Congenital juvenile xanthogranuloma with ulceration: a pediatric case report

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Abstract

Congenital juvenile xanthogranuloma (JXG) is an uncommon diagnosis and even more rarely presents with ulceration. We report such a case in a two-week-old girl. Biopsy was performed to rule out any concerning entities. Adequate treatment was provided with topical petrolatum and occasional miconozole or zinc oxide; the mass spontaneously regressed. Because congenital JXG has an excellent prognosis, insight into unique presentations such as this may provide useful information and avoid unnecessary surgical interventions.

Keywords: xanthogranuloma, congenital, juvenile, giant

Introduction

Juvenile xanthogranuloma (JXG) is an uncommon benign non-Langerhans cell histiocytosis, that typically occurs in children [1]. Approximately 20% are present at birth and most arise in the first year of life [2]. The etiology of JXG is unknown and the cell of origin has been proposed as a plasmacytoid monocyte, but this is a matter of ongoing debate. Juvenile xanthogranuloma generally carries a good prognosis, which includes spontaneous resolution over the course of a few years with minimal to no intervention [3].

Clinically, JXG can sometimes be difficult to diagnose owing to its myriad of subtypes and presentations [4]. Subtypes include classic, giant (>2cm), cluster, and extracutaneous. Although they can occur anywhere on the body, the majority of cases present as a solitary skin lesion on the head, neck, or upper trunk. A small percentage of patients may have ocular involvement, and in rare occasions, visceral or deep soft tissue involvement may be found. Juvenile xanthogranulomas commonly present as reddish or yellowish to brown papules, plaques, or nodules [5]. Early lesions appear red and raised; later lesions appear yellow and flattened owing to increased lipidization.

Congenital JXG is an uncommon diagnosis and even more rarely presents with ulceration. We report such a case in a two-week-old girl. Because congenital JXG has an excellent prognosis, insight into unique presentations such as this may provide useful information and avoid unnecessary surgical interventions.

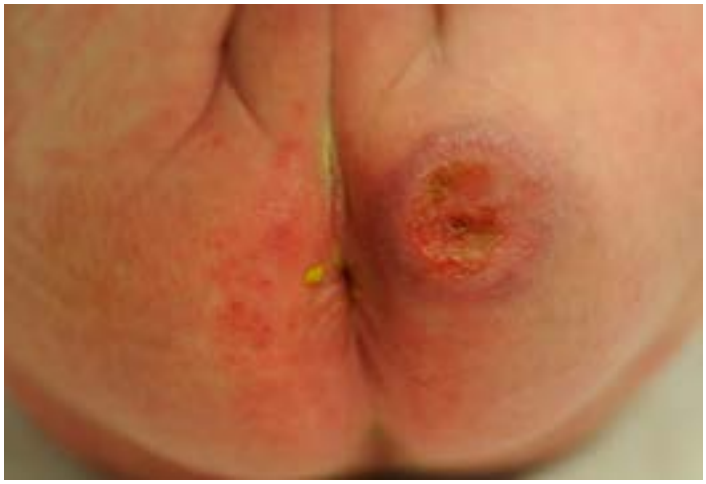

Figure 1. *Very firm, non-pulsatile pink-violaceous nodule over the left medial buttock, approaching the perianal rim with central ulceration.*

