

Apert's Syndrome - A Case Report and Review of Literature

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ABSTRACT

Apert's syndrome makes up approximately 4% of all cases of craniosynostosis. The incidence is reported to be 1/160,000 live births [1]. Radiology has an important role in the evaluation, management and follow up of Apert syndrome patients. Plain radiographs are sufficient for diagnosis but CT has added a new dimension to the evaluation of these disorders. Apert syndrome is a form of acrocephalosyndactyly, a congenital disorder characterized by malformations of the skull, face, hands and feet. It is classified as a branchial arch syndrome, affecting the first branchial (or pharyngeal) arch, the precursor of the maxilla and mandible.

HISTORY

In 1851 Virchow noted that there is a cessation of growth in a direction perpendicular to that of the affected suture while growth proceeds in a parallel direction. In 1894 Wheaton described Apert's syndrome [2]. In 1906, Eugène Apert, a French physician, described nine people sharing similar attributes and characteristics, "acro" is Greek for "peak", referring to the "peaked" head that is

common in the syndrome. "Cephalo", also from Greek, is a combining form meaning "head". "Syndactyly" refers to webbing of fingers and toes. Findings for the incidence of the syndrome in the population have varied, with estimates as low as 1 birth in 200,000 provided and 160,000 given as an average by older studies. A study conducted in 1997, however, by the California Birth Defects Monitoring Program, found an

incidence rate of 1 in 80,645 out of almost 2.5 million live births [3]. Another study conducted in 2002 by the Craniofacial Center, North Texas Hospital For Children, found a higher incidence of about 1 in 65,000 live births [4]

SIGNS AND SYMPTOM

The cranial malformations are the most apparent effects of acrocephalosyndactyly. Craniosynostosis with brachiocephaly being the common pattern of growth, another common characteristic is a high, prominent forehead with a flat posterior skull. A flat or concave face may develop as a result of deficient growth in the mid-facial bones, leading prognathism. Other features of acrocephalosyndactyly may include shallow bony orbits and broadly spaced eyes

CAUSES

Apert syndrome is an autosomal dominant disorder; approximately two-thirds of the cases are due to a C to G mutation at the position 755 in the FGFR2 gene,. The molecular basis of this syndrome appears 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 genes. Males and females are affected equally

CASE REPORT

A 14 month girl presented with abnormal shape of the head and the complaints of symmetric syndactyly of both hands and feet.

On Examination the findings of abnormal contour of the head (turribrachycephaly), , proptosis, exorbitism, high arched palate, mid-face hypoplasia, [Figure - A] symmetric syndactyly of second, third, fourth and fifth digits of the hands and all the toes of the feet [Figure – B&C] were found . A provisional diagnosis of Apert's Syndrome was made and radiological investigations were performed.



Figure A:- Abnormal shape of skull with mid-face hypoplasia & proptosis



Figure B & C:- Syndactyly of second, third, fourth and fifth digits of the hands and all the toes of the feet

Skull radiographs revealed fused coronal sutures, turribrachycephalic skull contour, elongated flat forehead with bitemporal widening [Figure - D]. Spiral CT of the brain was performed which confirmed these findings. Also, the ventricles were mildly dilated and proptosis noted [Figure – E&F]. Radiographs of both hands showed soft tissue syndactyly of second, third, fourth and fifth

digits and synostosis involving phalanges of third and fourth digits with deformed phalanges of first digit [Figure- G]. Radiographs of both feet showed soft tissue syndactyly of all the toes with synostosis involving metatarsals of first, second and third digits. Phalanges of great toe were deformed [Figure – H]



Figure D (Skull AP & LAT):- Turribrachycephalic skull contour, elongated flat forehead

Figure E & F (Axial CT IMAGE):- Mildly dilated ventricles and proptosis



Figure G (HAND AP Images)

Figure H (FOOT AP Images)

DISCUSSION

- Apert's syndrome makes up approximately 4% of all cases of craniosynostosis. In Apert's syndrome, or acrocephalosyndactyly, 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. The typical craniofacial appearance includes a flat, elongated forehead with bitemporal widening and occipital flattening. Premature fusion of sutures with continued brain growth can lead to increased intracranial pressure which can be seen as

increased convolutional markings on skull radiographs. There is also midface hypoplasia accompanied by orbital proptosis, downslanting palpebral fissures and hypertelorism. High arched palate, clefts of the secondary palate and crowding of the dental arch can also be seen. The nose is down turned at the tip, the bridge is depressed and the septum deviated.

- Plain skull radiographs are usually done. Now, 3D- CT scans added a further dimension in planning surgery of these patients and for objective assessment of operative outcome. It is important to evaluate the entire length of each suture as a small segment only may be involved, not seen on plain radiography, resulting in a functional synostosis of the whole suture. This is only effectively done with axial CT images on bone window settings. Several perspectives can be obtained enabling measurement of the skull width in the anterior, mid and posterior thirds as well as the biparietal diameter and interocular distance. The measurements and surface shaded images provide a baseline against which future interval growth and post-operative CT studies can be compared. The dataset also enables construction of three-dimensional models on which complex surgical corrective procedures can be tested [5].
- Other central nervous system abnormalities include malformations of the

corpus callosum, the limbic structures, or both, megalencephaly, gyral abnormalities, encephalocele, pyramidal tract abnormalities, hypoplasia of cerebral white matter and heterotopic gray matter [6]. CT can help in the detection of such abnormalities. There is also an increased incidence of delayed mental development in these children, but many of them develop normal intelligence.

- Cervical spine involvement in the form of variable degrees of fusion has been described, involving the articular facets, the neural arch or transverse processes, or block fusion of the vertebral bodies. In a study by Kreiborg et al, cervical fusions occurred in 68%, single fusion in 37% and multiple fusions in 31%. C5-C6 fusion was most common, alone or in combination with other fusions [7]
- The upper extremities are shortened. The usual hand abnormality in Apert's syndrome consists of a bony fusion of the second, third and fourth fingers, with a single common nail. Involvement of the first or fifth digits in this bony mass is variable. There can be a similar deformity involving the foot (mitten hand and sock foot). Other skeletal abnormalities have also been described in Apert's syndrome. These include limited mobility at glenohumeral joint and elbow joint, multiple epiphyseal dysplasia, very short or absent neck of scapula, small capitulum and flat radial head. [8].

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