Hallermann-Streiff Syndrome: A Rare Case Report

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ABSTRACT

The Hallermann-Streiff syndrome (HSS) is a rare congenital disorder characterized by distinctive craniomaxillofacial malformations and significant orodental abnormalities. A relative lack of dental literature regarding this syndrome has been noted. Most cases of Hallermann-Streiff syndrome occur randomly for unknown reasons and may be the result of mutations, or changes to the genetic material. A 15-year-old girl presented with almost all the classical signs of HSS except congenital cataract. A thorough medical and dental examination was performed. Opinion by the gynecologist, ophthalmologist and ENT specialist were considered. These patients require an interdisciplinary preventive treatment.

Keywords: Hallermann-Streiff syndrome, Dyscephaly.

INTRODUCTION

Hallermann-Streiff syndrome (HSS) is a rare genetic condition which involves multiple congenital abnormalities chiefly affecting the head and the face. Around 150 cases have been reported in the literature worldwide.1

HSS was first incompletely described by Aubry in 1893. It was then known as Oculomandibulo-Dyscephaly syndrome or Francois Dyscephalic syndrome. Later, Hallermann (1948) and Streiff (1950) acknowledged this entity as separate from progeria and mandibulofacial dysostosis and coined the syndrome as Hallermann-Streiff syndrome.2

In 1958, Francois reviewed 22 published cases and described diagnostic criteria for this syndrome. These include:3
1. Dyscephalia and bird face
2. Dental anomalies
3. Proportionate nanism
4. Hypotrichosis
5. Atrophy of the skin
6. Bilateral microphthalmia

Virtually all cases of the HSS are sporadic and are usually not associated with chromosomal anomalies.3 However, Gerinec et al4 have reported the occurrence of this syndrome in two generations; and Schanzlin et al5 reported a chromosomal defect associated with it. There is no sex predilection. The syndrome has been described as concordant and discordant in monozygotic twins, and an affected female giving birth to two normal children has also been reported.6

CASE REPORT

A 15-year-old female patient reported to the Department of Oral Medicine and Radiology with the chief complaint of missing teeth with no history of any past dental treatment. The pattern of eruption sequence of deciduous and permanent teeth was not known. The patient was born after an uncomplicated full-term pregnancy of a nonconsanguineous marriage with birth weight 1.5 kg. Her mother’s obstetric history revealed no record of systemic disease or drug administration. She is the eldest child with her parents and her only sister shows no specific findings. Her medical history revealed feeding problems during infancy, symptomatic treatment taken for recurrent respiratory tract infections since childhood, and decreased visual acuity for which no treatment was taken. Patient has not achieved menarche till now.

Physical examination of the patient’s face revealed frontal bossing, small and thin face, slanting palpebral fissures, bilateral microphthalmia, beak shaped nose accompanied with microstomia and retrognathia. Skin of the face appeared dry and thin. There was slight bluish tinge visible on sclera and hypotrichosis of the scalp and eyelashes are apparent (Figs 1 and 2). The patient was mentally retarded and had a proportionate nanism. Extremities did not reveal any significant findings.

Intraoral examination revealed partial anodontia. The existing teeth exhibited enamel hypoplasia, grayish yellow discoloration and were irregularly developed with altered morphology of the crowns thereby making their identification...


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There was gingival recession present in all the teeth and was markedly apparent in upper first quadrant. The palate was high arched and V shaped (Fig. 4). After thorough clinical examination patient was subjected to series of investigations. No significant finding was detected in hematological investigation.

Orthopantomography revealed absence of numerous permanent teeth and a retained deciduous tooth. The teeth present showed decreased thickness of enamel and dentin, markedly enlarged pulp chambers and canals, stunted roots; all features suggestive of ghost teeth. There was generalized horizontal bone loss present in both the arches. The mandibular body was small with no antegonial notch. The mandibular rami were hypoplastic. The trabeculae appeared to be dense with decreased marrow spaces giving the appearance of generalized opacification (Fig. 5).

Lateral cephalogram showed calvarial thickening, frontal bossing, midfacial hypoplasia with concave profile. Rickett’s cephalometric analysis showed skeletal and dental class III malocclusion due to maxillary retrognathia (SNA: 70°, SNB: 75°, ANB 5°, a mild overjet (1 mm), openbite (13 mm) and average growth pattern (SN-GoMe: 30°. Upper anteriors were proclined (U1-SN 116°) and lower anteriors retroclined (73°) (Fig. 6).

The radiographs of long bones showed no significant changes.

Based on the clinical findings, the possible differential diagnosis of: mandibulofacial dysostosis, HSS and progeria were formulated.

Patient was referred for ophthalmological, gynecological and ENT opinion for further evaluation. Ophthalmological findings reported were bluish sclera, nystagmus, microphthalmous, disorders of refraction and accommodation, sluggish and irregular pupil, glaucoma and microcornea. Gynecological investigations did not reveal any abnormality other than absence of axillary and pubic hair and non-commencement of menstrual cycle corresponding with the age. ENT findings reported were small nares, glossoptosis, tracheomalacia, narrow airway passage, and abnormal glottis closure.

The case reported here presented all the essential features of the HSS with the exception of congenital cataracts; hence the diagnosis of HSS was made.

Patient was immediately included in an interdisciplinary preventive-care program, subjecting her to detailed oral hygiene instructions, dietary recommendations, counselling of the parents and regular dental visits. Patient is advised oral prophylaxis, extraction of grossly carious and mobile teeth, RCTs in all the existing teeth followed by abutment preparations for overlay dentures.

**DISCUSSION**

Hallermann-Streiff syndrome is a rare genetic condition which causes characteristic facial features, visual abnormalities, tooth problems, short stature, and occasionally mental impairment.7

The etiology of HSS seems to be an asymmetric second arch defect which arises during the fifth or sixth gestational week.8 In literature, although there are only few described cases of HSS with chromosomal abnormalities, most studies revealed normal chromosomal analysis.5 We did not perform chromosomal analysis in our case. The possibility arises that a single sporadic mutant gene is responsible.9

Francois analysed clinical manifestations of 22 cases and gave the diagnostic criteria for HSS. Our case revealed dyscephalia and bird face, dental anomalies, proportionate nanism, hypotrichoisis, atrophy of the skin, bilateral microphthalmia except for congenital cataracts. Additionally our case showed mental retardation (IQ < 61/65) which is reported only in 15% of cases.6 Positive clinical features of HSS present in our patient with their % of occurrence is shown in Table 1.

The Hallermann-Streiff syndrome can be distinguished from the pseudoprogerial Hallermann-Streiff (PHS) syndrome,
Progeria, Wiedemann-Rautenstrauch syndrome, Seckel syndrome, Mandibulofacial Dysostosis, and Mandibuloacral Dysplasia. The PHS syndrome in addition has severe spastic quadriplegia and appearance at birth is normal except for absence of eyebrows and eyelashes. Progeria differs from the HSS because of premature arteriosclerosis, nail dystrophy, acromicria, and chronic deforming arthritis; the eyes are normal. Wiedemann-Rautenstrauch syndrome is a progeroid disorder characterized by large hands and feet as compared to HSS. Seckel syndrome is easily distinguished from the HSS on the basis of prominent eyes without cataracts, malformed ears, normal temporomandibular joints, cutaneous atrophy and absence of hypotrichosis. Mandibulofacial dysostosis differs from HSS as it usually has lower eyelid colobomas and associated ear anomalies. Mandibuloacral dysplasia shows for abbreviated terminal phalanges with acroosteolysis which is absent in HSS.6

Dental anomalies are common and may include natal and neonatal teeth, absence of teeth, premature eruption and persistence of deciduous teeth, supernumerary teeth, malformed teeth, enamel hypoplasia, severe and premature caries. Crowding, malocclusion and open bite occurs frequently due to micrognathia even though the teeth are smaller than normal. Even worse, severe agenesis of permanent teeth and delayed eruption of existing teeth are frequent findings.10-12 Hence, every effort should be made to preserve prematurely erupted deciduous teeth to facilitate future nutritional intake and prevent unfavorable sequelae, until the existence of successional permanent teeth can be confirmed.

There have been multiple case reports concerning the potential difficulties in airway management of HSS patients both intraoperatively; and long term. Upper airway obstruction may result from small nares and glossoptosis secondary to micrognathia and tracheomalacia. These conditions can predispose the patients to obstructive sleep apnea, respiratory insufficiency, pulmonary infection, cor pulmonale and feeding problems in infancy.8,10 Early deaths of the patients with HSS due to respiratory challenges necessitate prompt intervention by an ENT specialist.

Individuals with HSS and their families may also benefit from the information on the condition and recurrence risks for future.
There is no cure reported for HSS. In conclusion, in the symptomatic management of a patient with HSS, one should pay special attention to ophthalmologic, dental and upper respiratory problems. Periodic ophthalmologic and ENT examination should be performed for early diagnosis of eye and respiratory complications. The predisposition to severe dental caries and other dental problems makes it imperative that strong prevention program is performed, as early as possible. As with other genetic syndrome genetic counseling should be performed in all the affected patients.

REFERENCES


7. http://www.healthline.com/galecontent/hallerman-streiff-syndrome/5#ixzz1DLLD344DU


Table 1: Positive clinical features of HSS present in our patient with their percentage of occurrence

<table>
<thead>
<tr>
<th>Feature</th>
<th>Percentage</th>
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<tbody>
<tr>
<td>Low birth weight</td>
<td>36%</td>
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<tr>
<td>Proportionate nanism</td>
<td>45-68%</td>
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<tr>
<td>Hypogenitalism</td>
<td>10-12%</td>
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<tr>
<td>Mental deficiency</td>
<td>15-31%</td>
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<tr>
<td>Microphthalmia</td>
<td>78-83%</td>
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<td>Blue sclera</td>
<td>22-31%</td>
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<tr>
<td>Nystagmus</td>
<td>32-45%</td>
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<tr>
<td>Hypotrichosis</td>
<td>80-82%</td>
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<td>Skin atrophy</td>
<td>68-70%</td>
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<tr>
<td>Corneal abnormalities</td>
<td>9-14%</td>
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<tr>
<td>Down slanting palpebral fissures</td>
<td>12-13%</td>
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<tr>
<td>Glaucoma</td>
<td>7-11%</td>
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<tr>
<td>Dental abnormalities</td>
<td>80-85%</td>
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