HALLERMANN-STREIFF SYNDROME: A CASE REPORT

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Abstract
Hallermann-Streiff syndrome is a very rare condition characterized by proportionate dwarfism with characteristic facial appearance such as beaked nose, small mouth, dental abnormalities, bilateral microphthalmia, congenital cataracts, atrophied skin and Hypotrichosis. We present a case of a 15 month old patient who presented with poor vision in both eyes associated with nystagmus, but further examination and investigation confirmed Hallermann-Streiff syndrome

INTRODUCTION.

Hallermann-Streiff syndrome is known under a number of other names such as oculo-mandibulo dyscephaly with hypotrichosis, dyscephaly with congenital cataract and hypotrichosis[1].

This condition was first reported by Audrey in 1893, but by 1948 and 1950, Hallermann and Streiff described the conditions independently[2,3], hence the name Hallermann-Streiff syndrome. There are approximately 60 cases reported in the literature [4] and only a few have been reported by Ophthalmologists. No sex predilection has been reported in the literature[5]. The inheritance pattern of Hallermann-Streiff syndrome is still debated [6], the majority of the cases are sporadic, and autosomal dominant inheritance has been suggested while current data indicates autosomal recessive pattern[7]. The disease is characterized by abnormalities of the skull such as brachycephaly and microcephaly with prominent frontal and parietal prominences, malformation of the face, dental anomalies, localized hypotrichosis, congenital abnormalities of the eyes, dwarfism, motor and mental retardation[8] but the most severe complication is respiratory embarrassment due to narrow air passage with abnormal glottic closure[9]. Hallermann Streiff syndrome has never been reported in this environment and to the best of our knowledge this is the first of its kind.

Case Presentation:

A 15 month old female child was brought to the clinic with complaints of inability to see, and a whitish reflex in both eyes since birth. As the white reflex was increasing in density, the eyes began to oscillate by the age of 3 months. By the age of 6 months the parents noticed an inward movement of both eyes. No medical attention was sought elsewhere before presentation.

The child was the 5th of 5 children. The mother was 29 years old, while the father was 35 years old. Both parents were first cousins. They were healthy so also were the other 4 children. The child is a product of full term, uneventful but not supervised pregnancy. She was delivered at home and said to have cried immediately after birth. Milestones have been delayed because of her condition. and can only sit for now. No records of immunization against all the childhood diseases.

On examination, the child's height was appropriate for her age, mental assessment could not be done due to her age. There was

brachycephaly with bossing of the frontal and parietal bones, the fontanelles were still opened. The hair on her head was scanty, hypopigmented and thin, (see figure 1). The nose was pointed with atrophied skin, beak-shaped, with skin looking white and felt tough. There was malar hypoplasia and micrognathia. The teeth were disfigured and she had high arched palate. No deformities were seen in the external ears. She responded to sound stimulus.

Ocular examination revealed a child that was completely unresponsive to visual stimulus. She had madarosis,. There was obvious nystagmus, microphthalmia and microcornea. Both sclerae were blue in colour. There was convergent squint of about 20 degrees by Hirsberg test. She had bilateral lens opacities. No fundal reflex was seen on fundoscopy.

Skull X-ray showed small orbits, thin scalp bone and hypoplstic mandible (see figure 2).

She was however sent to the Paediatricians for further assessment and fitness for surgery under general anaesthesia. She had bilateral lens matter aspiration and was given aphakic glasses after refraction. She was seen one week after discharge but was lost to subsequent follow up.

Figure 1

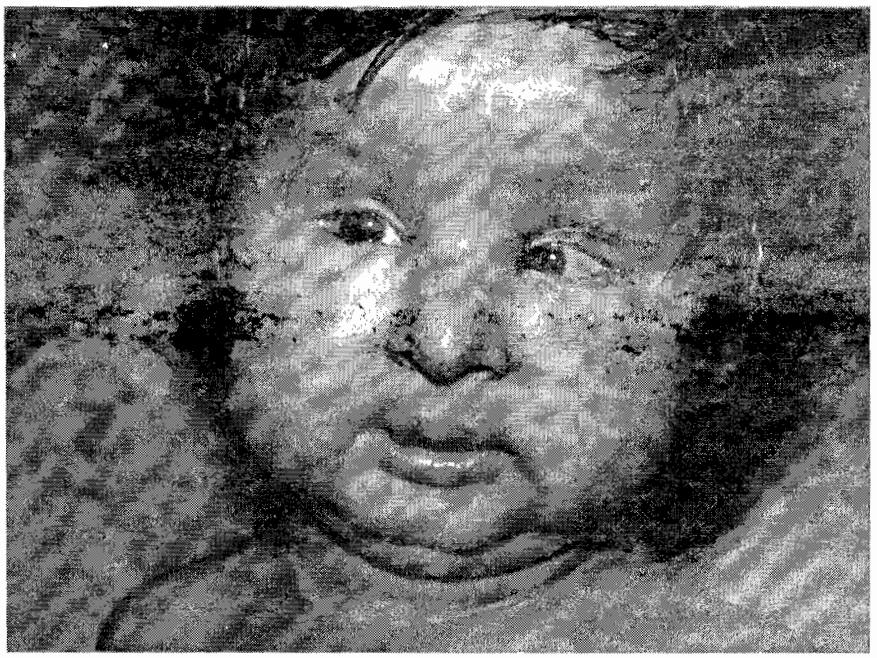
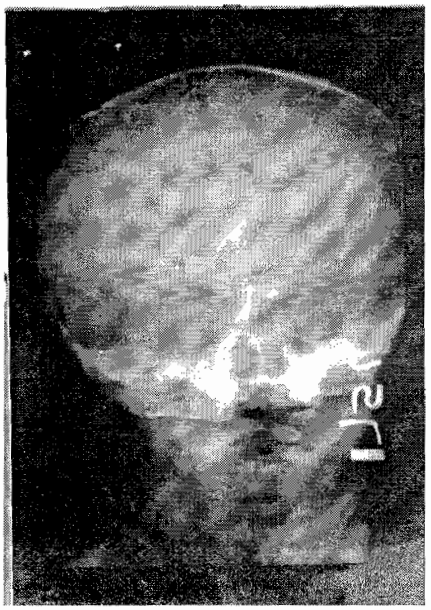


Figure 2



DISCUSSION

Hallermann-Streiff syndrome is a rare disorder that is characterized by systemic, head and face abnormalities in which patients show bird-like facies, dental abnormalities and hypotrichosis with various ophthalmic abnormalities[10,11]. Growth failure occurs in 50% 0f the cases and mental retardation in 15%[2]

Though the majority of cases are said to be sporadic, other workers [12] have shown the inheritance pattern of the disease. The case presented in this study is a product of consanguinous marriage, though the parents and other siblings were healthy and showed no sign of the disease. No chromosomal analysis was done on the parents or the siblings. The main presentation was cataract, which was noticed as a white reflex at birth and continued to increase in density associated with nystagmus. The frequency of cataracts is about 90% in Hallermann-Streiff syndrome with spontaneous resorption of the lens in 8% of the cases[3,13], hence ocular ultrasonography should be done on all cases of Hallermann-Streiff syndrome to prevent unnecessary anaesthesia and surgery[13].

This patient had mandibular hypoplasia and micrognathia, but induction was not difficult for the anaesthetist. Other workers[14] had a different experience in their study and recommended that the anaesthetist should be alerted. Another major problem faced with mandibular hypoplasia and micrognathia is feeding and respiratory difficulties in early infancy[12].

Hallermann-Streiff syndrome has not been reported by many Ophthalmologists despite the ocular signs associated with it. Ophthalmologists should be aware of this condition especially when they are presented with children with bilateral cataracts.

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