



## Case Report

# Hallermann-Streiff Syndrome in a Neonate: - A Case Report

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### ABSTRACT

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 literature world wide. We report a 24 hour old baby which presented to our hospital with to an abnormal facial appearance and feeding difficulties. Baby had frontal and parietal prominence with a pointed and beak-like nose, micrognathia and neonatal teeth. The prognosis for this condition is not exactly known and therefore, a multidisciplinary approach is essential for management.

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## 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 literature worldwide<sup>1</sup>.

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<sup>2</sup>.

In 1958, Francois reviewed 22 published cases and described diagnostic criteria for this syndrome. These include - Dyscephalia and bird face, Dental anomalies, proportionate nanism, hypotrichosis, atrophy of the skin, bilateral microphthalmia, congenital cataract. Virtually all cases are sporadic and usually not associated with chromosomal anomalies and obvious pattern of inheritance.<sup>3</sup> However, Schanzlin *et al*<sup>4</sup> reported a chromosomal defect associated with it and no predilection to sex.

## Case Report

A 24 hour old male baby presented to our hospital with to an abnormal facial appearance and feeding difficulties. He was a first born child to a non- consanguineous married couple with an antenatal period and no exposure to teratogens. His family history was unremarkable. The baby was born was born at term, cried immediately after birth and weighed 2300g. He was referred to our hospital on the following day for feeding difficulty.

On physical examination, body weight was 2400 grams, length was 48cm, and the head circumference was 33 cm. Physical Examination (figure 1 and figure 4)

showed frontal and parietal prominence, cutaneous atrophy of the forehead and over the nose, which was thin, pointed and beak-like. The baby had micrognathia and microtia with relative macroglossia, neonatal teeth (figure 2) and a high arched cleft palate. He also had microphthalmia and bilateral congenital cataract. He had micropenis, underdeveloped scrotum and bilateral cryptorchidism (figure 3). Abdomen, thorax, neurological, cardiac and ophthalmic examination appeared to be normal.

The baby had severe respiratory distress and was not able to take feeds. He was put on ventilator support for the respiratory difficulty and started on intravenous fluids.

## Discussion

The reported case has the 6 out of the 7 diagnostic criteria required.

Diagnostic Criteria for Hallermann-Streiff Syndrome<sup>3</sup>-

1. Dyscephalia and bird face
2. Dental anomalies
3. Proportionate nanism
4. Hypotrichosis
5. Atrophy of the skin
6. Bilateral Microphthalmia
7. Congenital Cataract

Hallermann-Streiff syndrome is a rare congenital anomaly, primarily characterised by head and face abnormalities. The etiology appears to be unknown with very few cases in literature being associated with chromosomal abnormalities and most having normal chromosomal analysis. We did not do a chromosomal analysis on our case. The possibility of a single sporadic mutant gene cannot be ruled out. The syndrome appears to be related to an asymmetric brachial arch defect arising between the 5th to 7th week of gestation<sup>3</sup>.

The faces of these children have a characteristic bird like appearance with a small pointed thin nose (beak-like) and a receding chin. The head appears to be larger with frontal and parietal prominence giving a parrot like appearance. Dental abnormalities tend to be common and 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<sup>7,8</sup>. Hypotrichosis is common, predominantly cephalic and with scanty eyebrows and eyelashes, as in our case. Cutaneous atrophy is seen where the skin becomes thin and shiny, and is often accompanied by telangiectasia. Cafe-au-lait spots may also be seen<sup>6</sup>.

Ocular findings are congenital cataract and microcornea. Other findings which maybe present include blue sclera, glaucoma, raised IOP, defects in the ciliary body and nystagmus. Proportional dwarfism is seen in most cases, including ours. Delayed bone age, failure of skeletal development, scoliosis and spina bifida are the other features that maybe seen<sup>5</sup>.

Hallermann Streiff syndrome can be distinguished from pseudo progerial Hallermann Streiff syndrome (PHS) Progeria, Wiedemann-Rautenstrauch syndrome, Seckel syndrome, Mandibulofacial dysostosis and Mandibuloacral dysplasia. PHS has the additional features of spastic quadriplegia and appearance at birth is normal save for absence of eyebrows and eyelashes. Progeria is differentiated by the presence of premature arteriosclerosis, nail dystrophy and chronic deforming arthritis. Wiedemann-Rautenstrauch syndrome is a progeria-like disorder with large hands and feet as compared to HSS. Seckel syndrome is distinguished based on features of prominent eyes without cataracts, malformed ears, normal temporomandibular joints, cutaneous atrophy and absence of

hypotrichosis. Mandibular facial dysostosis has lower eyelid colobomas and ear abnormalities. Mandibuloacral dysplasia has abbreviated terminal phalanges and acroosteolysis.

A patient with Hallermann-Streiff Syndrome should have special attention paid to ocular, dental and respiratory complications. Feeding difficulties, respiratory embarrassment, obstructive sleep apnoea and recurrent respiratory tract infections are common in these children. Endotracheal intubation and airway management may pose difficulties in many of these cases. Securing and maintaining a proper airway would be important in the treatment. Repeated respiratory infection would be quite common. CPAP has been found to be effective in those with obstructive sleep apnoea<sup>10</sup>. Regular visual assessment and maintenance is important. Early surgical correction is a must in cases with congenital cataract<sup>11</sup>. Dental assessments and hygiene are of utmost importance in these children with a good oral hygiene regimen and nutritional recommendations. Rhinoplasty, facial contouring plastic surgery and mandibular contouring surgery could be performed for the face.

## Conclusion

The prognosis for this condition is not exactly known. Hence a multidisciplinary approach must be used for these children and parents must be counselled accordingly.

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**Figure 1.** Dyscephalia, bird like face with bilateral Microphthalmia



**Figure 2.** Presence of neonatal teeth



**Figure 3.** Micropenis with underdeveloped scrotum and cryptorchidism



**Figure 4.** Proportionate nanism with hypotrichosis and skin atrophy