Apert syndrome
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Section: Paediatric radiology
Case Type: Clinical Cases
Patient: 3 days, female

Clinical History:
A day 3 female child presented with multiple congenital deformities. She had depressed nasal bridge, prominent eyes, complete syndactyly and craniosynostosis.

Imaging Findings:
A day 3 female child presented with multiple congenital deformities. She was a full term normal vaginal delivery with birth weight of 2.2 kg and cried immediately after birth. She had depressed nasal bridge, prominent eyes, and complete syndactyly of all 4 limbs. There was no history of consanguinity in her parents. A CT of the brain on 3D study showed complete fusion of coronal sutures with partial fusion of lambdoid suture with normal brain parenchyma. A radiograph the hand showed syndactyly, the 4th and 5th metacarpals was fused. A radiograph of the feet also revealed syndactyly, polydactyly. 1st, 2nd and 3rd metatarsals were fused.toes are webbed.

Discussion:
Apert syndrome (AS) or acrocephalosyndactyly is a rare autosomal dominant malformation characterized by craniosynostosis, symmetric severe syndactyly, abnormalities of skin, skeleton, brain, and other organs. AS is a rare malformation described by Wheaton in 1894 and later by Apert in 1906. Apert syndrome makes up approximately 4% of all cases of craniosynostosis. The incidence is 1/160,000 live births.

Apert syndrome is usually a spontaneous mutation. The molecular basis is remarkably specific: two adjacent amino acid substitutions (either S252W or P253R) occurring in the linking region between the second and third immunoglobulin domains of the fibroblast growth factor FGR-2 gene. Mutations of the FGFR-2 gene have also been associated with several other craniosynostosis malformation syndromes, including Crouzon, Jackson–Weiss, Pfeiffer, and Beare–Stevenson cutis gyrata syndromes. The common skin manifestations in AS include hyperhidrosis, brittle nails, and synonychia. This combination often leads to candidal infection and colonisation.

Radiology has an important role in the evaluation, management and follows up of these patients. Radiological examination of the skull showed that the vault was large in size, with increased antero-posterior and transverse diameter. The cranial vault deformity is variable but most often presents as a short anteroposterior dimension with craniosynostosis involving the coronal sutures resulting in a turribrachycephalic skull. Three-dimensional CT scans have a role in planning surgery and for objective assessment of operative outcome. Other central nervous system abnormalities include malformations of the corpus callosum, the limbic structures, or both, megalencephaly, gyral abnormalities, encephalocle, pyramidal tract abnormalities, hypoplasia of cerebral white matter and heterotopic gray matter. Magnetic resonance imaging is the preferred investigation to demonstrate the anomalies of the brain. Cervical spine involvement is a fusion, involving articular facets, neural arch, transverse processes, or vertebral
There is a complex syndactyly (bone and soft tissues) with webbed fingers or toes. The upper extremities are shortened. The usual hand abnormality in AS consists of a bone fusion of the second, third and fourth fingers, with a single common nail. There can be a similar deformity involving the foot. Limited mobility at glenohumeral joint and elbow joint, multiple epiphyseal dysplasia, very short or absent neck of scapula and small capitulum and flat radial head were described in AS.

**Differential Diagnosis List:** Apert syndrome

**Final Diagnosis:** Apert syndrome

**References:**


Figure 1

Description: complete fusion of coronal sutures with partial fusion of lambdoid suture Origin:
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Origin:
**Description:** complete fusion of coronal sutures with partial fusion of lambdoid suture

**Origin:**

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The image depicts a medical scan, possibly a CT scan, showing the fusion of cranial sutures. The text provides a description of the fusion patterns observed in the skull.
Figure 2

Description: Big head, mid-facial anomaly and syndactyly

Origin:
**Description:** Syndactyly with webbing of the fingers, the 4th and 5th metacarpals was fused.
Description: Syndactyly, polydactyly. 1st, 2nd and 3rd metatarsals were fused. Toes are webbed.

Origin: