Isolated Forefoot Juvenile Xanthogranuloma: Unique Case Study And Treatment In A Pediatric Patient

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Purpose

Juvenile xanthogranulomas (JXG) are a benign histiocytic cell proliferative disorder in early childhood. They most commonly present within the first two years of life with papular or nodular changes to the skin on the head, neck or upper trunk. Xanthogranulomas are mostly self-limiting, can be singular or multiple, and rarely can infer a systemic disease process[1]. The purpose of this study was to publish a unique finding of an extra-tendinous solitary mass in an otherwise healthy 17-year old, with surgical and medical treatment after diagnosis.

Literature Review

Juvenile xanthogranulomas are the most common of the non-Langerhans cell histiocytoses[1]. In a large tumor registry comprised of over 24,000 juvenile soft tissue masses, the incidence of JXG is approximately 0.5%. However, this could underrepresent the incidence because some of these diagnoses were made without histological confirmation[2]. Histologically JXG appear with multinucleated giant cells, and spindle cells in the younger population. In more mature tumors, there is an increased lipid composition with Touton giant cells. These immunohistochemically stain positive for Factor XIIIa, CD68, CD163, CD14, and are negative for S100[3]. Clinically, the most common appearance of these masses are nodules in subcutaneous or deep tissue or yellow plaques or papules. Solitary skin lesions are found in 67-71% of all cases, multiple lesions are seen in 7-10% of patients with JXGs[4].

Treatment is typically performed with excisional biopsy and treatment, most of these lesions are self-limiting and the majority resolve on their own through childhood[5]. Most resolve into hyperpigmented lesions, which are asymptomatic. Surgical intervention is indicated when conservative care fails, or the lesions appear malignant in nature. Rarely, these cases have systemic manifestations, but associated conditions to JXG are neurofibromatosis type 1 (NF-1) or juvenile myelomonocytic leukemia[9,10].

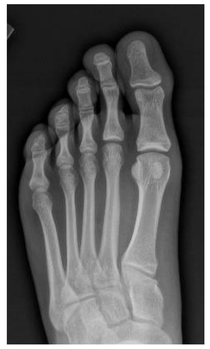

Figure 1. Normal AP radiographic view

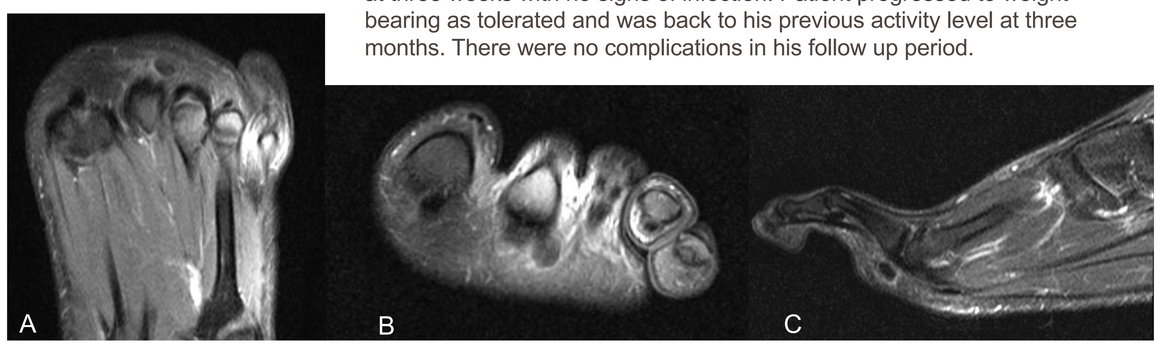

Figure 2.
A. Axial PD fat saturated MRI
B. Coronal PD fat saturated MRI
C. Sagittal T2 MRI

Case Study

A healthy 17-year-old male presented to clinic with atraumatic 2nd metatarsophalangeal joint (MTPJ) pain on his left foot. Physical examination revealed pain to palpation of the plantar 2nd MTPJ with no callus or dermatological abnormalities. There was a negative Lachman's test and no subluxation of the 2nd MTPJ present. Patient had pain on extension of the 2nd MTPJ but passive and active ROM were WNL. End extension was the most painful. Plain film radiographs were taken (Figure 1) which did not show any bony abnormality or signs of predislocation syndrome. Conservative care with modification of shoegear and orthotics were suggested.

