CASE REPORT

Congenital Juvenile Xanthogranuloma of Foot, a Nodular Lesion: An Unusual Case in 2-month-old Infant

RAM AVTAR*, ONKAR KAUR†, ANAND K VERMA‡, RAJ KUMAR CHANDOKE#

ABSTRACT

A 2-month-old infant presented with a circumscribed nodule on left foot since birth. Excision biopsy showed juvenile xanthogranuloma, an uncommon diagnosis in an unusual site; common sites being head and neck. Uncommon sites are groin, genital organs, limbs and even internal organs. It carries a favorable prognosis.

Keywords: Juvenile xanthogranuloma, nodule, fibrohistiocytic tumor

Juvenile xanthogranuloma (JXG) is a disorder of non-Langerhan's cell group of histiocytic proliferative diseases.[1] It is an uncommon benign soft tissue lesion first reported by Adamson.[1] It presents as nodular or plaque like lesion in soft tissue sites. Although head and neck are common sites; it can involve all parts including groin, scrotum, genitals, trunk, and proximal part of limbs, even internal organs like lungs, liver and bone (Erdheim-Chester disease). It is present at birth in approximately 5-35% of total cases and rest usually present within 6-9 months of life.[2]

This case is being reported as it presented on dorsum left foot, which is a rare site and was present since birth.

CASE REPORT

A 2-month-old child presented with a well-defined globular swelling of size 3 × 3 cm over dorsum of left foot just distal to the ankle joint (Fig. 1). It was noted since birth and had remained stationary in size since then. There were no other similar lesions and no associated symptoms were present. Surface of the swelling was shiny although there were no signs of inflammation.[3]

Palpation of mass did not cause the infant any distress. It was firm in consistency with well-defined edges, non-fluctuant, nonpulsatile, not adherent to the skin and underlying structure. No associated lymphadenopathy was present. All routine blood investigations were within normal limits. On X-ray, no bony lesion was present. Magnetic resonance imaging report mentioned a well-defined rounded mass in the subcutaneous plain in dorsum of foot with low signal intensity on T_1-weighted images and high signal intensity on

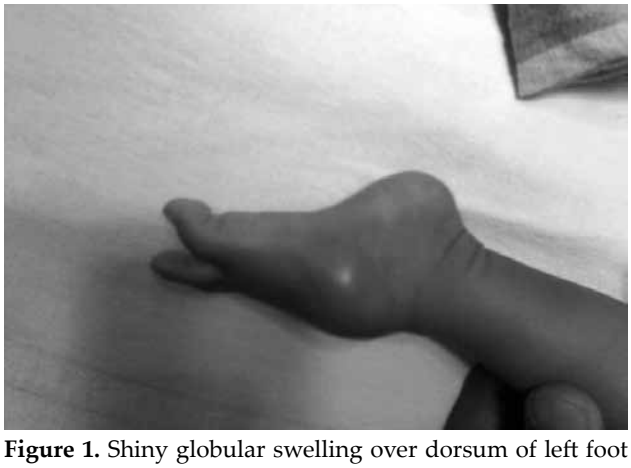

Figure 1. Shiny globular swelling over dorsum of left foot showing flexion of great toe.

*Head
Dept. of Orthopedics
†Senior Medical Officer
‡Senior Specialist
#Consultant and Head
Dept. of Pathology
ESI-PGIMSR and Model Hospital, Basaidarapur, New Delhi
Address for correspondence
Dr Raj Kumar Chandoke
Consultant and Head
Dept. of Pathology
ESI-PGIMSR and Model Hospital, Basaidarapur
Ring Road, New Delhi - 110 015
E-mail: drchandoke@gmail.com

T_2-weighted images, indicating no evidence of high flow vessels with signal voids on these sequences.

Aspiration cytology done from lesion showed scattered spindle cells with bland nuclei, foamy cells suggestive of benign mesenchymal lesion. Excision biopsy was advised for definitive nomenclature. Sharply defined noncapsulated globular mass of size 2.5 × 2.5 cm was excised from the subcutaneous plane. It was not involving any of the neighboring structures (Figs. 2 and 3). Microscopy revealed small fascicles composed of spindle-shaped cells with presence of inflammatory cells, plump foamy histiocytes and Touton giant cells. The findings were consistent with JXG (Figs. 4 and 5).

