Case report: orofacial characteristics of Hallermann-Streiff Syndrome

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ABSTRACT

Background Hallermann in 1948 and Streiff in 1950 described patients characterised by “bird face”, congenital cataract, mandibular hypoplasia, and dental abnormalities. The new syndrome was later defined as Hallermann-Streiff Syndrome (HSS), underlining the differences with regard to Franceschetti’s mandibulofacial dysostosis. Case Report Examination of a white male affected by Hallermann-Streiff Syndrome revealed facial characteristics typical of the “bird face” in HSS. The nose appeared thin, sharp and hooked; the prominence of the chin was absent in the lateral view; a marked microstomia was evident as well. Radiographic records showed aplasia of teeth 14, 13, 12, 22, 24, 35, 34, 33, 32, 31, 41, 42, and 43. As for orthodontic diagnosis, the following dento-skeletal features were assessed: skeletal Class II malocclusion, narrow upper arch, bilateral posterior crossbite, and anterior open bite. Lateral cephalogram showed hypoplasia of the mandible, a typical sign of HSS. The mandible revealed a clockwise rotation growth pattern with an opening of the gonial angle, skeletal anterior open bite, and an excessive vertical dimension of the lower third of the face. Radiographs at the age of 13 years showed an anomaly in shape of the lower right first bicuspid, which appeared with a double crown. Treatment Orthodontic treatment started at 10 years of age with rapid maxillary expansion in the early stages of the mixed dentition, in order to correct the posterior crossbite due to the narrow maxillary arch. A subsequent phase of the orthodontic therapy consisted of a functional appliance with the goal of maintaining the transverse dimension of the dental arches and of controlling the tongue thrust in the anterior open bite. Surgical and prosthetic interventions were scheduled at completion of growth to solve the skeletal discrepancy and for occlusal rehabilitation. Follow-up The therapeutic protocol used in the patient presented here was able to reach the orthodontic goals during the developmental ages. Further treatment is planned as there are multiple missing permanent teeth and prosthodontic/restorative care is needed until the patient reaches full adulthood.

KEYWORDS: Hallermann-Streiff Syndrome, Face, Dental.

HSS results from a developmental disturbance that arises between the 5th and 7th week of embryonic life and affects the cephalic ventral extremity at the moment when development of facial bones and of lenses is at the highest degree, thus involving both ectoderm and mesoderm [Jones, 1988; Gorlin et al., 1990].

In the past, some authors [Francois, 1958; Carones, 1961; Suzuki et al., 1970] hypothesized that HSS corresponded with other diseases caused by exogenous or endogenous agents, in particular prenatally contracted viral infections. Nowadays, however, a genetic transmission seems to be the most probable hypothesis. A dominant pattern of transmission has not been demonstrated but the sporadic prevalence [Gorlin et al., 1990] does not support a recessive transmission. Most probably this syndrome could be the result of a single mutation.

Introduction

Hallermann in 1948 and Streiff in 1950 described patients characterized by “bird face”, congenital cataract, mandibular hypoplasia, and dental abnormalities. The new syndrome was later defined as Hallermann-Streiff Syndrome (HSS) [1948, 1950], underlining the differences with regard to Franceschetti’s mandibulofacial dysostosis. Polymorphism of clinical signs is typical of HSS. Therefore, even if it is a rare disease, it requires an exact diagnosis in order to evaluate prognosis and implications of a probable genetic transmission [Yuksel et al., 1990; Ioan et al., 1997; Pugliese et al., 1997].

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Case report

Diagnosis. GC, a 6 years old male, was referred to the Department of Orthodontics of the University of Florence, Italy, because of a severe malocclusion. His family history was not significant. The clinical history revealed that he was born after uneventful pregnancy by eutocic delivery at full term. The patient was hospitalized for respiratory troubles immediately after birth and for ocular problems a few months later. Following a diagnosis of congenital cataract associated with hypermetropia, nystagmus, and high intraocular pressure, the patient underwent a surgical intervention.

The dental clinical examination revealed a deficiency in stature and body weight, hypotricosis and cutaneous atrophy, particularly in the nasal region. Psychomotor development appeared slightly retarded, probably because of severely defective sight.