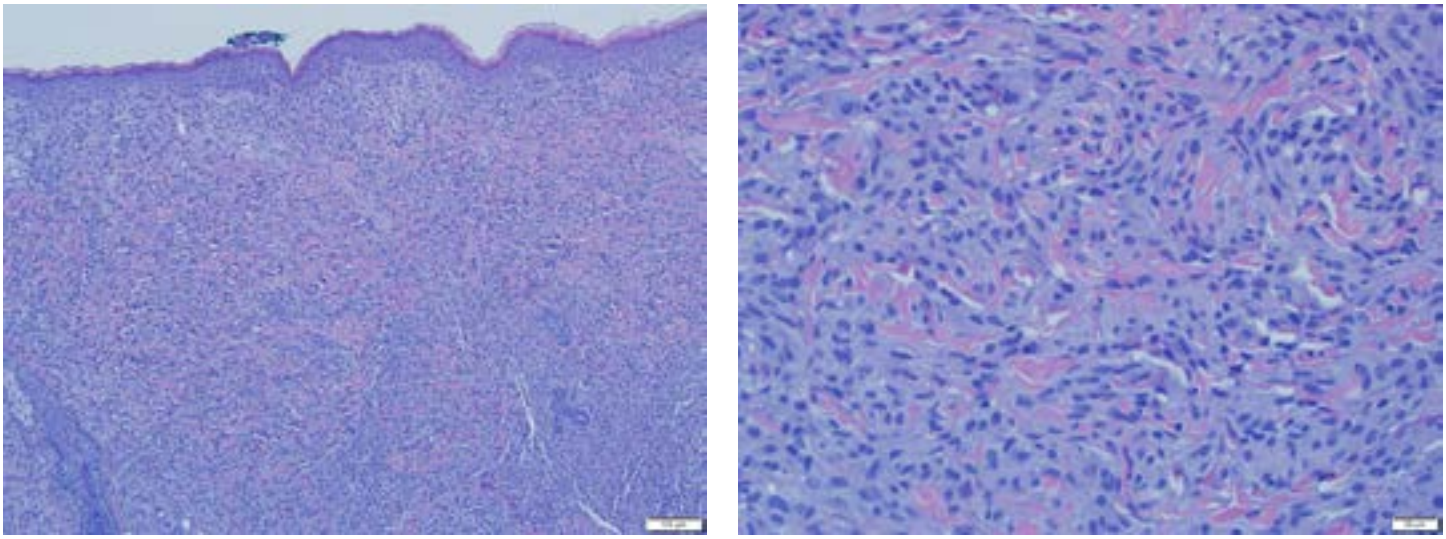

Figure 2. *Hematoxylin-eosin stain showed diffuse dermal infiltrate of monomorphous foamy histiocytes with bland cytology and amphophilic cytoplasm with indistinct borders. A) 10x, B) 40x.*

Case Synopsis

A term 2-week-old girl presented for evaluation of a solitary congenital mass on her left medial buttock. It had been present since birth without any appreciable changes in size, texture, or color, but developed recent central painful ulceration. Parents denied lesions elsewhere on her body. She was born large for gestational age via prolonged vaginal delivery without other complications and was otherwise thriving. There was no family history of skin issues.

Ultrasound showed a solid mass without inherent vascularization. Physical exam revealed a relatively well-defined 4cm x 4.5cm, very firm, non-pulsatile pink-yellow nodule with a dusky-violaceous border (**Figure 1**) over the left medial buttock, approaching the perianal rim. At the center of the nodule there was a shallow ulceration with clean exudate. A 4mm punch biopsy of the lateral edge of the lesion was taken; findings are shown in **Figure 2**.

Histopathological examination revealed a diffuse dermal infiltrate of monomorphous foamy histiocytes with bland cytology and amphophilic cytoplasm with indistinct borders. Rare typical mitotic figures were present and there were numerous admixed eosinophils within the infiltrate. CD68, CD163, and CD31 immunohistochemical stains highlighted the cells of interest, whereas immunohistochemical stains for S100, CD1a, CD117, tryptase, CD34, pancytokeratin and CD45 were negative. The histological findings and immunophenotypic profile were consistent with the diagnosis of giant congenital juvenile xanthogranuloma. The biopsy was sent to a soft tissue pathology expert for confirmation of the above diagnosis.

Supportive therapy was the treatment of choice, including petrolatum ointment with occasional use of topical miconazole and zinc oxide as barriers. Ophthalmologic evaluation was recommended. At the four-month follow-up, parents reported marked regression of the mass and no new skin lesions. Examination revealed a smaller, sessile, tan to somewhat yellow, firm round plaque at the left medial buttock with several surrounding satellite small yellow-tan papules (**Figure 3**). At the one-year follow-up, parents reported the mass had almost completely regressed, flattened, and softened leaving only a scar-like area of lighter skin (**Figure 4**). They denied subsequent issues with ulceration or pain but did note mild irritation of the lesion with associated "diaper rashes."

