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CASE IMAGE

Clinical manifestations of Apert syndrome

Qaisar Ali Khan[1] | **Christopher Farkouh**[2] | **Muhammad Uzair**[3] | **Bikona Ghosh**[4]

[1]DHQ & Teaching Hospital KDA Kohat, Khyber Pakhtunkhwa, Pakistan

[2]Rush Medical College, Chicago, Illinois, USA

[3]KMU Institute of Medical Sciences Kohat, Khyber Pakhtunkhwa, Pakistan

[4]Dhaka Medical College and Hospital, Dhaka, Bangladesh

Correspondence
Bikona Ghosh Dhaka Medical College and Hospital, Dhaka, Bangladesh.
Email: bikonaghosh.01@gmail.com

Abstract

Apert syndrome presents similarly to the one we presented in this image, and a genetic study is used for confirmation. This image shows 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, craniosynostosis, syndactyly, synonychia

1 | CASE PRESENTATION

An 11-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 4-year-old sister who has similar problem since birth. What is the most likely diagnosis?

2 | 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 (Panels A and B), and a tall, pointed head (acrocephaly), a high prominent forehead, hypertelorism with an extruded brow, protruded eyes (exorbitism) slanting downward, and a depressed nasal bridge.

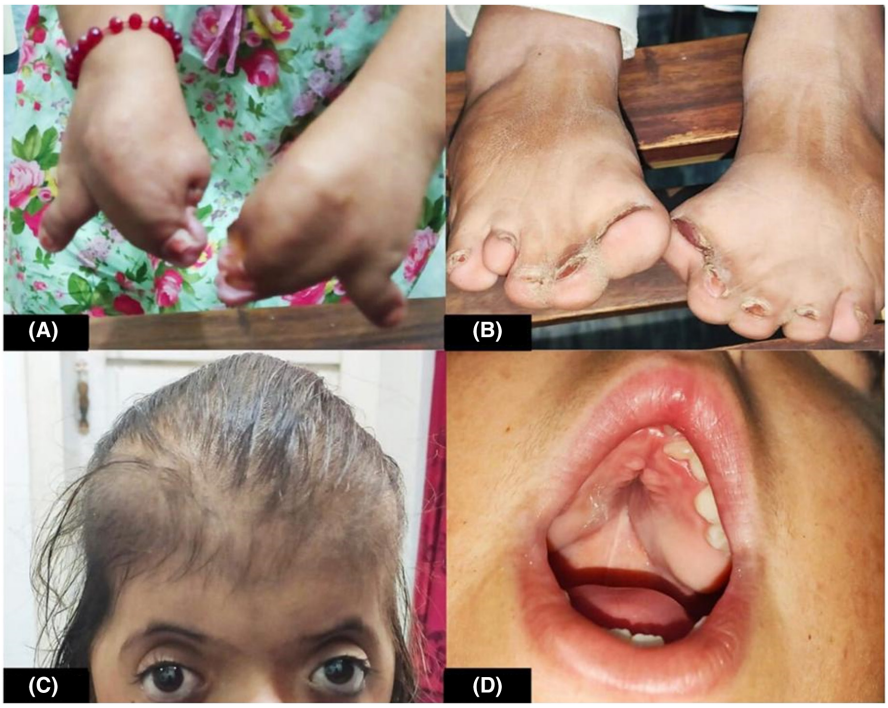

FIGURE 1 Clinical diagnosis of Apert syndrome. (A, B) Mitten's hands and sock feet (syndactyly), i.e., the fusion of the digits of hands and feet. (C) Tall, pointed head (acrocephaly), a high prominent forehead, hypertelorism with an extruded brow, protruded eyes (exorbitism) slanting downward, and a depressed nasal bridge. (D) High-arched palate and missed teeth.

(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]

AUTHOR CONTRIBUTIONS

Christopher Farkouh: Conceptualization; writing – original draft; writing – review and editing. **Mohammad Uzair:** Conceptualization; data curation; writing – review and editing.

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None.

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CONFLICT OF INTEREST STATEMENT

There is no conflict of interest to declare.

DATA AVAILABILITY STATEMENT

Data can be available on reasonable request.

ETHICS STATEMENT

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.

GUARANTOR

Qaisar Ali Khan.

ORCID

Qaisar Ali Khan https://orcid.org/0000-0001-8660-9551
Bikona Ghosh https://orcid.org/0000-0003-0703-9656

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