Hallermann-Streiff Syndrome Without Cataract: Case Report From Kosova

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Hallermann-Streiff syndrome is a rare congenital disorder characterized as a complex association of developmental anomalies involving structures of ectodermal origin (the face, the skull, the hair, the skin, the eyes, the teeth) and overall growth and development. Ophthalmic abnormalities included bilateral congenital cataract. Glaucoma is uncommon. We report a case with Hallermann-Streiff syndrome having all the main features of the syndrome, however associated with juvenile glaucoma and without congenital cataract. An 16 year-old-boy was admitted in our hospital for surgical treatment of juvenile glaucoma. His ophthalmic features included microphthalmos, enophthalmos, short upper lid, and thin conjunctiva, edematous corneas with fine epithelial bulla, atrophic-colobomatous iris in the left eye, slightly dilated pupils and clear lenses. The right optic disc was pale and cupped (0.9-1). The left optic disc was pale. The IOP was 70 mmHg in the right eye and 62 mmHg in the left eye. There was no light perception on the right eye and 1/60 in the left improving to 0.1 with +7.0 Dsph. Antiglaucomatous therapy failed to reduce the pressure and a left trabeculectomy was carried out. Protective eye shields during sleep were recommended to protect the corneas from dryness because of the short upper lids. In the left eye the IOP has fallen to within normal limits and the vision has improved 0.3-0.4 with +7.0 Dsph. The medical therapy was continued in the right eye but the pressure still remains high (40 mmHg).

Key words: Hallermann-Streiff Syndrome, juvenile glaucoma, no cataract

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1. INTRODUCTION

Hallermann-Streiff Syndrome (HSS) is a rare genetic disorder first described in 1948 by Hallermann, and then in 1950 by Streiff. The syndrome was characterized primarily by head and face abnormalities presented in approximately 150 case reports in world literature (1, 2, 3). Diagnostic criteria for this syndrome described Francois in 1958 (4. 5. 6). As a result of many life-threatening complications, such as respiratory and cardiac difficulties, many patients die in infancy (2, 4, 6, 7).

We report a rare case of this syndrome, a 16-year-old schoolboy with juvenile glaucoma, having all the main features of the syndrome but without congenital cataracts.

2. CASE REPORT

A 16 year old schoolboy was admitted at the Clinic of Ophthalmology at the University Clinical Centre of Kosova because of treatment of juvenile glaucoma. He was born after an uncomplicated full-term, normal pregnancy, by normal spontaneous delivery. His parents were not consanguineous and were healthy. His aunt (father’s sister) is blind as a result of microphthalmos, microcornea and glaucoma. He had two brothers who died at the age of two and three months due to multiple anomalies. He has two other brothers and a sister who have no specific findings.

Physical examination: Revealed he is of short stature and slender physique (Figure 1). His head was small and fore-frontal bossing and scantly hair at the temples. His face was small with an undeveloped jaw, a small mouth, no teeth and a prominent and pointed parrot-beak nose (Figures 2 and 3). He had clawed hands and an S-shaped deformity of clavicles (Figures 4 and 5). The patient was cooperative and intelligent.

Ophthalmic examination: The IOP was 70 mmHg in the right eye and 62 mmHg in the left eye (Goldmann application tonometer). There was no light perception on the right eye and 1/60 in the left improving to 0.1 with +7.0 Dsph. There was microphthalmos, enophthalmos, the upper lid was shorter and the conjunctiva was thin (Figure 6). The corneas were edematous with fine epithelial bulla. The anterior chambers were edematous with fine epithelial bulla. The anterior chambers were edematous with fine epithelial bulla.
The syndrome is a genetic condition, but specific gene responsible has not yet been identified (1). It affects both sexes equally with high frequency of parental consanguinity (8, 9). The condition is sporadic due to a gene mutation during the early months of pregnancy (10). The inheritance pattern of HSS is still debated and the etiology still questioned. An autosomal dominant (AD) inheritance with variable expression or a new mutation has been mentioned (1, 7) but some reports have suggested the possibility of autosomal recessive inheritance (7, 8, 11, 12). In view of the present case, positive family history, our case could be presumed to be due to autosomal recessive inheritance. Potential causes of this syndrome include an asymmetric second bronchial arch defect that arises during the fifth or sixth gestational week (3, 7) maternal viral infections, toxin exposure and paternal age (2, 7).

The syndrome is characterized as a complex association of developmental anomalies involving structures of ectodermal origin (the face, the scull, the hair, the skin, the eyes, the teeth) and overall growth and development. The features are: a bird like face with a parrot-beaked nose (13, 14, 15, 16). The main features of the syndrome are dyscephaly with mandibulofacial malformation, bilateral congenital cataracts and dental anomalies (17). Frequently present are: hipotrichosis, cutaneous atrophy, microphthalmus and proprionate dwarfism. Inconstant ocular features are numerous-iris atrophy, blue sclera, malformations of drainage angles, peripheral anterior synechiae, posterior synechiae, pale optic discs and glaucoma. In addition, this syndrome also has many life-threatening manifestations, such as respiratory difficulties and cardiac defects (2, 7).

Fortunately, our patient did not have suffered from these life-threatening difficulties. In the presented case there were no cataracts, but there were some inconstant ocular features like a short upper lid, enopthalmos, thin conjunctiva and glaucoma. Only a small minority of patients reported in literature were found to have glaucoma (8, 17). In most patients glaucoma usually occurs after the lens has been needled or undergone spontaneous rupture. The glaucoma is caused by complications of intraocular inflammations as a granulomatous anterior uveitis. Hopkins and Horan (17) reported two cases with a classical manifestation of the HSS and secondary glaucoma after cataract surgery. They presented a short review of 5 cases reported in literature which had glaucoma as a complication of intraocular inflammation after cataract surgery. In our case there were no cataracts or evidence of intraocular inflammation. There were no recognisable abnormalities on gonioscopy, but there were some iris defects on the left eye. We supposed that, in our case, undetectable abnormalities of the drainage angle were the cause of the juvenile glaucoma onset. The onset of glaucoma in patients with the HSS presents a most difficult problem. We hope that the successful trabeculectomy carried out on the left eye is long lasting and will avert blindness. Additionally, there was some dryness of the cornea on both eyes due to the short upper lids failing to cover the eye during sleep. The patient was advised to wear protective shields over the eyes when sleeping. The parents reported that since wearing the shields the patient had, to their relief, switched to sleeping on his back rather than on...
his front, which he had inadvertently been doing to protect the eyes.

In conclusion, HSS is a rare disorder whose prognosis is not optimistic. It is important for ophthalmologists to distinguish the features of HSS and to treat these patients promptly to save lives and improve the quality of life.

REFERENCES: