



Hallermann-Streiff Syndrome—Confluence of Systems!

CASE REPORT

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ABSTRACT

The purpose was to report a case of Hallermann–Streiff syndrome (HSS) with atypical presentation and present a brief literature review. 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 HSS, underlining the differences with regard to Franceschetti’s mandibulofacial dysostosis. HSS is associated with developmental anomalies involving structures of ectodermal origin (the face, skull, hair, skin, eyes, and teeth) and overall growth and development. Approximately 150 cases have been reported in the literature worldwide.

Keywords: Hallermann–Streiff syndrome, dyscephaly, bird face

INTRODUCTION

Hallermann–Streiff syndrome (HSS) is an uncommon congenital anomaly featuring oculo–mandibulo–cranial malformation with hypotrichosis. HSS was first incompletely described by Aubry in 1893. It was then known as oculomandibulodyscephaly syndrome or Francois dyscephalic syndrome. Later, described by Hallermann in 1948 and Streiff in 1950, the term “bird face” was used because of the peculiar facial appearance (1). Because this anomaly is accompanied by dyscephaly, micrognathia, dental anomalies, microphthalmia, congenital cataract, microstomia, mandibular hypoplasia, high-arched palate, and a large tongue in relation to the oral cavity, there are difficulties in maintaining a patent airway. There is no sex predilection. We present the rare case of a patient who was admitted to our department with birth asphyxia, who was born of a third-degree consanguineous marriage, and who was subsequently diagnosed with HSS because of distinctive clinical features with ventricular septal defects. Approximately 150 cases of HSS have been reported in the literature worldwide (2).

CASE REPORT

A 4-day-old male child born of a third-degree consanguineous marriage with a birth history of G3P3L1A0D2, late preterm, LSCS delivery, weighing 2.26 kg, did not cry immediately after birth came to us with chief complaints of decreased activity and poor feeding since the past 3 days. The baby had episodes of desaturation. Intravenous antibiotics and anticonvulsants were given. He had to be intubated because of resistant seizures and was kept on a ventilator for 5 days. Intravenous antibiotics were stepped up, and the baby was progressively weaned off the ventilator. He had some typical features as dyscephaly, micrognathia, dental anomalies, microphthalmia, congenital cataract, microstomia, high-arched palate, and a large tongue (Figures 1, 2) in relation to the oral cavity, with additional features of ventricular septal defect, micropenis, and undescended testes (Figure 3). On investigations, the hemogram was within normal limits. Serum electrolyte levels were suggestive of hypernatremia. CSF analysis was normal. Thyroid function tests and growth hormone levels were normal. MRI of the brain was suggestive of corpus callosum agenesis with a classical “bat wing” appearance (Figures 4-6). There was no blood test or molecular genetic test that could confirm the diagnosis. Therefore, we diagnosed the baby as having HSS.

Diagnostic Criteria:

In 1958, Francois reviewed 22 published cases and described diagnostic criteria for this syndrome (3). These include the following:

1. Dyscephalia and bird face
2. Dental anomalies
3. Proportionate nanism
4. Hypotrichosis
5. Skin atrophy
6. Bilateral microphthalmia
7. Congenital cataract

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Figure 1. Featuring oculo-mandibulo-cranial malformation



Figure 2. Showing neonatal teeth

Treatment:

Treatment is symptomatic.

Prognosis: Unknown

Differential diagnosis:

Criteria

1. Progeria (Hutchison's Gilford type 2)
2. Cleidocranial dystosis
3. Franceschetti's mandibulofacial dystosis
4. Oculodentodigital dysplasia

DISCUSSION

Hallermann–Streiff syndrome is an uncommon congenital anomaly featuring oculo–mandibulo–cranial malformation. The exact etiology of this syndrome is undetermined, but the following possibilities exist. The most likely possibility is that of a single mutant dominant gene. Most cases have been reported as presenting a new mutation. In almost all cases, HSS randomly occurs because of unknown reasons (sporadically), and this syndrome is thought to be the result of a mutation (4). However, Gerinec (5) reported the occurrence of



Figure 3. Showing micropenis and undescended testis

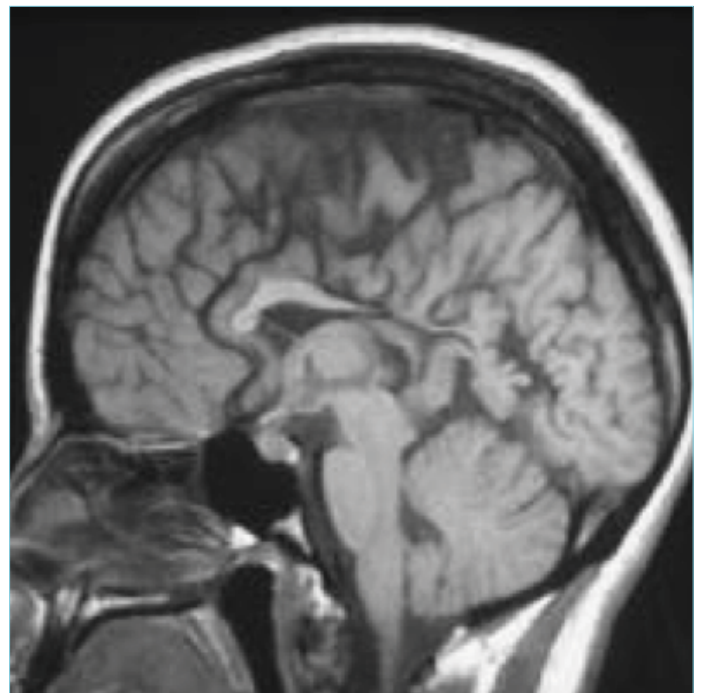


Figure 4. Midsagittal view of MRI brain showing agenesis of corpus callosum

this syndrome in two generations; Schanzlin et al. (6) reported associated chromosomal anomalies of the syndrome. The syndrome has been described as concordant and discordant in monozygotic twins and an affected female giving birth to two normal children (7).