Case Discussion

Congenital giant JXG is a rare subtype with less than twenty reported cases, including the current. Associated ulceration is infrequent [6]. Although JXGs more commonly affect males, the giant subtype is more common in females. The differential diagnosis for JXG includes other dermal neoplasms such as: mastocytoma, Langerhans cell histiocytosis, xanthoma, and melanocytic neoplasm (e.g. Spitz nevus or tumor), among other entities. Biopsy may be

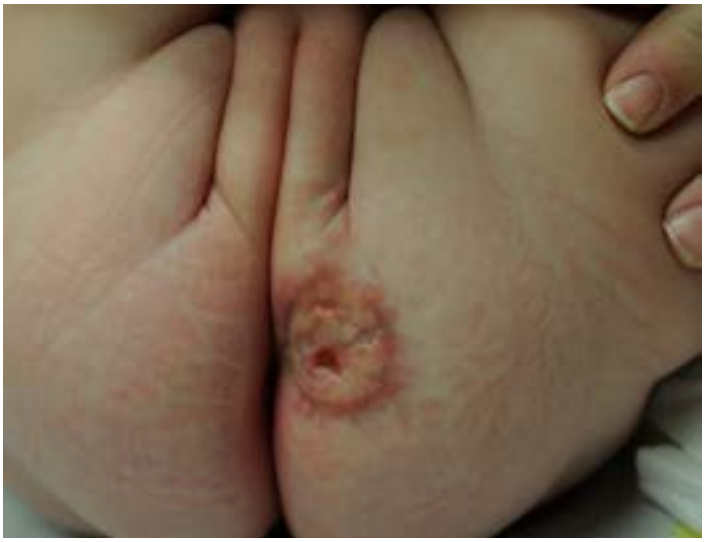

Figure 3. *Four-month follow-up; smaller, sessile, tan-yellow plaque at the left medial buttock with surrounding satellite papules.*

considered, especially with unique JXG presentations such as ulceration [7].

Histologically, JXG contains a nodular, densely cellular dermal infiltrate of small histiocytes with spindled to round morphology and variable amounts of eosinophilic to foamy cytoplasm. The histologic findings vary somewhat with the age of the lesion, with early lesions appearing relatively monomorphous, without significant foam cells, and mature lesions typically containing numerous foamy histiocytes and Touton giant cells [8]. Admixed lymphocytes, eosinophils, neutrophils, and mast cells may be seen, as well as rare non-atypical mitoses [9]. The biopsy in this case did not contain significant foamy cells or Touton giant cells, compatible with an early lesion of JXG.

Conclusion

This case highlights a rare presentation of JXG. Biopsy may be necessary to differentiate from other more worrisome entities, but the overall prognosis is benign and necessary intervention is typically minimal [10].

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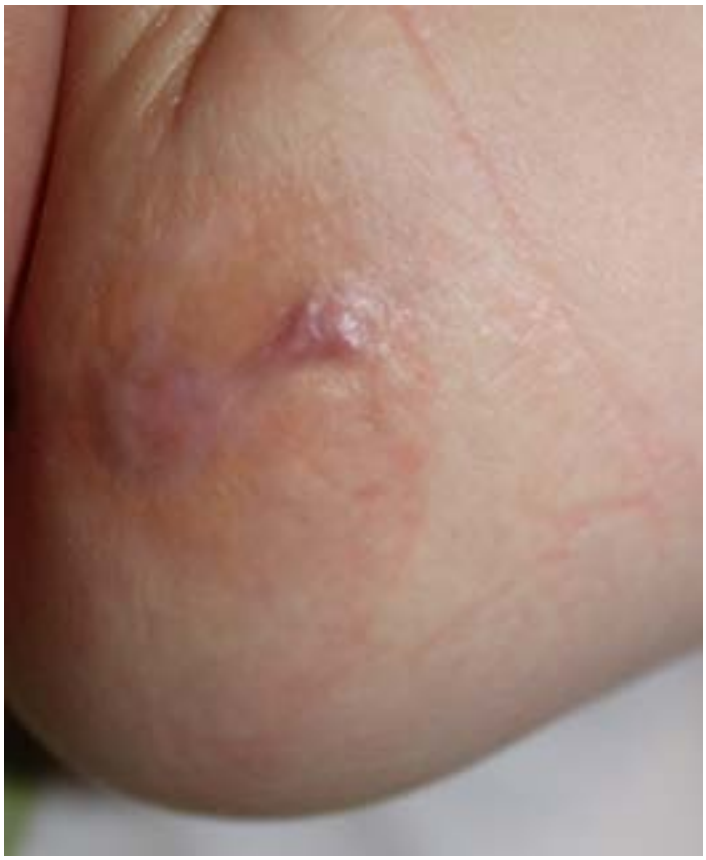

Figure 4. *One-year follow-up; two coalescing shiny, pink sclerotic papules in area of prior ulceration within a* **3.***0 x* **3.***5cm yellow-tan patch.*

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