

Hallermann Streiff Syndrome-The Oral Manifestations in a Child

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Abstract

Hallermann-Streiff syndrome (HSS) is a rare genetic disorder that is primarily characterized by distinctive malformations of the skull and facial region, sparse hair, eye abnormalities, dental defects, atrophic skin changes and a proportionate short stature. Here we discuss a case of 9 years-old female child who presented with abnormal facial features, dental problems and associated cardiac problems.

Keywords: Hypoplasia; Aplasia; Syndrome; Orodontal; Dysplasia

Introduction

Hallermann-Streiff-François (HSF) syndrome is marked by a characteristic facies with hypoplastic mandible and beaked nose, proportionate short stature, hypotrichosis, microphthalmia with congenital cataract, hypodontia, hypotrichosis, skin atrophy of the face and hypoplasia of the clavicles and ribs [1]. About 15% of cases display intellectual deficit [2]. Neonatal teeth may be present. Upper airway obstruction may result from small nares and glossoptosis (tongue falling backwards) secondary to micrognathia, and these may lead to cor pulmonale [3].

Tracheomalacia is a complication that can lead to chronic respiratory insufficiency, resulting in biventricular cardiac failure and early death. It is a rare clinical entity of unknown etiology that affects growth, cranial development, hair growth, and dental development [4]. It is probably due to a developmental disorder in the 5th-6th gestational week that results in an asymmetric second branchial arch defect [5]. Most cases are sporadic but some have mutations in the GJA1 gene (6q21-q23.2) [6]. Both autosomal dominant and autosomal recessive inheritance have been postulated. Reproductive fitness may be low but rare affected individuals have had affected offspring. Males and females are equally affected. The facies are sometimes described as 'bird-like' with a beaked nose. HSS is characterized by seven essential signs, as described by Francois: Dyscephaly (scaphocephaly or brachycephaly with frontal bossing) and typical facies (micrognathia, condylar aplasia, and thin pointed nose); dental anomalies; proportionate nanism; hypotrichosis; atrophy of the skin localized to the head and nose; bilateral microphthalmia; and congenital cataracts. It is known that the dental abnormalities are present in 50%- 80% of the cases; these abnormalities include malocclusion; open bite; severe caries; enamel hypoplasia; supernumerary and neonatal teeth; hypodontia; premature eruption of primary teeth; agenesis of permanent teeth; and maxillary hypoplasia, with poor development of the paranasal sinuses [7]. Our main objective here is to report a case of HSS in 9 years old female child with physical and oral features consistent with the syndrome as well as detection of mutism in this child which has not been reported earlier in cases of HSS.

Case Report

Nine years old female child visited the outpatient department of pediatric dentistry with complain of multiple carious teeth which were asymptomatic. The patient was pre term (34 weeks) low birth weight (2.2 kg) first child of non-consanguineous parents. The parent also gave a history of child having frequent cough and cold for which symptomatic treatment was being given. The mother reported of the child having cardiac problem (ventricular septal defect) for which



Figure 1: Short stature and thin built.



Figure 2: Thinning of hair.

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Received July 30, 2015; Accepted August 31, 2015; Published September 07, 2015

Citation: Acharya S, Mohanty M, Acharya S (2015) Hallermann Streiff Syndrome-The Oral Manifestations in a Child. J Genet Syndr Gene Ther 6: 268. doi:10.4172/2157-7412.1000268

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surgery had been performed. The mother's obstetrics' history did not reveal any systemic disease or drug intake during pregnancy. Physical examination revealed a height of 95 cm (<3rd percentile, <5 Std deviation) with thin built (30 kg) (Figure 1). There was also presence of thin, brittle hairs on scalp (Figure 2). The patient was not able to speak (mute) but she could hear properly. There was no major abnormality detected on routine hematological investigations and ultrasonography of abdomen. Examination of the face revealed frontal bossing, small and thin face, beaked nose with atrophy of skin near nose (Figure 3,4).

Intra-oral examination showed all deciduous dentition, multiple caries, enamel hypoplasia (Figure 5). The palate was high arched and V shaped (Figure 6). The orthopantomograph revealed multiple carious deciduous teeth, missing permanent teeth, hypoplastic coronoid and condylar processes (Figure 7). The overall findings suggested Hallermen Streiff syndrome and full mouth rehabilitation was planned. Unfortunately the patient had to be admitted to hospital with acute respiratory distress and did not turn up for dental treatment despite multiple reminders.



