

Case Report

# A Subcutaneous Juvenile Xanthogranuloma in a 4-Year-Old Girl Who Presented with a Lower Eyelid Mass

Amjad A. Saifaldein<sup>a</sup> Faeqah H. Almahmoudi<sup>b</sup> Rafea I. Babgi<sup>b</sup>  
Alaa A. Alsammahi<sup>c</sup>

<sup>a</sup>Department of Ophthalmology, KAMC – Ministry of Health, Jeddah, Kingdom of Saudi Arabia; <sup>b</sup>Department of Ophthalmology, King Fahad Armed Forces Hospital, Jeddah, Kingdom of Saudi Arabia; <sup>c</sup>King Abdulaziz University Hospital, Jeddah, Kingdom of Saudi Arabia

## Keywords

Infant · Juvenile xanthogranuloma · Eyelid

## Abstract

Juvenile xanthogranuloma (JXG) is a relatively uncommon, benign, histiocytic proliferative cutaneous disorder that typically affects children, with the head and neck being the most common sites. The present case report describes an isolated subcutaneous JXG in a 4-year-old girl who presented with a circumscribed oval mass located in the lower eyelid of the right eye. This lesion was histologically diagnosed as JXG after a surgical resection of the mass.

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## Introduction

Although an uncommon condition, juvenile xanthogranuloma (JXG) is the most common form of non-Langerhans cell histiocytosis [1]. The first reported case of JXG was reported in 1905 [2], which was generally described as a benign, self-limited histiocytic disorder of the

skin [3]. The incidence is unknown but has been reported to represent 0.5% of all paediatric tumours [4]. JXG may present at birth (5–17% of cases) or in the first year of life (40–70% of cases). During infancy, male predominance can be seen in the ratio of 1.5:1, with a tendency of spontaneous regression [5]. Up to 80% of JXG cases can present as a solitary lesion, with fewer cases presenting as multiple well-demarcated lesions [3]. Although the head, neck, and trunk are the most common sites for JXG, it can appear anywhere on the body, including the groin, scrotum, penis, clitoris, toenail, palm soles, and lip [6–8]. However, it may also occur in the bone, genitourinary tract, tympanic membrane, spinal column, brain cortex, bone marrow, lung, and liver [9–15]. Extracutaneous involvement is usually restricted to the eye area. Ocular involvement of JXG includes the orbit, eyelid, conjunctiva, limbus, uvea, retina, and optic disc [16–23]. Although mainly a cutaneous lesion, some rare cases have been reported as pure subcutaneous lesions. However, very few cases have been reported to involve the eyelid [9, 24–27].

We describe a case of an isolated subcutaneous JXG located in the lower eyelid without any additional cutaneous or systemic nodules in a 4-year-old girl.

## Case Report

A 4-year-old girl presented to the ophthalmology outpatient clinic with history of painless right lower eyelid mass for 3 weeks. There was no history of bleeding or discharge from the surface of the lesion and no history of fever or trauma. The patient was not on any topical treatment or systemic medication.

She had a known case of lactose intolerance. Additionally, she had a previous history of abdominal distention at the age of 6 months and a history of poor appetite. The patient had no family history of similar illness. The patient's mother had a normal pregnancy and normal labour.

On examination, visual acuity without correction was 20/20 in both eyes (OU). Pupils were round, regular, and reactive to light with no afferent pupillary defect. Orthoptic evaluation showed orthophoria with full ocular motility OU. Her refraction was not significant OU.

Results of bilateral anterior and posterior segment examinations were within the normal range. The right lower eyelid mass was 1 × 1 cm in size, firm, not tender, mobile, and well circumscribed. The skin above the lesion was red in colour (Fig. 1). There was a chalazion-like lesion on the left lower eyelid margin. Laboratory investigations including a complete blood count, liver function test, and renal function test were negative.

Magnetic resonance imaging (MRI) of the orbit showed an oval-shaped soft tissue lesion of the right lower eyelid which measured 0.6 × 0.6 × 0.7 cm, with low signal on T1 and high intensity on T2. The lesion was confined to the subcutaneous tissue, not exerting any mass effect on adjacent structures and not arising from the bone. The globe, extra-ocular muscle, optic nerve, and retrobulbar fat were normal with no evidence of intra-orbital mass (Fig. 2). The patient was referred to a paediatrician for general investigation, including skeletal X-ray and abdominal ultrasound, which showed normal bones and visceral organs, respectively.

The patient had an excisional biopsy of the right lower eyelid mass (Fig. 3) and the specimen was sent for histopathology. Histopathology of the right eyelid mass showed multiple fragments of fibrovascular and granulation-like tissue, heavily infiltrated by inflammatory cells. Histopathology also revealed a non-caseating granuloma and collection of foamy histiocytes and rare Touton-like cells (Fig. 4). The specimen was negative for atypia and signs of malignancy, as well as being negative for CD1 and S100. According to these results, the patient

was diagnosed with JXG. Histopathology of the left eyelid mass lesion showed a nonspecific chronic inflammatory response, suggestive of chalazion.

## Discussion

JXG is a rare, benign lesion which is usually confined to the skin [25]. Ocular JXG may occur without concomitant skin involvement, with 92% of patients being younger than 2 years of age [28]. The ocular tissues affected by JXG include the iris (68%), conjunctiva (19%), eyelid (6%), choroid (6%), and orbit (3%) [17]. Eye involvement is usually, but not always, unilateral and commonly presents with an asymptomatic iris tumour, a red eye with signs of uveitis, unilateral glaucoma, spontaneous hyphema, or heterochromia iridis [6]. Eyelid involvement by JXG is an uncommon presentation [21] and has only been reported in a small number of cases, with only 2 known cases involving the eyelid [13, 25, 29]. One such case was a congenital subcutaneous eyelid JXG with orbital involvement which was treated with an intralesional steroid [26]. The other case was reported in a 10-month-old infant, which resulted in visually significant astigmatic amblyopia [27].

We describe a case of solitary subcutaneous JXG involving the lower eyelid, without any other cutaneous or systemic lesions and non-significant refraction. JXG can be differentiated from xanthoma by the distribution of the lesion and absence of lipid abnormalities. Other differential diagnoses include molluscum contagiosum (pearly, dome-shaped papule with central umbilication), haemangioma, and neurofibroma (firm lesion with associated cafe-au-lait spots) [28].

The diagnosis of JXG is mainly based on characteristic clinical features and confirmed by histopathology. Histopathology of JXG is characterised by Touton giant cells which are seen in 85% of JXG cases. S-100 protein immunoreactivity, a marker for the diagnosis of Langerhans cell histiocytosis, is typically absent [30].

The lesion is sometimes associated with neurofibromatosis type 1, juvenile chronic myelogenous leukaemia, Niemann-Pick disease, diabetes insipidus, lytic bony lesions, and urticaria pigmentosa [3, 6, 30].

JXG is a self-limiting disease that often spontaneously regresses [8, 30]. Lesions may resolve completely or may leave a residual atrophic or hyperpigmented scar. Conservative management of these lesions has been advocated [31]. Despite the likelihood of spontaneous regression, excision of the lesion(s) is often decided for aesthetic or diagnostic reasons, as was done in our case. Excision of the lesion is an adequate treatment and recurrence is uncommon, although it has been reported [32].

## Conclusion

Juvenile xanthogranuloma is typically a benign and self-limiting disease, and treatment may be necessary to prevent amblyopia if the eyelid is involved. Early multidisciplinary evaluation for systemic lesions and associated malignant conditions should be considered.

## Statement of Ethics

The authors have no ethical conflicts to disclose.

## Disclosure Statement

The authors report no conflicts of interest.

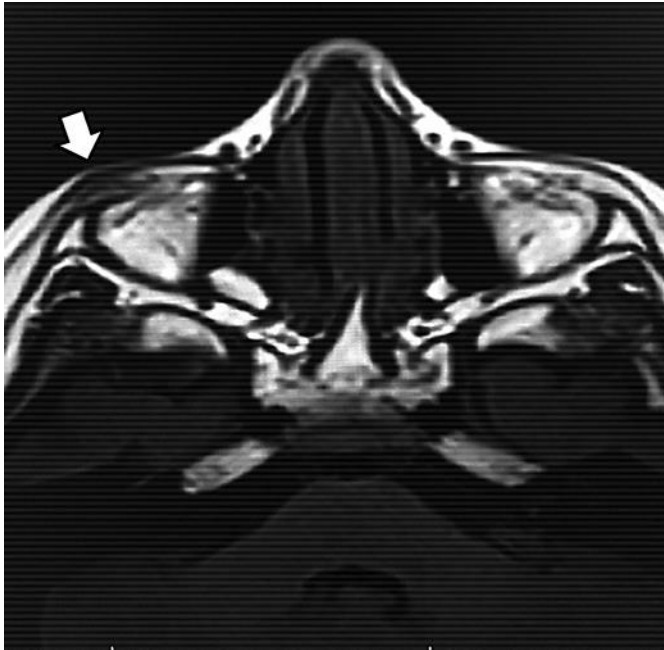
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**Fig. 1.** Right subcutaneous lower eye lid mass 1 month after the initial presentation.

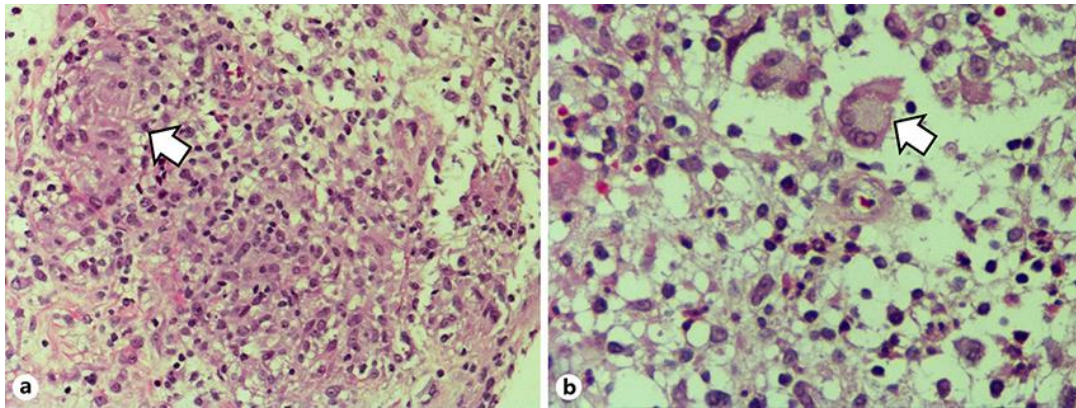


**Fig. 2.** MRI T1 axial plane, the arrow points to the mass – an oval-shaped soft tissue lesion of the lower eyelid of the right orbit, measuring  $0.6 \times 0.6 \times 0.7$  cm with low signal.



**Fig. 3.** Postoperative week 6.





**Fig. 4.** Right eye. **a** Non-caseating granuloma. **b** Touton-like giant cells.