

## Hallermann-Streiff syndrome A case report from Egypt

Fatma A. Atwa\*, Mona M.A. Farag\*, Mona N. Mansour\* & Abeer F. Albadry\*

\*Ophthalmology department, Faculty of Medicine (For Girls), Al Azhar University

### Abstract

Hallermann-Streiff syndrome (HSS) is a rare genetic disorder that is primarily characterized by distinctive malformations of the skull and facial (craniofacial) region; sparse hair (hypotrichosis); eye abnormalities; dental defects; degenerative skin changes (atrophy), particularly in the scalp and nasal regions; and proportionate short stature. Here we describe a case with HSS.

**Keywords;** Hallermann-Streiff syndrome, congenital cataract, Microphthalmos.

### Introduction

Hallermann-Streiff syndrome (HSS) (also known as oculo-mandibulo-facial syndrome) is a rare congenital disorder characterized by dyscephaly, dental anomalies, proportionate nanism, hypotrichosis, cutaneous atrophy limited to the head, bilateral congenital cataracts and bilateral microphthalmia. <sup>[1]</sup>

Various ocular findings and fundus anomalies have been reported, including vitreous degeneration, retinal folds, coloboma, and Coats' disease. <sup>[2]</sup>

Cataract and microphthalmos are the most apparent ocular features, but retinal abnormalities may be the primary cause of poor vision. <sup>[3]</sup>

### Case report

1.5 years-old male infant was referred to the Ophthalmology Department of Faculty of Medicine, Al Azhar University because of congenital cataract.

He was the 3<sup>rd</sup> child of non consanguineous parents with old paternal age. His parents were healthy. He had one brother & one sister both of them was totally healthy. He was born by normal vaginal delivery after uneventful pregnancy. His family history was irrelevant. On genetic counseling there was no detectable anomaly.

Following the initial diagnosis of congenital cataract, the patient underwent a surgical intervention in one eye (the Left) at 6 months of age.

The general examination revealed the typical features of the Hallermann-Streiff syndrome (six out of the seven diagnostic criteria), including dyscephaly (an unusually

prominent forehead and sides of the skull) with parrot-beak nose (a thin, pinched, tapering nose) and micrognathia (a small, underdeveloped lower jaw; hypoplastic mandible), with receding chin (retrognathia), sparse hair of the scalp, eyelashes & eyebrows (hypotrichosis), skin atrophy and proportionate dwarfism {Fig. 1}. He has normal intelligence, with no neurologic abnormalities.

Ophthalmic examination revealed right congenital cataract & microphthalmos, nystagmus, strabismus, down slanting palpebral fissures & madarosis.

The measured visual acuity (Lea Gratings: 253300 depending on preferential looking) was 0.25(Rt. Eye) & 0.50(Lt. Eye) {Fig. 2}. The used distance was approximately 57cm (at this distance 1cm equals 1 degree of visual angle). On plotting the measured visual acuity of this infant against his age on the curve, it was below normal.

The average corneal radius of curvature was 5.92mm (right) and 5.74mm (left); extreme micro-corneas.

A-scan Biometry (Mentor-Advent™ Digital B 2000) showed bilateral extreme microphthalmia (axial length, 8 mm OU) {Fig.3}, with the use of the contact technique where the probe was placed on the center of the cornea with caution not to compress the cornea till 5 readings were obtained. The provided measurements in the automatic mode reflect the axial length from the corneal gate backwards to the termination of the orbit instead of the retina (17.54mm OD & 18.06 mm OS instead of 8mm OU) reflecting extreme microphthalmos that was

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out of proportion of the instrument calculation mode.

B-scan Ultrasonography (axial view) revealed acoustic evidences of remnant of lenses (patient surgery could explain this sign in the left but in the right it could be a sign of lens matter absorption). Both vitreous bodies are echolucent & there was evidence of optic disc coloboma in both eyes with remnant of glial tissue (Bergmeister's papillae) {Fig.4}.

