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Saethre-Chotzen Syndrome: case report

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Abstract

This study analyses a case of Saethre-Chothzen syndrome (cranio-facial stenosis), which affects the coronal structures and the spheno-basilar synchondrosis in an asymmetric way.
This syndrome has an autosomal dominant pattern of inheritance with a male/female ratio of 1 to 1 and an incidence of about 1 in 25,000-50,000 newborn children.
This article analyses in detail the distinctive features of this syndrome, which affects the cephalic region, the limbs and the psychophysical development.
The case reported is a 10 year-old female patient. Clinical and radiological aspects are carefully described, and an intra-oral analysis is reported with an orthodontic assessment. By analyzing the therapeutic prospects, the authors' conclusion is that the treatment is primarily a surgical one, but the functional and aesthetic aspect of the patients will later have to be taken into account.

1. Introduction

The Saethre-Chotzen Syndrome (S-CS), known also as acrocephalosyndactyly type III is included in the craniofacialstenosis group, of which the most widely known is Apert Syndrome(1).
This syndrome, whose eponym descends from the two European psychiatrists who identified it in the beginning of 1930's, has a rate of 1 in 25,000-50,000 newborns.
In S-CS there is an asymmetric appearance of the two coronal sutures and the sphenobasilar syncondrosis. The importance of these structures for harmonic growth of the neuro and splacnocranium is well known, especially for the different contributions: while the cranial sutures can be considered growth places, and influenced by the surrounding soft tissues and internally from the encephalon, the basicranic synchondroses are the real "growth centers" mostly independent from the functional matrix and influenced by genetic factors (2).
These considerations are essential to understand the complex clinical aspect of this syndrome, especially for the negative influence of a growth defect of the cranial base on the shape and dimension of the maxillofacial structure.

2. General aspects

The Saethre-Chotzen syndrome is described (3), as a congenital hereditary dysmorphosis, with turricefaly (or acrocephalia) with craniofacial asymmetry, ocular, auricular and occlusal anomalies, syndactylia and/or clinodactylism.
The pattern of inheritance is autosomal dominant with high penetration and wide expressive variability (mapped gene: 7p21.2). The frequent mutations are correlated with the father's increasing age. The sexual rate is: 1:1.
Symptoms first appear very early and when not known from the morphologic fetal sonographs, the syndrome is always recognized at the time of birth.
Even though the pathogenic mechanism involves the early fusion of the coronal suture and the sphenobasilar syncondrosis, the reason for this is still unknown.
In the literature there are many pathologic appearances and the clinical overview is not homogeneous. The signs that are always present consist of:

> ***Cephalic area:*** turricefaly with craniofacial asymmetry, plagiocefaly from monolateral sutural defect, (4), low hair insertion, hypertelorism , palpebral rimas with antimongoloid shape, exophthalmous, palpebral ptosis mono or bilateral, myopia, eagle nose with septum deviation, little and malformed ears, (concha

with a prominent pillow starting from the helix root), with low insertion; maxillary retrusion with mandibular prognathism, dental malocclusion (upper lateral incisors sharp or missing).

At the extremities: partial dermal syndactylia between the 2°-3° and sometimes 4° hand finger (dermatoglifi anomalies and monkey furrow in half of the cases); brachydactylia with 4° metacarpus short; clinodactylia of the 5° finger; wide thumb; valgus hallux (sometimes bifid); dermal sindactylia between 2°-3° or 4°-5° foot finger; limited articular excursion in extension movements of elbows and knees.

Psychophysical development:: normal intelligence (sometimes reduced by early craniostenosis). Low stature.

Occasionally reported: strabismus, stenosis and atresia of dacryosyrinx, ogival palate,sometimes fissured, deafness, optical nerve atrophy, cardiac and renal anomalies, cryptorchidism, *(5)*.

2.1 Radiological Aspect

The cranium radiograph, (6), shows rivela microcephaly with acro-brachicephaly; accentuation of the digited impressions; absence or low development of frontal and mastoid sinus; maxillary hypoplasia with relative mandibular prognathism and and temporo-mandibular joint ankylosis.
In the rachid occasionally we can find vertebral anomalies, while it is frequent to find short clavicula with ipoplasic extremity.
The basin presents small iliac wings and big ischium, with valgus coxa.

2.2 Prognosis
The quoad vitam prognosis is good. With regards to esthetics, we frequently see an improvement of facial dismorphism with the growth. Often the case is complicated by ocular infections due to the dacryosyrinx anomalies.

2.3 Therapy
The therapy is limited to the surgical correction of craniostenois, of facial asymmetry, of the palpebral ptosis and of the dacryosyrinx anomalies(7).

3. Case report
The patient was 10 years old.
No former family anomalies. At the eight month ultrasound a malformation syndrome was seen localized on the head. A later amniocentesis shows a normal karyotype 46XX.
Born at full term weighing 1850 g., the baby shows a bilateral clinodattily of the V finger, ipotony, blepharophimosis, craniofacial dismorphism.
The cranium axial tomography done at birth, confirmed a severe syndrome with a wide hypodense liquor content probably connected with ventricular chambers with vertical development in the left fronto-occipito-parietal area. Due to the severe malformation no surgical intervention was indicated.
A later recovery, at the age of 8 months, had shown a 4.150 g. weight, normal blood values, including the serial dosage of cortisol in the blood. The radiological and neurosurgical re-evaluation confirmed the clinical impression of a complex craniosynostosis by the early complete closure of the coronal sutures and the partial closure of the left lambdoid suture, associated with cerebral malformation (holoprosencephalic type).
At the age of one year the weight is 4.5 kg and the length is 57 cm. (5th growth percentile). The magnetic resonance confirmed the holoprosencephalic semilobar case, also for the only posterior cerebral falx. And more: lateral ventricular fusion only anteriorly separated, talams separation and corpo callosum absent. Big dorsal cysts in communication with the left lateral ventricle cause the controlateral shift of the median line.

4. Clinical general and orthodontic aspect

4.1 Morphology and esthetics:

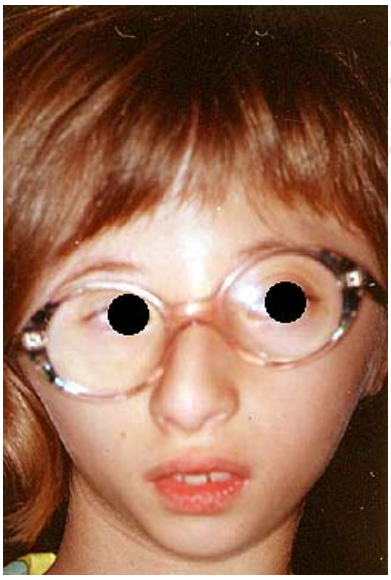

Fig. 1

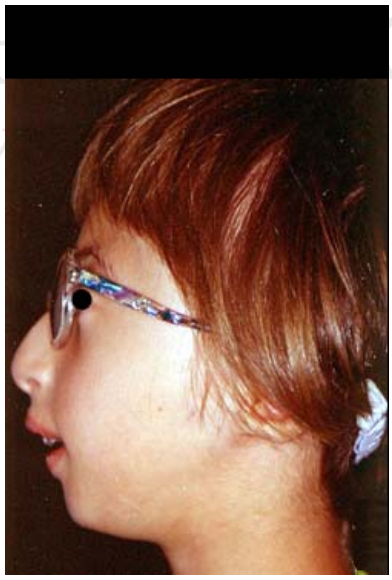

Fig. 2

4.1.1 Frontal Evaluation *(fig. 1 and 2)*:

MicrocephalySevere cranium growth asymmetry with reduced convexity of the profile on the abnormal side, orbit and ears un-leveling (lower on the abnormal side) and nose deviation with convexity forward the abnormal side.Convergent strabismus.Upper bilateral palpebral ptosis, bigger on the abnormal side.Palpebral rima with anti-mongoloid shape.

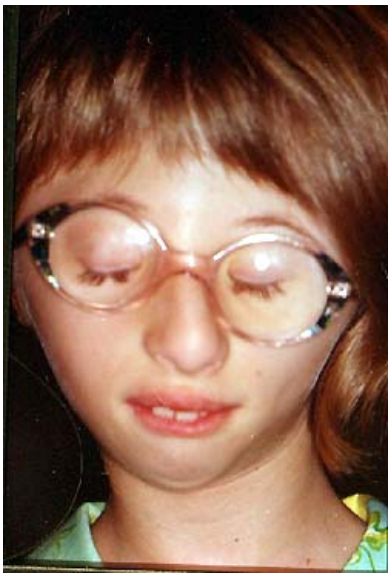

Fig.3

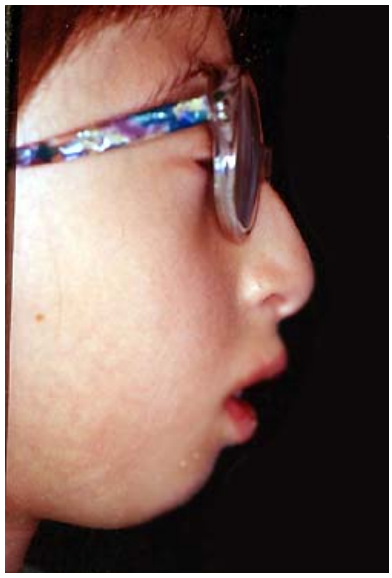

Fig. 4

4.1.2 Profile *(fig. 3 and 4)*:

Convexity due to mandible retrusion.