Figure 3: Beaked nose and skin atrophy.



Figure 4: Frontal bossing.



Figure 5: Multiple caries, hypoplasia and retained teeth.



Figure 6: High arched palate.



Figure 7: Orthopantomograph.

Discussion

The first record of this syndrome is reported by Aubry in 1893 [8]. 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), underlining the differences with regard to Franceschetti's mandibulofacial dysostosis [1]. Variability of clinical signs is typical of HSS. Mostly the cases are sporadic in nature and such inheritance pattern is found. The etiology of the syndrome is unknown but it has been stated that this syndrome results from a developmental disturbance affecting 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.5 HSS affects both the sexes equally. According to the literature this syndrome is characterized by seven classical signs, of which our patient had most: i.e., dyscephalia and bird-like face; dental abnormalities; hypotrichosis; atrophy of skin, especially on nose but there were no ocular abnormalities, short stature. Patients with HSS are at high risk of recurrent respiratory tract infections; obstructive sleep apnea; corpulmonale due to anatomical abnormalities of the upper airway; mandibular hypoplasia; and microstomia. In our case also the patient was reported to have recurrent respiratory infections. Other anomalies reported in such patients are skeletal defects, cardiac defects (ventricular septal defect reported in our patient), hematopoietic abnormalities, and pulmonary anomalies [9]. In addition to dental anomalies, TMJ anomalies have been reported in these patients, including aplasia/hypoplasia of the condyles and

coronoid processes, anterior displacement of the condyles, and anterior disk displacements [10]. The hypoplasia of condyles and coronoid processes also was found to be present in our case. The case reported here was found to be mute who has not earlier been reported in this syndrome although no correlation to this syndrome could be established. Orodonal anomalies reported in the literature (50-85% of the cases) are microstomia, a small and retracted tongue, mandibular hypoplasia, a high arched palate, class II malocclusion, open bite, hypoplasia of deciduous and permanent teeth, absence of teeth, persistence of deciduous teeth, supernumerary teeth, natal teeth, malformed teeth, and severe and premature caries [10,11]. The orodental abnormalities in the present case (confirmed by radiographic examination) were in the form of severely carious hypoplastic teeth, missing permanent teeth, persistent deciduous teeth, bilateral hypoplastic condyles and coronoid processes. The presence of these abnormalities is of great help in differentiating this condition from oculo-dento-osseous dysplasia. The differential diagnoses to be considered are progeria (Hutchinson-Gilford), mandibulofacial dysostosis and cleidocranial dysostosis, pyknodysostosis, Franceschetti mandibulofacial dysostosis, ectodermal aplasia and dysplasia, and oculodontoosseus dysplasia [12]. The differential diagnosis of HSS from progeria and progeroid syndromes, mandibulofacial dysostosis, and pseudoprogeria is as follows. Progeria differs from HSS by having premature atherosclerosis, nail dystrophy, chronic deforming arthritis, acromicria, and normal ocular findings. Mandibulofacial dysostosis usually has ear anomalies and lower eyelid colobomas [13]. Five negative signs were also described by Francois as differential diagnostic criteria for HSS. These include the absence of (I) auricular anomalies, (II) palpebral anomalies, (III) premature arteriosclerosis, arthrosis, deformities of joints, muscular atrophy, (IV) nail and extremity anomalies, and (V) mental retardation [2]. No negative signs were seen in our patient.

Medical treatment in these patients is not necessary after patients reach adulthood, though some ophthalmologic problems may need attention. The dental problems need thorough treatment with an interdisciplinary approach. Early preventive care protocols, detailed oral hygiene instructions, and regular dental visits are essential for patients with this syndrome

Conclusion

Despite being a rare syndrome, this syndrome has to be considered in differential diagnosis of other syndromes. Interdisciplinary approach has to be undertaken for the benefit of the patient. Our case had the classical signs of HSS as well as the striking orodental features. The uniqueness of our report is the presence of mutism in this syndrome which has not been reported earlier in literature.

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