

It is important to note that various other types of craniosynostosis, such as Pfeiffer syndrome, can also be caused by a mutation in FGFR2. However, a mutation in FGFR1 is more common. This condition is significant for broad thumbs and brachydactyly, which were absent in this patient. Differing genetic mutations can cause other craniosynostosis- Carpenter syndrome has a RAB23/GTPase mutation, Saethre-Chrotzen syndrome has a TWIST gene mutation, and Muenke has a FGFR3 mutation [1].

Apert syndrome has a broad spectrum of clinical features, including the obliteration of coronal and sagittal sutures, acro cranium flattening, a prominent forehead, hypoplastic maxilla with pseudo prognathism, hypertelorism, divergent strabismus, and dental malocclusion [2]. Diagnostic confirmation typically requires radiographic assessments of the skull, spine, and hand. Skull radiography may reveal sclerosis of the suture line, most commonly the coronal sutures, and hypoplasia of the midfacial bones [3]. In contrast, spinal X-rays typically depict cervical fusion, mostly found in C5/C6, which was present in this patient [4].

Pediatric patients can significantly benefit from early intervention services to help with any developmental or intellectual disabilities. Early intervention requires prompt and accurate diagnosis when assessing a patient with possible craniosynostosis syndrome. Understanding the differences between these conditions and correctly identifying them, made more accessible through the patient report photos presented below, can benefit physicians and healthcare professionals when assessing patients.

**Authors' contributions:** Nayab Munib and Qaisar Ali Khan conceptualized the idea, Alondra M. Robles was responsible for image editing and manuscript writing/review, and Naod F. Belay, Raivat Shah, and Muhammad Afzal were responsible for writing the manuscript. All the co-authors approved the manuscript before submission.

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