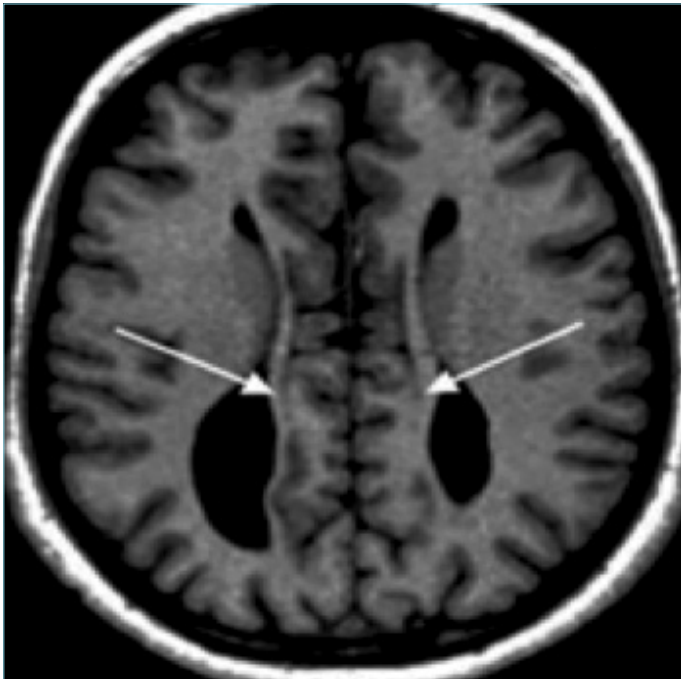


Figure 5. Axial view of MRI brain showing the presence of non decussating fibre bundles of Probst medial to the bodies of lateral ventricles (shown by arrows)

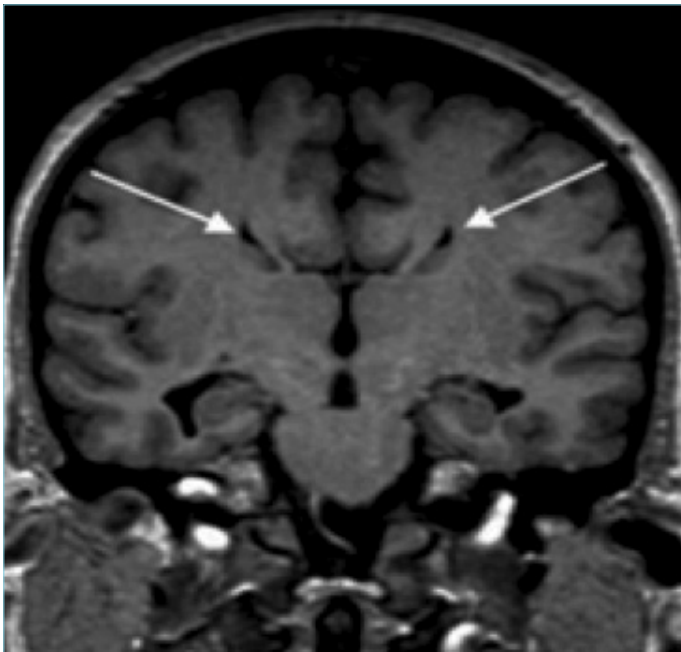


Figure 6. Coronal view Of MRI brain showing classical “bat wings” appearance of lateral ventricles (shown by arrows)

No consanguinity of the parents has been reported, but in our case, third-degree consanguinity was present, which was atypical. However, these facts do not necessarily implicated.

Hallermann–Streiff syndrome is a rare genetic condition causing characteristic facial features, visual abnormalities, teeth problems, short stature, and occasionally, mental impairment (8). The etiology of HSS seems to be an asymmetric second arch defect arising during the fifth or sixth gestational week (9).

There have been many cases reported concerning with the difficulties in airway management intraoperatively and associated with long term morbidities. Upper airway obstruction may result from small nares and glossoptosis secondary to micrognathia and tracheomalacia, which in turn leads to obstructive sleep apnea, respiratory insufficiency, pulmonary infection, cor pulmonale, and feeding problems during infancy (9, 10). Early deaths have been reported due to respiratory challenges.

CONCLUSION

This report helps delineate the specific clinical phenotype of HSS and provides insight into the frequency and spectrum of clinical problems encountered and their outcome. Identifying the cause of HSS is essential to clarify whether this clinical phenotype is a single entity and whether more varied phenotypes are because of the same cause but without characteristic features. With the advent of next-generation sequencing technologies, it is anticipated that the basis of HSS, and other rare disorders of unknown etiology, will become apparent in the near future and lead to an improvement in our understanding of the molecular processes underpinning them.

Informed Consent: Written informed consent was obtained from the parents.

Peer-review: Externally peer-reviewed.

Authors' Contributions: Conceived and designed the experiments or case: PB, SJ, MA. Performed the experiments or case: PB. Analyzed the data: PB, SJ, MA. Wrote the paper: PB, SJ. All authors have read and approved the final manuscript.

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