**Clinical Manifestations of Apert syndrome**

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**Abstract:**

Apert syndrome presents similarly to the one we presented in this image and a genetic study is used for confirmation. Through this image, we pretend to show the typical findings of physical examination, so that if this appears in the outpatient department the diagnosis of Apert syndrome should be assumed.

**Keywords:** Apert syndrome, syndactyly, craniosynostosis, synonychia.

**Abbreviation**; Fibroblast growth factor

**Case presentation:** An eleven-year-old female patient presented to the outpatient department for deformities in her hands, feet, and face. On examination, she had congenital abnormalities including complete fusion of the second, third, and fourth digits of both the upper and lower limbs, the forehead was high and prominent, the eyes were down slanting, the nasal bridge was depressed, the skull sutures were fused, and the palate was high arched. (Figure 1) She also has a four-year-old sister who has similar problem since birth. What is the most likely diagnosis?



**Figure 1: Clinical Diagnosis of Apert syndrome.**

(Panel A & B). Mittens hands and Sock feet (Syndactyly) i.e. the fusion of the digits of hands and feet. (Panel C).Tall, pointed head (acrocephaly), a high prominent forehead, hypertelorism with an extruded brow, protruded eyes (exorbitism)slanting downward, and a depressed nasal bridge. (Panel D). High arched palate and missed teeth.

**Discussion/conclusion:**

Apert syndrome is an autosomal recessive genetic condition caused by a mutation in FGR genes and mainly targets the skull sutures, hands, and feet resulting in various congenital deformities. (1) The image that we present shows the typical presentation of Apert syndromes like Mitten’s hands and Sock feet (syndactyly) i.e. the fusion of the digits of hands and feet (Panel A and B), and a tall pointed head (acrocephaly), a high prominent forehead, hypertelorism with an extruded brow, protruded eyes (exorbitism) slanting downward, a depressed nasal bridge. (Panel C) Intraorally: Dentition tarda, crowding, and severe high-arched palate can be seen (panel D). These clinical signs especially the fusion of fingers and toes distinguish Apert syndrome from other craniofacial disorders. The deformities in Apert syndrome are generally cosmetic but can affect various functions such as hearing, visual abnormalities, swallowing, writing, etc. so a multidisciplinary approach is needed for its management. (2)

**Authors contribution:**

Dr. Qaisar Ali Khan and Dr. Christopher Farkouh prepared, organized, wrote, and edited all aspects of the manuscript. Dr. Uzair Wazir prepared all the figures in the manuscript. Dr. Bikona Ghosh participated in the conception and design of the study. All authors contributed equally to preparing the manuscript and participated in the final approval of the manuscript before its submission.

**Ethical approval:**

Not required

**Consent:**

Written informed consent was obtained from the patient for the publication of these images. A copy of written consent is available for review by the Editor-in-Chief of this journal.

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**Conflict of interest:**

There is no conflict of interest to declare.

**Guarantor**

Qaisar Ali Khan

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