Parrot nose *(5)*

Labial incompetency

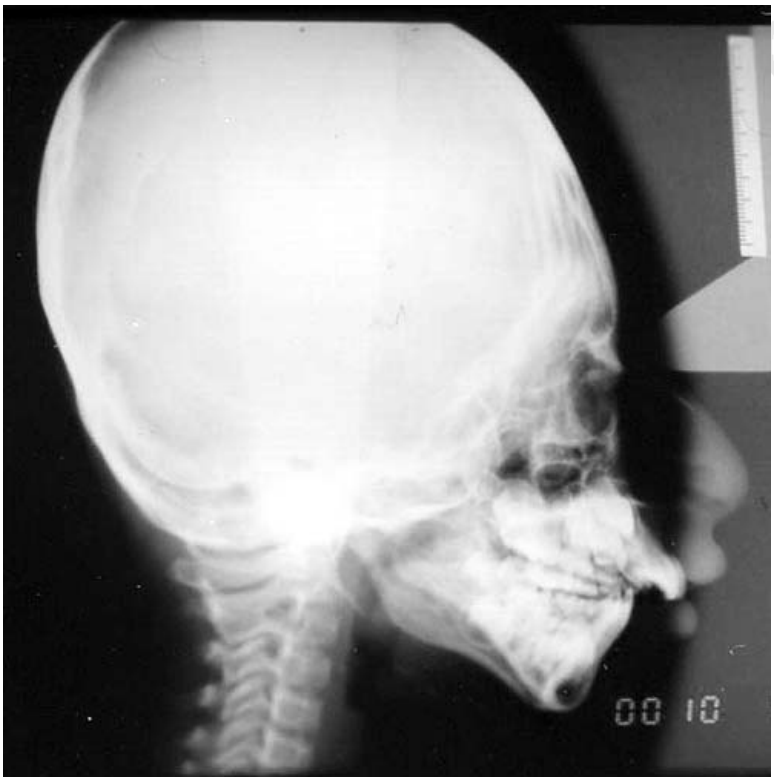

Fig. 5

4.2 Radiology

4.2.1 Lateral view*(fig. 5)*:

Microcephaly

Turricephaly (or acrocephaly)

Sella turcica hypoplasia

Frontal sinus hypoplasia

Orbit reduced deepness

Mandibular retrusion

Secondary un-leveling of mandibular planes

Skeletal Class II relationship indicated by mandibular retrusion

Normodivergency

Maxillary hypoplasia with low sagittal development

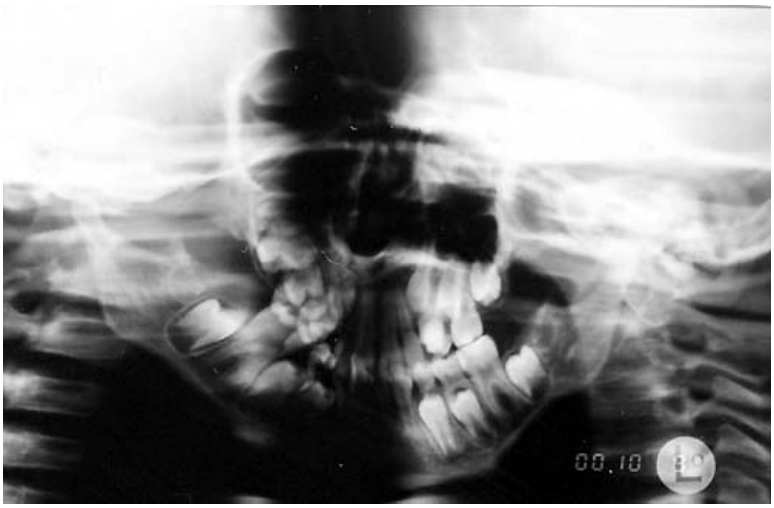

Fig.6

4.2.2 Orthopantogram

(fig. 6):

Nasal septum deviation

Maxillary sinus atresy on the abnormal side

Total mandibular hypoplasia and bilateral coronoid apophysis

Normal dental formula, regular permutation phase.

