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CASE REPORT

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 scull, 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 trabeculecthomy 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 forehead was wide with frontal bossing and scanty 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.

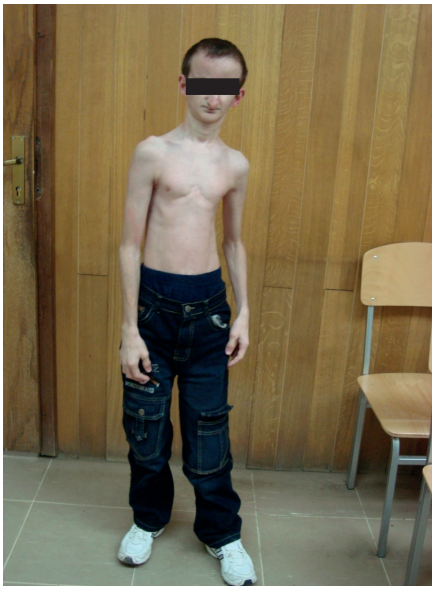

Figure 1. The stature is short and slender. The clavicles are S – shaped

Ophthalmic examination: The IOP was 70 mmHg in the right eye and 62 mmHg in the left eye (Goldmann applanation 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

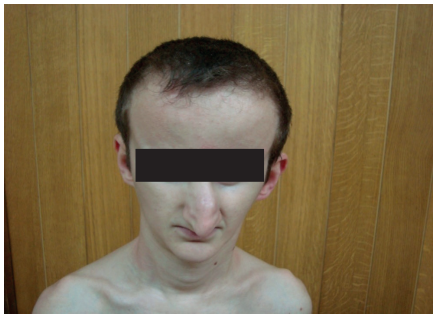

Figure 2. The head is small with a wide forehead and frontal bossing. The nose is prominent and parrot-beaked. The jaw is undeveloped. The hair is scanty at the temples; the skin is thin and atrophic.

were of normal depth with no signs of inflammation. The gonioscopic examination revealed that the drainage angles were opened. The left iris was atrophic-colobomatous (Fig.7). The pupils were slightly dilated. A red reflex was present. The lenses were clear. The right optic disc was pale and cupped (0.9-1). The left optic disc was pale.

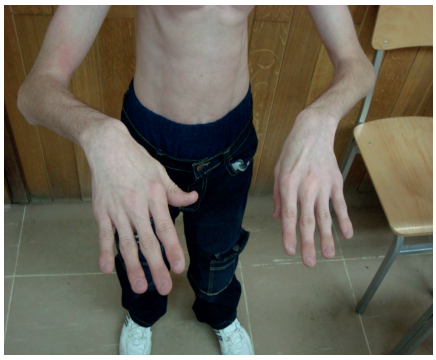

Figure 3.The hands are clawed.

Treatment and results: Anti glaucomatous 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 +7.0Dsph. The medical therapy was continued in the right eye but the pressure still remains high (40 mmHg).

3. DISCUSSION

The Hallermann-Streiff Syndrome is a rare disorder, synonyms: Francois dyscephaly syndrome, Hallermmann-Streiff-Francois syndrome, Oculomandibulodyscephaly with hipotrychosis, Oculomandibulofacial syndrome, Hallermmann' syndrome, Francois' syndrome. 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 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).

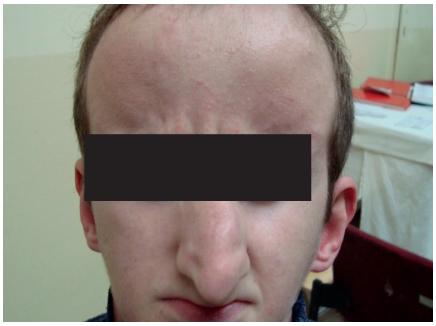

Figure 4.The upper lids are short and don't cover the eye even when shut

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 hypoplastic mandible and a prominent 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).

Figure 5. The left iris is coloromatous.

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, enophthalmos, 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 cataracts surgery. They presented a short review of 5 cases reported in literature which had glaucoma as a complication of intraocular inflammation after cataracts 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.

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BOOK REVIEW

SCIENCE EDITING IN BIOMEDICINE AND HUMANITIES

Izet Masic and Enes Kujundzic. Sarajevo: Avicena, 2013. 272 pages; ISBN 978-9958-720-49-9

The book presents an original effort to summarize the role of academic periodicals and relevant media for communication of scientific knowledge, ideas and new scientific discoveries, various systems of the knowledge classification and categorization, methods for preparation of the papers with scientific and professional aspirations, including insight into the different types of information resources and instruments on access to sources of information in scientific communication.

The book is conceptualized upon contemporary knowledge, methods and methodological approaches toward bases of knowledge and new technologies. The book presents a common ground of an interdisciplinary matter in the research in medicine laboratory, clinical and public health fields, integrating two important areas of scientific inquiry–Biomedicine and Humanities by such key words as Bioethics, Bioinformatics, Social Justice and Human Solidarity. All those various types of research could be of benefit to the society and the welfare of the individual in the community by gathering of evidence to improve clinical and public health practices and policies, identification of health problems and/or methods to promote health and prevent disease and disability. The expansion of scientific literature lays the foundation for the future scientific research, health policy and public health practice making the material accessible to all those who are pursuing or planning to carry out research in medicine, biologic and social sciences. Personal well-being, in general, relate to adoption of new knowledge in society, community for creating healthy environment development and improvement of new skills and leading healthy life styles.

The book is fulfilling an important gap and dedicated towards improving the rank and relevance of domestic academic periodicals thus hopefully contributing to the general scientific competitiveness, first of all, in Bosnia and Herzegovina, as well as in Southeastern Europe and broader.

The publication is a result of a rich scientific knowledge of authors and more than 30-year teaching and research experience in social medicine and public health, community and family medicine at the University in Sarajevo Faculty of Medicine.

The 272 pages are distributed in 17 chapters with references and consulted literature added to each chapter, detailed contents and useful explanation of terms and abbreviations in publishing.

The book is directed toward a certain group of professionals in biomedicine and humanities: for those who intend to carry out scientific research, especially the young doctors and students on specialization, preparing graduate papers, master thesis and doctoral dissertation. The book can be used by students at all levels, from undergraduate to postgraduate master and doctoral studies, and professionals in various clinical disciplines and public health who tend to writing research proposal, selecting a proper research strategy, conducting the research itself and submitting a final report. The book can also be useful as a guideline for all medical and other professionals in biomedicine and humanities in conducting and promoting of their professional and research work.

Prof. Dr. Doncho Donev