Apert syndrome: A case report with discussion of craniofacial features

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Apert syndrome is a rare congenital anomaly characterized by acrocephaly, syndactyly, and abnormalities of other organs. It has characteristic features in the orofacial region, affecting the eyes, palate, middle third of face, and uvula. In this case report, the features of Apert syndrome, particularly in relation to the orofacial region, are discussed. (Quintessence Int 1999:30:423–426)

Key words: acrocephalosyndactyly, Apert syndrome, craniosynostosis, syndactyly

A pert syndrome, or acrocephalosyndactyly type I, is a rare, congenital anomaly with an incidence of 1 in 160,000 live births.1 This syndrome was described in detail in 1906 by Apert, hence the name. The syndrome had been mentioned as early as 1842 by Baumgartner and also in 1894 by Wheaton.2 In 1960, Blank1 established distinct criteria for the syndrome. Apert syndrome is a variant among the craniosynostoses, characterized by acrocephaly and syndactyly of limbs, minimally involving the second, third, and fourth digits, and often is combined with anomalies of various other organs. The majority of cases of Apert syndrome are sporadic, possibly representing new mutations. Many of the cases reported reveal familial tendencies, and some cases are associated with genetically determined anomalies such as cleft palate and oxycephaly. The syndrome seems to be transmitted as an autosomal-dominant trait.3

CASE REPORT

A 27-year-old man reported to the Department of Oral Medicine with the presenting complaint of missing maxillary anterior teeth. The teeth had been extracted 4 months prior to the visit because of caries. The patient also complained of an inability to brush his teeth because his fingers were malformed. The patient's medical history revealed that his hands had exhibited abnormalities since his birth.

He was born of a nonconsanguineous marriage. No illness or use of medications during pregnancy was reported. The delivery was full term and uneventful. Postnatally, he had experienced no serious illnesses or hospitalization. The developmental milestones were normal, although a slight delay in the eruption of both dentitions was reported. The family history revealed no similar conditions in any of the members.

The patient was well built and well nourished. All vital signs were within normal limits. Extraoral examination revealed a brachycephalic skull with a flat occiput. The patient also exhibited a depressed nasal bridge with septal deviation, hypertelorism, a hypoplastic middle third of face, a slightly prognathic mandible, and large, low-set ears. Proptosis and exotropia were also present (Figs 1 and 2). Syndactyly of all fingers and toes of both hands and feet were prominent features (Figs 3 and 4).

Intraoral examination revealed poor oral hygiene with varying degrees of periodontal involvement. The patient had a high, arched palate with a cleft in the middle of the anterior part of the hard palate (Fig 5). Bifid uvula was a prominent intraoral feature (Fig 6). Severe malocclusion and posterior crossbite were noted. The 6 maxillary anterior teeth and both mandibular first molars were missing.
Fig 1  Hypertelorism and brachycephaly.

Fig 2  Mandibular prognathism, hypoplastic middle third of the face, and large, low-set ears.

Fig 3  Syndactyly of hands.

Fig 4  Syndactyly of feet.

Fig 5  High, arched palate with a cleft.

Fig 6  Bilid uvula.
Roentgenographic examination included panoramic (Fig 7) and posteroanterior (Fig 8) views and hand radiographs (Fig 9). Radiographic findings were consistent with the clinical findings and, based on these, a diagnosis of Apert syndrome was made.

**DISCUSSION**

Apert syndrome is characterized by craniofacial malformations, syndactyly of hands and feet, dysplasia in other skeletal regions, as well as visceral anomalies. Patients exhibit varying characteristics, depending on the severity of involvement.

Craniofacial malformations include hyperacrobrycephalic skull; steep, wide, and flat forehead; and flat occiput. The skull is wide, and it bulges at the temporal region. Asymmetry is common in Apert syndrome and may be related to constant combination of true megalencephaly, premature fusion of coronal sutures, and patency of calvarial sutures, fontanelles, and synchondroses. Cloverleaf skull, or Apert skull appearance, depends on the degree to which the temporal bones are obliquely placed.
Other characteristic features include shallow orbits with ocular proptosis, hypertelorism, downward-slanting palpebral fissures, and interruption of the eyebrows. Exotropia is also a characteristic feature. The V pattern is common, with divergent upward gaze and esotropic downward gaze. Myopia and astigmatism are found frequently. There is an overall tendency for the ears to be large and low set. The nose is short and wide, the nasal bridge is depressed, and the nasolabial angle is diminished.

There is syndactylyism in the patients. The expression can range from partial fusion of the skin to a true osseous syndactyly of fingers and toes. When the fingers are completely fused, there is often a common nail. The hands might resemble that of children affected by thalidomide.

During infancy, the lips of patients with Apert syndrome have a trapezoidal configuration because the upper lip is lifted in the midline. The palate is highly arched and constricted and has median furrow in almost all patients. The hard palate is shorter than normal, and the soft palate is longer and thicker than normal. Nearly 75% of patients have cleft soft palate or bifid uvula. Dental anomalies include delayed and ectopic eruption, shovel-shaped incisors, crowding of teeth, especially in the maxilla, anterior open bite, bilateral crossbite, mandibular overjet, and midline deviation.

In the present case, the patient's craniofacial anomalies included a brachycephalic skull with a flat occiput, hypertelorism, a hypoplastic maxilla, and exotropia. Intraorally, the features were a bifid uvula, a high, arched palate with a cleft, posterior crossbite, lip incompetency, and malocclusion. Complex osseous syndactyly, involving all the fingers and toes, was also found. These features are consistent with those reported by other authors. The poor oral hygiene and periodontal status could be attributed to the patient's inability to maintain oral hygiene because of his fused fingers.

In this patient, a steep forehead and interruption of the eyebrows were absent, but other features correlated with the classic findings of Apert syndrome. The absence of a positive family history suggests a possibility of new mutation in this patient.

REFERENCES