Juvenile Xanthogranuloma Causing Bilateral Eyelid Swelling and Severe Ptosis in a Nine-Year Old Male

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Background: Juvenile Xanthogranulomas (JXG) are rare benign lesions that belong to the large group of non-Langerhan cell histiocytoses.[1]The most common ocular presentation is on the Iris.[2] They occur predominantly in children (40%-75%) in the first year of life[2] with >15-20% having lesions at birth.[3]Eyelid lesions are extremely rare and tend to present later in life.[3] There are two existing variants: a) multiple 2 – 5 mm dome shaped papules and b) one or more large nodules 1 - 2 cm in size. There is a slight male preponderance with a male to female ratio of 1.7:1 and prevalence of 1 in 1 million. [2]

Case Report: A 9-year-old child presented to the eye clinic with a 4-year history of bilateral upper eyelid swelling. It initially started on the left eye and then involved the right eye a few months later. The swelling was painless, slowly progressive with no associated history of redness, itching**.** Blurring of vision was observed with progressive increase in size of the swelling. On examination, visual acuity was 6/9-1 in the right eye and 6/9+3 in the left eye. Both eyelids were ptotic with palpebral fissure height of 3mm.

Palpable in the right upper eyelid were two firm, fluctuant, nodular masses with smooth surface measuring (3cm x 2cm and 2cm x 2cm). There was no attachment to the underlying and overlying structures and no skin discolouration. In the left upper eyelid, there were 4 masses each measuring 1.9cm x 1.7cm with similar characteristics to those on the right. Intraocular pressure was 17.6mmHg on the right and 21.3mmHg on the left. Fundoscopy was done with much difficulty and found to be normal for both eyes. Systemic examination did not reveal any systemic involvement. Complete Blood Count, Liver Function Test and Renal Function Test were normal. Computed tomography scan showed a superolateral lobulated isodense mass with contrast enhancement with no orbital extension.

A clinical diagnosis of Juvenile Xanthogranulomas was made. The differential diagnoses included Lymphoma, Rosai Doffman syndrome, Langerhan histiocytosis, Multiple myeloma, Malignant fibrous histiocytoma. The patient underwent excision biopsy of the right upper eyelid mass. Intraoperatively, a pinkish-yellow mass was found between orbicularis muscle and orbital septum with surface telangiectasia. Complete excision was done. Histology revealed pseudo-tumorous benign granulomatous proliferation with numerous histiocytes, confirming a diagnosis of giant juvenile xanthogranuloma of the eyelid. Excision biopsy was also performed for the lesions in the left eye.Visual acuity returned to 6/6 in both eyes and ptosis markedly improved from first day post-operatively. The patient has remained stable and no recurrence has occurred.

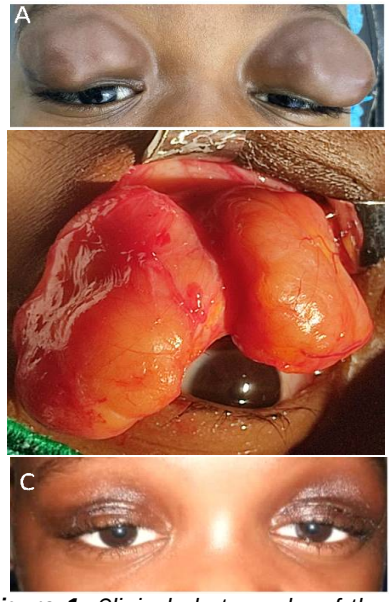

Figure 1: *Clinical photographs of the patient. A- Preoperative; B- Intraoperative (Right eye); C – postoperative*

Discussion: JXG is a rare and typically benign disease that presents in children. Small isolated forms are under-diagnosed.[3] Our case typically belongs to the 2nd variant and can be a cause psychosocial disability. The aetiology is unknown – Non neoplastic process characterized by an abnormal response to a non-specific injury e.g trauma or viral infection. Common systemic associations are Juvenile chronic myelogenous leukemia, Neurofibromatosis, urticaria pigmentosa, Insulin-dependent DM, Aquagenic pruritus, CMV infection. Serious complications can arise when systemic juvenile xanthogranuloma is involved. Simple excision biopsy yields good results in uncomplicated cases.

Conclusion: Bilateral eyelid swelling could result from Juvenile Xanthogranuloma. Histopathologic diagnosis and careful examination to rule out systemic association is required. Complete resolution can be achieved after excision biopsy.

References

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Goldenhar Syndrome: A Case Report

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Introduction: Goldenhar Syndrome, (GS) also called oculoauriculovertebral dysplasia is a rare congenital disease arising from abnormal development of the first and second branchial arches[1-3] Incidence is 1 in 3500-5600[4,5], male to female ratio of 3:2[1], 85% unilateral presentation[1,5.] Multifactorial risks factors implicated are genetic[1,6]; deletion of 5p, 14q23 duplication, Chr18,22 anomaly and 14q23 duplications are implicated. Others are gestational diabetes, second trimester bleeding, multiple gestation and maternal ingestion of vasoactive drugs: aspirin, pseudoephedrine, accutane, ibuprofen[6-8] and alcohol.[9] Viral[5], environmental and other unknown factors are suspected as well. Clinical features include hemifacial microsomia, facial asymmetry maxillary hypoplasia, malar flattening[4]. Ears may show, accessory tragi, tags, meatal atresia, low set ears, microtia and [6.] laryngomalacia[4-6,10]. Spine may have scoliosis and hemivertebrae[10]. Cardiac defects include ventricular septal defects (VSD) and transposition of great vessels[4,6,10]. Tracheo-esophageal fistula and gastrointestinal (GIT) defects are associated[4,6,10]. Central nervous system (CNS) defects include hydrocephalus, hypoplastic corpus callosum[4,6,10]. Ophthalmic features occur in 60%[11] and include epibulbar dermoid, usually located infero-temporally. Others are upper lid coloboma, unequal palpebral fissures, subconjunctival dermoid, uveal coloboma, retinal coloboma, cataract, strabismus, microphthalmia and anophthalmia[4,6]. There is an association with glaucoma[4]. Diagnosis is usually by phenotypic appearanceand radiological investigations (Computed Tomography, Magnetic Resonance Imaging)[3,4]. Genetic tests help to support the diagnosis and Ultrasound scans aid prenatal diagnosis[1]. Treatment is multidisciplinary with interventions depending on age, severity and extent[1]. Early intervention may be required in airway problems such as obstructive sleep apnoea,