An examination of the head and face revealed characteristics that are typical of the “bird face” in HSS. The nose appeared thin, sharp and hooked; the prominence of the chin was absent in the lateral view; a marked microstomia was evident as well (Figs. 1 and 2). Radiographic records (Fig. 3) showed aplasia of teeth 14, 13, 12, 22, 24, 35, 34, 33, 32, 31, 41, 42, and 43. The orthodontic assessment showed dentoskeletal features as shown in Figure 4. A lateral cephalogram (Fig. 5) showed hypoplasia of the mandible, a typical sign of HSS. The mandible revealed a clockwise rotation growth pattern with an opening of the gonial angle, skeletal anterior open bite, and an excessive vertical dimension of the lower third of the face.

FIG. 1 - Photograph showing frontal view of the patient’s face.

FIG. 2 - Photograph showing lateral view: the nose appears thin, sharp and hooked, and there is absence of the chin prominence.

FIG. 3 - Orthopantomogram at the age of 6 years showing aplasia of teeth 14, 13, 12, 22, 24, 35, 34, 33, 32, 31, 41, 42, and 43.

FIG. 4 - Intraoral photograph at the age of 6 years showing narrow upper arch, bilateral posterior crossbite, and anterior open bite. Teeth 61 and 62 were extracted in order to encourage eruption of 21.
Radiographs taken at the age of 13 years showed an anomaly in shape of the lower right first bicuspid, which appeared with a double crown (Fig. 6).

**Treatment.** Orthodontic treatment started at the age of 10 years by means of rapid maxillary expander in the early stages of the mixed dentition, in order to correct the posterior crossbite. A subsequent phase of orthodontic therapy consisted of a functional appliance with the goal of maintaining the transverse dimension of the dental arches and of controlling the tongue thrust in the anterior open bite. Surgical and prosthetic interventions were scheduled at completion of growth to solve the skeletal discrepancy and for occlusal rehabilitation.

**Discussion**

*Polymorphism of clinical aspects.* François [1958] had already underlined the multiplicity of characteristic signs of HSS by analysing 22 cases and suggested (Table 1) to make a distinction of the various clinical elements in 7 positive signs, always present, and 5 negative signs, certainly absent. The case described here presented with all these signs. Furthermore, the patient exhibited severe dentofacial characteristics that could be helpful in detailing the generic terms of “discephaly and bird face” as indicated by François. These signs can be characterised as follows:

- **craniomaxillar signs** - zygomatic-maxillary hypoplasia, hypoplasia of the nose, which is small, sharpened, and beak-shaped, mandibular hypoplasia, with underdeveloped mandibular body and ramus and excessive width of the gonial angle;
- **ocular signs** - microphthalmos with bilateral total or partial congenital cataract;
- **oro-dental signs** - multiple aplasia of deciduous and permanent teeth and anomalies in tooth shape;
- **systemic signs** - proportioned dwarfism, cutaneous atrophy, slight deficiency in psychomotor development and hypogonadism and cryptorchidism.

**Differential diagnosis.** The typical HSS characteristics that were found in the patient described here allow for differential diagnosis with respect to other syndromes of first branchial arch and ectodermic dysplasiae, especially when associated with a reduced volume of craniofacial structures. In progeria there is a more generalized cutaneous atrophy, and ocular alterations are absent. In oculo-dento-digital dysplasia the face is similar to
HSS, but anomalies of fingers (such as syndactyly and absence of the phalanxes) are present. In mandibulofacial dysostosis the appearance of the face is analogous to HSS, but there are typical ear anomalies. In Seckel syndrome or bird-face dwarfism there are differences in auricular deformities as well.

From the analysis of the literature and the observation of the present case it has to be emphasized that craniofacial and dental signs may play a major role within HSS polymorphism. A detailed appraisal of craniofacial and dentoalveolar characteristics appears to be of valuable assistance in the diagnosis of the syndrome.

As for treatment of the dentoskeletal anomalies associated with HSS, the therapeutic protocol used in the patient presented here was designed to reach the orthodontic goals during the developmental ages. More specifically, the use of rapid maxillary expansion followed by a functional appliance is recommended in those patients with HSS in order to correct the skeletal and dentoalveolar transverse deficiency and to control the tongue thrust. The severity of the skeletal discrepancy in both vertical and sagittal planes, however, requires surgical intervention at the completion of growth.

References