**Flash visual evoked potential (VEP)** of both eyes showed significantly reduced wave amplitudes of N1,P1,N2 &P2, with great implicit time delays indicating markedly impaired retino-cortical transmission, more evident in the right eye. **Pattern VEP** of both eyes showed severely reduced, but not extinguished responses, indicating an organic basis along the visual pathway –from the optic nerve to the cortex- for the visual impairment in both eyes. The right eye is relatively more affected {Fig.5}.



**Fig.1.Physical features of the syndrome**



**Fig. 2.Estimation of Visual Acuity**

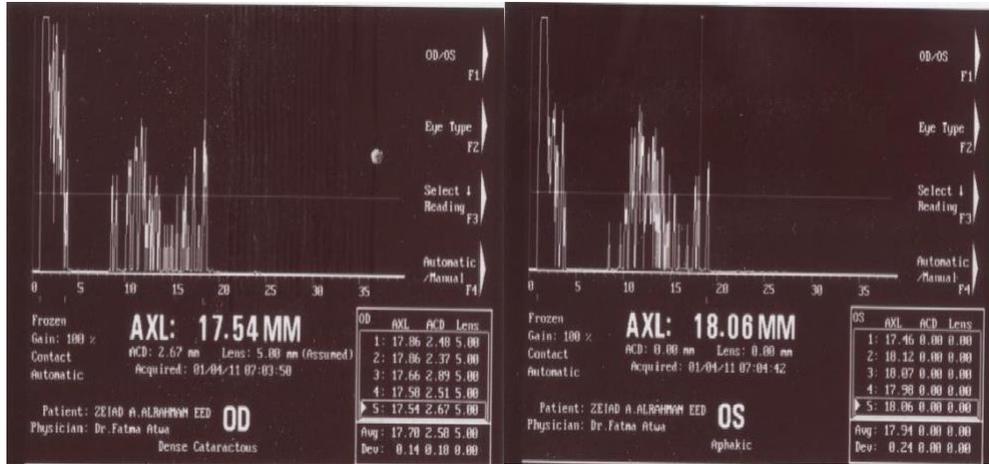


Fig.3.A scan Biometry

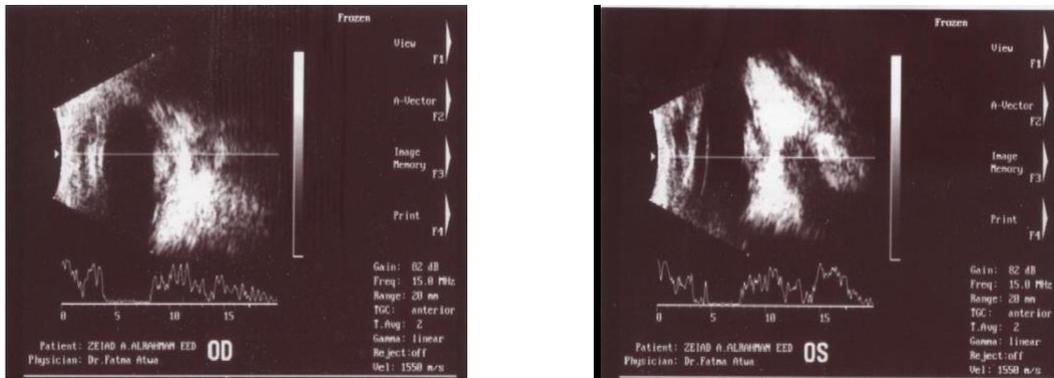


Fig.4. B scan Ultrasonography (Axial view) findings

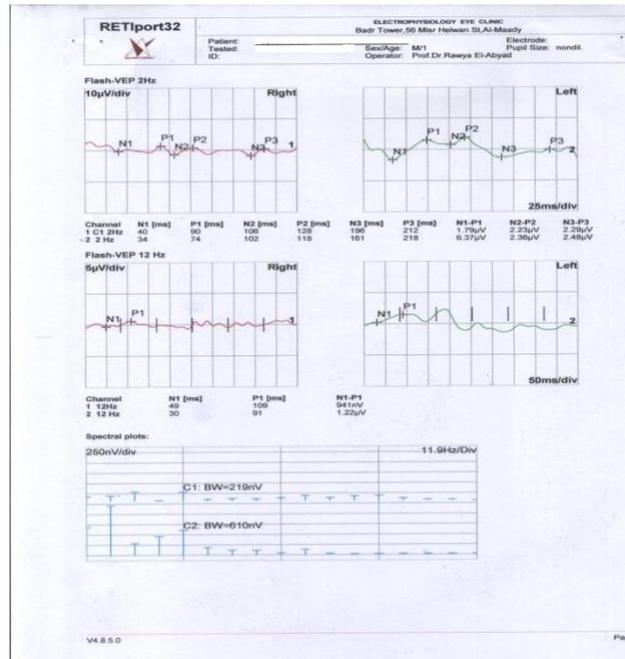


Fig.5.VEP findings

## Discussion

The Hallermann - Streiff syndrome (HSS) variously known as sutural congenital alopecia, Francois syndrome, and oculo-mandibulo dyscephaly, is a rare condition. <sup>[4]</sup>

HSS is characterized by seven main signs: dyscephaly with bird-face and hypoplasia of the mandible, proportioned dwarfism, dental anomalies (absent, malformed or irregularly set teeth), hypotrichosis (particularly obvious in the region of the scalp, the eyelashes and eyebrows), cutaneous atrophy (usually affecting the skin of the scalp and face and especially that of the nose), bilateral microphthalmia, and spontaneous resorption of the congenital cataract. <sup>[5]</sup>

The five signs which distinguish this condition from other related mandibulo-facial dysostoses and ectodermal aplasia and dysplasia are absence of anomalies of ears, nails and extremities, musculo-arthritis anomalies and neuropsychological deficit. <sup>[4]</sup>

Most cases of HSS are sporadic. Autosomal dominant inheritance has been suggested in some cases. <sup>[3]</sup>

