Juvenile Xanthogranuloma A Case with Rare Ocular Manifestations

Dr. Reena A. DO MS, Dr. Nandakumar MD, Dr. Sony Siraj E., Dr. Tinu Mary Thomas

Introduction

Juvenile xanthogranuloma is a benign self limiting dermatological disorder which is rarely associated with systemic manifestations. The eye, particularly the uveal tract is the most frequent site of extracutaneous involvement. There are a few case reports in the literature describing juvenile xanthogranuloma with some ocular features mainly hyphaema. Juvenile xanthogranuloma is the most frequent cause of spontaneous hyphaema in childhood. We report an interesting case with rare ocular manifestations, characterised by the absence of hyphaema.

Case Report

A four year old boy presented to our institute with generalized pruritic skin lesions of one month duration with photophobia and watering of both eyes for one week. There was no significant antenatal and postnatal history. His best corrected visual acuity could not be checked at the time of presentation due to intense photophobia and watering. General examination revealed multiple non-tender discrete mobile lymph nodes involving bilateral cervical, axillary and inguinal areas.

Systemic examination showed multiple papules and plaques varying in size between two mm and two cm involving the trunk, limbs, palms and face including the eyelids which were either skin coloured or erythematous.[Fig 1a, 1b & 1c]

The child had severe photophobia and blepharospasm, preventing proper ocular examination. Examination under anaesthesia was done and this showed severe conjunctival and circumcorneal congestion with multiple conjunctival granulomas involving both palpebral and bulbar conjunctiva [Fig 2]. Corneal stroma was studded with greyish white infiltrative lesions with a hypopyon of three to four mm in the anterior chamber. Iris also showed numerous granulomatous lesions. There was limited view of the posterior segment due to severe anterior chamber reaction and vitreous haze. Red glow was present bilaterally. Clinical examination of other systems were within normal limits.

Investigations included complete blood count, peripheral blood smear, erythrocyte sedimentation rate, liver function lists, renal function tests. Erythrocyte Sedimentation Rate (ESR) was elevated at 100 mm/hour. Thrombocytosis was identified. Serology was negative for HIV and VDRL. A chest X-ray was also taken and was found to be normal. Immunohistochemistry was done. S-100 was found to be positive.

Histopathology of the conjunctival lesions showed mild hyperplasia of lining epithelium, the deeper areas showing collection of plasma cells, lymphocytes and foamy histiocytes.

Lymph node biopsy showed partial effacement of the architecture with infiltration of the subcapsular and paracortical region with sheets of cells with abundant eosinophilic cytoplasm and bland looking nucleus. [Fig 3a & 3b].
Skin biopsy from one of the eyelid lesions showed sheets of foamy macrophages with moderate to abundant eosinophilic cytoplasm and vesicular nucleus filling papillary and mid-dermis and scattered in the lower dermis. Lymphocytes, neutrophils and touton giant cells are seen in between. Histological appearance were confirmatory of xanthogranulomatous process.[Fig 4a & 4b]

The child was treated with topical and systemic steroids, topical cycloplegics and antiglaucoma medications. He responded very well to steroids with marked reduction in inflammatory signs and infiltrative lesions. At six weeks follow up there was significant clearing of the corneal and iris lesions. Skin lesions showed a gradual improvement. The child had two episodes of exacerbation of skin lesions in the past one month for which he was put on systemic steroids and antibiotics. On systemic steroids he showed excellent improvement and the drug was gradually tapered. The child is still having recurrences and is on maintenance dose of steroids. He is on regular follow up.

**Discussion**

Juvenile xanthogranuloma is a benign proliferative disorder of the non-Langerhans type of histiocytosis. It is characterised by benign, usually asymptomatic self healing red to yellowish papules and nodules composed of histiocytic cells. It predominantly occurs in infancy and childhood presenting at less than one year of age in 70% of cases. There is a male predominance in all categories of clinical presentation but especially noticed in the group with multiple cutaneous lesions.

Its mainly a skin disorder characterised by a typically raised orange skin lesions occurring either singly or in crops which regress spontaneously. Most frequent site of occurrence is the head and neck region followed by trunk and upper extremities.

Extra cutaneous form is rare and the most commonly involved site is the eye and periorbital region. The commonest ocular site is anterior uvea but any part of the eye may be affected. Although the lesions are histologically benign they bleed easily and are the commonest cause of spontaneous hyphaema in children. It was characteristically absent in our case. Progressive deterioration of vision caused by secondary glaucoma is a serious potential complication of uveal involvement.

Occasionally, the lesions may present in other areas such as cornea, lids and orbit. Our case of interest had multiple corneal infiltrates with hypopyon. It may also
be seen on the surface of ciliary process and are rarely seen in the vitreous. Involvement of the optic disc is a rare complication of juvenile xanthogranuloma.

Histological diagnosis depends on the demonstration of xanthoma cells infiltrating beyond the dermis. Touton giant cells, lymphocytic infiltration and focal hyaline necrosis may also be present. Old lesions demonstrate fibrosis. Histiocytes contain pleomorphic nuclei with few or absent mitotic figures and irregular dense bodies. In juvenile xanthogranuloma, histiocytes are positive to antibodies against Factor XIIIa, HAM 56, HHF35, CD 68 and vimentin and are generally negative to CD 1a and S 100. S 100 was found to be positive in our case. Other tests could not be performed due to lack of facilities.

Therapy depends on the condition of the eye. Early treatment of iris involvement is mandatory because if untreated it can lead to uncontrolled glaucoma, corneal blood staining and amblyopia. Antiglaucoma therapy can be added whenever necessary. Conservative management is justified because spontaneous regression of ocular lesions might occur. Topical steroids may not prevent recurrent hyphaemas and glaucomas. Supplemental therapy with periocular steroids, systemic steroids or low dose radiotherapy or a combination might be necessary. The iris lesions like the skin lesions may be self-limiting and resolution may not be related to the therapy.

Both cutaneous and extracutaneous lesions involute spontaneously with in three to six years. The relapse rate is found to be 7%. Correct diagnosis is especially important because of the possibility of successful eradication of the lesions and control of complications. Ocular, neurologic and hepatic diseases are rare but may have serious long term consequences.

References


