University Journal of Surgery and Surgical Specialities

ISSN 2455-2860 2021, Vol. 7(4)

HALLERMAN-STREIFF SYNDROME: A CASE REPORT

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ABSTRACT

We report a 14 year old boy presented with complaints of defective vision in both eyes since birth. On examination oculomandibulardyscephaly, bird face, proportioned dwarfism, hypotrichosis, cutaneous atrophy, bilateral microphthalmos and bilateral congenital cataract was found. A clinical Diagnosis of Hallerman Streiff Syndrome was made. Patient underwent LensAspiration and removal of Persistent Pupillary Membrane with Anterior Vitrectomy was done under general anaesthesia in both eyes. Post-surgery his visual acuity improved significantly. Patient was able to attend normal school like any other child of his age. This case is presented because of its rarity and to emphasis the pivotal role played by ophthalmologist in treatment and rehabilitation of patients with Hallermann Streiff syndrome.

KEYWORDS: Hallermann Streiff syndrome, Cataract, Microphthalmos

INTRODUCTION

Hallermann Streiff Syndrome is an oculomandibulardyscephaly. This condition was first reported by Hallermann in the year 1948 and by Streiff in 1950. Hallerman Streiff Syndrome mainly occurs due to developmental disturbance during 5^{th} to 7^{th} week of embryonal life. Hallerman Streiff Syndrome presents with constellation of ophthalmic and systemic features. Prominent features include bird like faces, proportioned dwarfism, cutaneous atrophy, bilateral microphthalmos andbilateral congenital cataract. Treatment of this condition which caused congenital blindness is challenging and needs multidisciplinary approach.

CASE REPORT

A 14 year old boy presented to us with complaints of defective vision in both eyes since birth, he was born out of non-consanguineous marriage, delivered after full term of pregnancy and cried immediately after birth. No similar illness was seen among his family members.

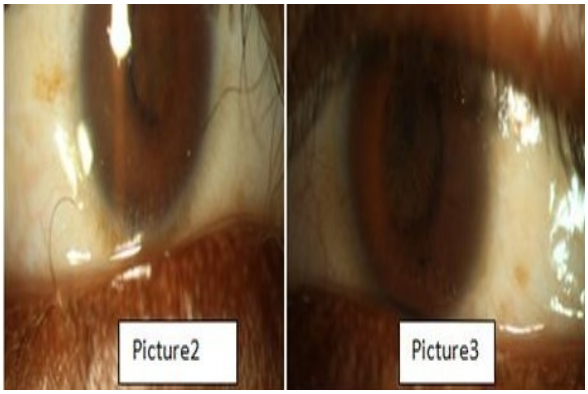
On examination oculomandibulardyscephaly, bird face, proportioned dwarfism(his height was 142cm as compared to an average 14 year old Indian boy who would measure 160cm of height), hypotrichosis, cutaneous atrophy and bilateral microphthalmos was noted (Picture1). His cardiovascular system, respiratory system and central nervous system were found to be clinically normal.

On ocular examination his best corrected visual acuity in Right Eye was Hand movements (HM) with projection of light rays (PR) accurate in all quadrants. His best corrected visual acuity in Left Eye was 1/60 with projection of light rays (PR) accurate in all quadrants. No abnormal head posture was noted. Bilateral pendular nystagmus which was high in frequency, moderate amplitude, no latent component and no dampening on convergence was noted. Both Eyeshad 1.epicanthal fold, 2.

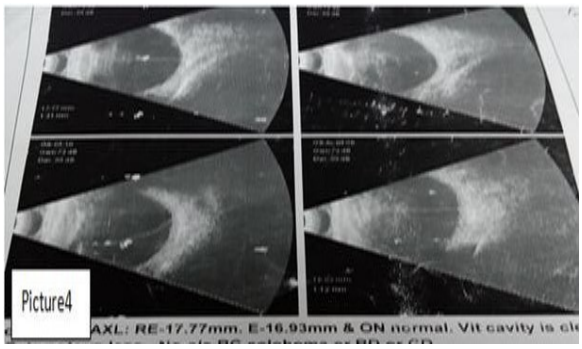
microcornea (horizontal diameter in both eyes were 7mm), 3. Iris atrophy and 4. Persistent pupillary membrane. Inner details like lens status and fundus could not be made out due to thick persistant pupillary membrane in both the eyes. Right eye had 30 degree esotropia. Intraocular pressures were normal in both eyes.

Picture2: Picture of Right eye with microcornea and persistant pupillary membrane.

Picture3: Picture of Left eye with microcornea and persistant pupillary membrane.



Patient was subjected to investigation like USG B-scan which revealed bilateralmicrophthalmos and bilateral cataract. Right eye axial length was 17.66mm and Left eye was 18.20mm (Picture4)



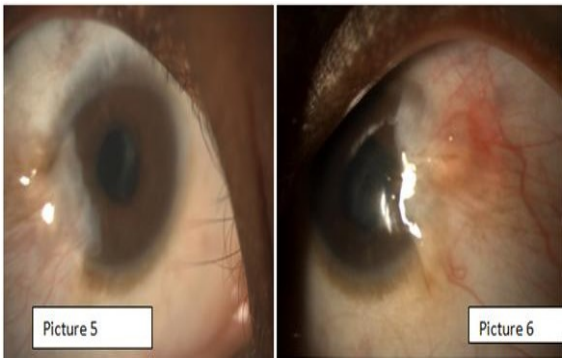
As our patient had all feature like bird faces, proportioned dwarfism, cutaneous atrophy, bilateral microphthalmos and bilateral congenital cataract as suggested by Joule Francois in his criteria for Hallerman Streiff syndrome[1] , a diagnosis of Hallerman Streiff Syndrome was made.