HSS results from a developmental disturbance that arises between the 5th and 7th week of embryonic life and affects the cephalic ventral extremity at the moment when development of facial bones and of lenses is at the highest degree, thus involving both ectoderm and mesoderm. <sup>[6]</sup>

Numerous inconstant ocular features have been reported, including blue sclera, anti-mongoloid slanting of the palpebral fissures, keratoglobus, iris atrophy, peripheral anterior synechiae, posterior synechiae, persistent pupillary membrane, amorphous retro-lenticular membrane, vitreous opacities, pale optic discs, optic nerve coloboma, chorioretinal pigmentary changes, retinal folds, and glaucoma. <sup>[5]</sup>

Diagnosis of HSS is based on clinical findings. Ocular abnormalities are a major problem, with the most common ocular features being microphthalmia and cataracts. <sup>[7]</sup>

Our patient had the clinical and ophthalmic features typical of Hallermann-Streiff syndrome. In addition to the systemic clinical features of HSS, there were sparse eyelashes and eyebrows,

bilateral microcornea, bilateral congenital cataract (was operated for congenital cataract in one eye with membranous cataract or partially absorbed lens with capsular remnant in the other) and bilateral extreme microphthalmia with bilateral optic disc coloboma. VEP was performed revealing poor retino-cortical conduction more in the right non operated eye.

Differential diagnoses include oculo-dento-digital dysplasia (ODDD), mandibulo-facial dysostosis, cleido-cranial dysostosis, progeria and other progeroid syndromes. <sup>[8]</sup>

Recognition of this syndrome at birth is very important, as potential complications are related to the narrow upper air-way associated with the craniofacial configuration: early pulmonary infections, sometimes lethal, respiratory and feeding difficulties, obstructive sleep apnea and anesthetic risk (difficult intubation). <sup>[5]</sup>

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