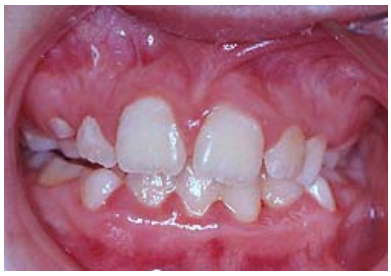

Fig. 7

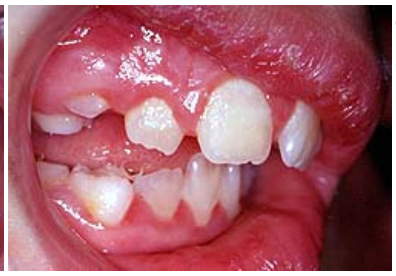

Fig. 8

4.3 Clinical Evaluation

The intraoral examination*(fig. 7, 8, 9)* and model analysis *(fig. 10, 11, 12, 13)* show:

Bimaxillary micrognatism

Normal soft tissues and teeth

Bimaxillary crowding

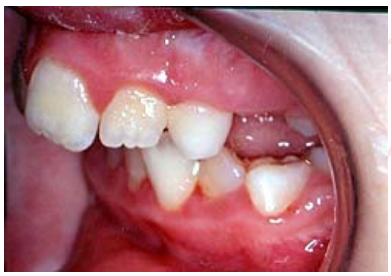

Fig. 9

Fig. 10

Medline deviation

II class, I div. dental relationships.

Relative macrodontia

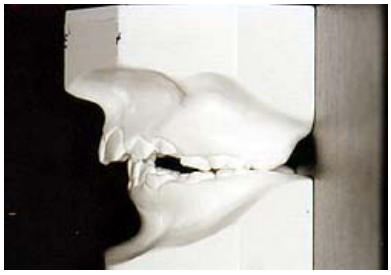

Fig. 11

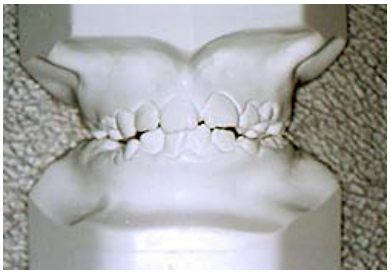

Fig. 12

Fig. 13

5. Differential diagnosis

The differential diagnosis must be done first of all with the other acrocephalosyndactyly (ACS) pathologies, among which the most widely known is the type I, also called Apert syndrome, with the same autosomal dominant transmission. The ACS type I is characterized by a severe and complex syndactylia of hands and feet, with a so called "mitten hands" complex (8).

Congenital central nervous system anomalies must be investigated, as the corpo calloso agenesis.

The other four acrocephalopolysyndactyly (ACPS) types are also known. Polydactylia is always present, and mental underdevelopment (type II or Carpenter syndrome), tibial hypoplasia (type III or Sakati-Nyhan syndrome), cardiac anomalies (type IV or Goodman syndrome). All of them have a recessive autosomal transmission(9).

The Crouzon syndrome (SC), must be also differentiated (recessive autosomal transmission). It has craniostenosis due to early closure of coronal and sagittal suture, by severe maxillary deficency in the infraorbital area due to the fusion of posterior and upper sutures along the roof orbit (7). To this aspect contribute also the basicranic syncondrosis growth anomalies: Hypertelorism, exophthalmos, relative mandibular protrusion, ogival palate and enlargement of the root of the nose. In the SC there is facial symmetry.

As major diagnostic criteria for SC-S, it is important to remember craniosynostosis, syndactylia, low hair insertion on the front, septum deviation and palpebral ptosis.

6. Therapeutical prospects

The treatment indication is mainly surgery with timing imposed by the risk of endocranial hypertension caused by craniostenosis. In a second step functional and esthetics consideration can be considered to correct facial deformities, palpebral ptosis, and dacryosyrinx anomalies, antero-posterior and trasversal relationships of the jaws. The orthodontic treatment is conditioned by the amount of dental crowding. It is complicated by multiple extractions. In the mixed dentition attention should be concentrated on palatal expansion and on the management of Leeway space.

7. Final considerations

The phatogenetic moment of the development of the skeletal malocclusion in SC-S, must be reconduct probably to the influence of the spheno-basilar syncondrosis. The praecox fusion of the cartilages at this level greatly reduces the

translation process of the maxilla. In other words, it is, with the statural growth and the superficial remodeling, very important for the normal growth of the facial complex(10).
No mistake must be made reading the literature about the mandibular prognathism, due to the maxillary deficiency, and the Class II skeletal relationship. The posterior position of the mandible is in our opinion provocate by the need to articulate with the basicranium.
Excluding aligning and leveling in the arches, only surgical-orthodontic treatment can offer correction for the maxillo-facial discrepancy. For this reason it is critical to define a therapeutical priority schedule as early on as possible between several specialists. The esthetic problems must not be underestimated in these patients because of their full ability to have relationships with others.

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