After three months of failed conservative care, the patient returned to clinic and an MRI revealed a 2nd MTP plantar mass measuring 6mm x 6mm x 4mm, directly plantar lateral to the 2nd MTPJ (Figure 2A,B,C). The mass was hypointense on T1, T2, PD and PD fat saturated imaging. At this point in time, the patient electively wanted to undergo soft tissue mass removal. Surgical intervention was successful and a well circumscribed nodule measuring 1.1 x 0.8 x 0.6 cm was removed.

Pathological evaluation included hematoxylin and eosin (H&E) (Figure 3) and CD68 immunohistochemical staining (Figure 4). After positive CD68 staining and foam cells seen on H&E staining, the mass was diagnosed as a tuberous xanthoma. A lipid panel was then ordered for the patient to determine if he had familial hypercholesteremia. Under the American Heart Association, he did not have familial hypercholesteremia and was not put on statin therapy despite an elevated cholesterol level. The pathologic specimen was then sent to Brigham and Women's hospital (Boston, MA) and further evaluated. It was shown to have stained negative for S100, and positive for PU.1 and CD163. This showed there were bland histiocytes and no signs of malignancy. The final pathologic diagnosis was determined to be xanthogranuloma with infarction.

The patient was followed for one year and postoperatively healed his incision at three weeks with no signs of infection. Patient progressed to weight bearing as tolerated and was back to his previous activity level at three months. There were no complications in his follow up period.

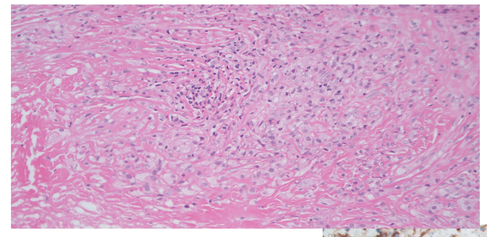

Figure 3. (Left) Hematoxylin and eosin staining with foam cells

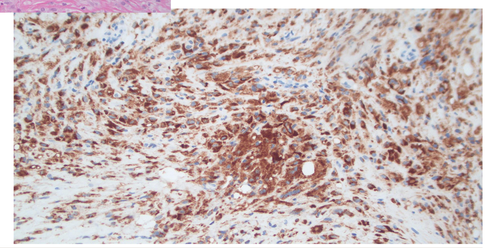

Figure 4. (Right) CD68 positive immunohistochemical stained image of the soft tissue mass

Analysis and Discussion

Although exceedingly rare, the majority of these lesions are self-limiting and resolve in the pediatric patient population. Two large case series have been performed for JXG. Dehner's study of 174 JXG lesions showed that only about 16% of masses were in the lower extremity, and four on the lower leg. With complete excisional biopsy, there was a 0% recurrence rate[4]. Janssen and Harms, in their study of 129 patients with JXG, showed only a 10% incidence on the lower extremity and only 2 patients with lesions on the sole of their foot. Their recurrence rate was 7% with total excision of the solitary masses. These recurrences were in the same surgical site and did not migrate[2]. More recently, there are scant case studies of juvenile xanthogranulomas on the soles of the feet, posterior heel and only one case subcutaneously of the 1st metatarsophalangeal joint[6-8].

In conclusion, we present a unique soft tissue tumor with a very unique presentation from the majority of JXG's in the literature. There was no dermatological component, the patient was slightly older than the average patient population, and the patient was relatively normolipidemic. We hope this adds to the body of literature and urge foot and ankle surgeons to use a medical team approach if they were to ever encounter one of these rare tumors.

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Financial Disclosures: None for this poster