Patient underwent surgery for Left Eye under general anaesthesia, Lens aspiration and removal of persistant pupillary membrane with Anterior vitrectomy was done. Intra operative period was uneventful; patient did not have any complication due to anaesthesia. As the eye was microphthalmic, intraocular lens was not implanted and eye was left aphakic. Postoperative period was uneventful and fundus was found to be normal. At the time of discharge, Best Corrected Visual Acuity (BCVA)in the left eye was 6/24 with +16.00DS Lens.

After 3 months patient underwent similar surgery for RIGHT EYE under general anaesthesia, Lens Aspiration, removal of Persistent Pupillary Membrane with Anterior Vitrectomy was done. Intraoperative period was uneventful. As the eye was Microphtalmic Intraocular lens was not implanted and the eye was left aphakic. Postoperative period was uneventful and fundus was found to be normal. At the time of discharge Best Corrected Visual Acuity (BCVA) in the right eye was 2/60 with +17.00DS Lens. The vision improvement was poor in right eye due to strabismicamblyopia.

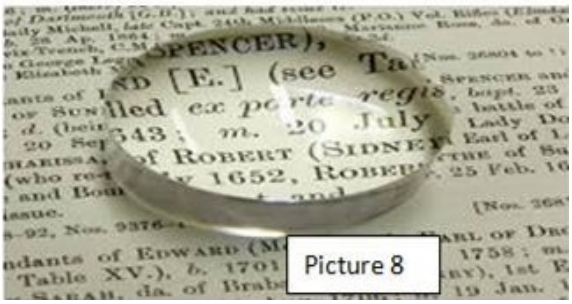
Patient was advised to attend Low Vision Clinic, patient was advised to use 4X Dome Magnifier for reading. Patient was able to read N8 with dome magnifier. Post-surgery our patient is now attending normal school like other children in his age group.

Picture 5&6: Post-operative picture of Right and Left eye respectively. Both showing clear visual axis (persistent pupillary membrane removed) and Aphakia.



Picture 7: Post-operative picture with Aphakic correction.

Picture 8: 4x Dome Magnifier used by the patient for reading.



DISCUSSION

Hallerman Streiff Syndrome is an Oculomandibular Dyscephaly. This condition was first described by Arbry in the year 1893.Clinical findings were first reported by Wilhelm Hallerman in the year 1948[2] followed by Enrico Streiff in 1950[3]. Diagnostic criteria for Hallermann Streiff Syndrome was proposed by Belgian Ophthalmologist Jule Francois in

POSITIVE SIGNS	NEGATIVE SIGNS
Dyscephaly	No Auricular Anomaly
Bird Face	No Muscular Anomaly
Proportioned Dwarfism	No Nail And Limb Anomaly
Hypotrichosis	No Mental Retardation
Cutaneous Atrophy	No Palpebral Anomaly
Bilateral Microphthalmos	
Bilateral Congenital Cataract	

the year 1958[1]. He proposed 7 positive signs and 5 negative signs. Negative signs are mainly to differentiate Hallermann-Streiff syndrome from other OculomandibularDyscephaly.

Hallermann Streiff syndrome occurs mostly due to sporadic single dominant gene mutation[4]. No chromosomal anomaly had been identified, yet few familial cases had been reported. HallermannStreiff Syndrome mainly occurs due to developmental disturbance during 5th to 7th week of embryonal life, during development of second branchial arch. Systemic features associated are (i) Skin anomalies: Cutaneous Atrophy above the nose, Hypotrichosis.(ii) Oro dental anomalies: Cleft Palate, Micrognathia, restricted jaw movements, Hypodontia, adontia, disalignment, enamel hypoplasia.(iii) Respiratory anomalies: Tracheomalacia, Upper Airway Obstruction, Sleep Apnea. Respiratory anomaly in Hallermann Streiff syndrome would lead to severe morbidity and even mortality in these patients. Tracheomalacia is very common in these patients. Ophthalmologist and Anaesthesiologist must be aware of this complication before subjecting these patients to general anaesthesia.Congenital cardiac anomalies: Atrial septal defect and ventricular septal defect. Associated Ocular featuresare Microcornea, bilateral congenital cataract, Nystagmus, Strabismus, Glaucoma, Aniridia, Iris atrophy, Persistent pupillary membrane and Coloboma.

CONCLUSION

Hallermann Streiff Syndrome is an oculomandibular discephaly and is a cause for congenital blindness. Treatment of this condition is challenging and needs multidisciplinary approach. Awareness of systemic complications and anaesthetic complications before subjecting the patient to surgery under general anaesthesia is important. Ophthalmologists play a pivotal role in treatment and rehabilitation of these patients.

REFFERENCES

1. François J. A new syndrome: dyscephalis with bird face and dentalanomalies, nanis, hypotricosis, cutaneous atrophy, microphtalmiaand congenital cataract. Arch Ophtalmol 1958;60:842-62.

2. Hallermann W. Voghelsicht and cataractacongenita. KlinMblAugenhlink 1948;113:315-8.

3. Streiff EB. Dysmorphiemandibulo-faciale (teted'oiseau) etalteration oculaires. Ophtalmologica 1950;120:79-83.

4. Bucak et al. Hallermann-Streiff Syndrome Erciyes Med J 2014 36(3): 130-2