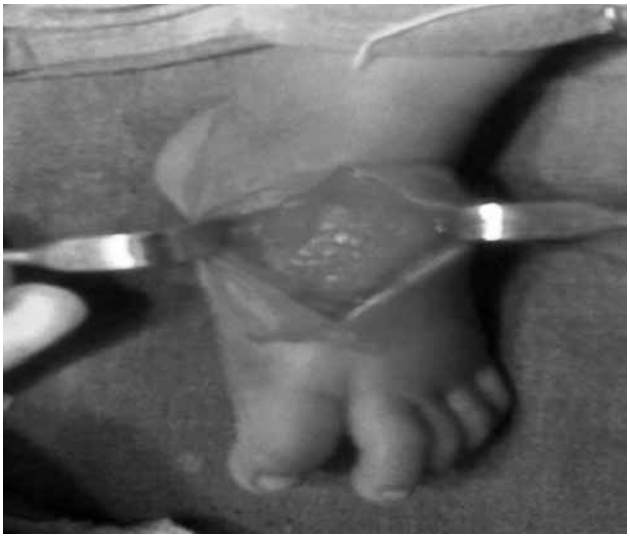

Figure 2. Operative photograph showing tumor.

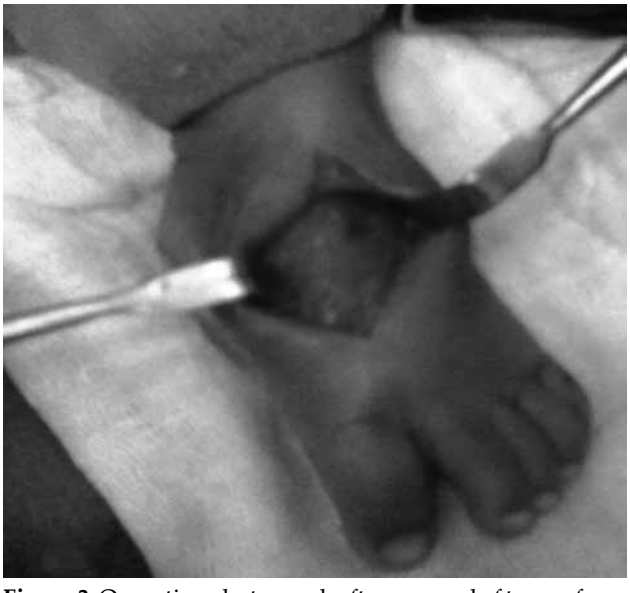

Figure 3. Operative photograph after removal of tumor from subcutaneous plane.

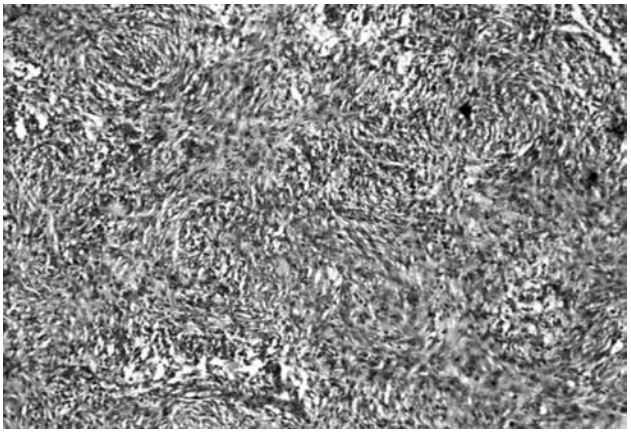

Figure 4. Soft tissue tumor: Fascicular arrangement of spindle cells and histiocytes (H&E 100x).

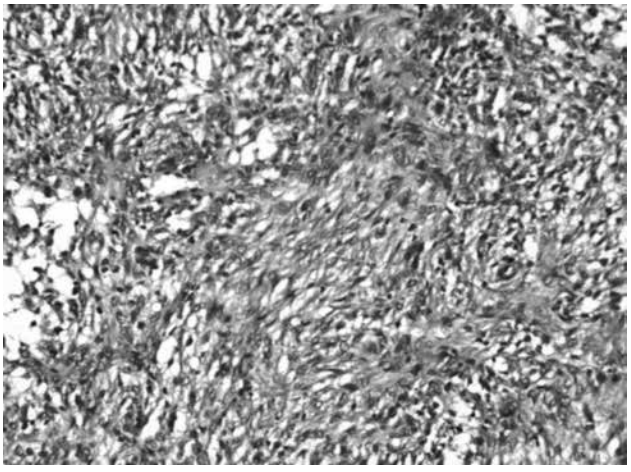

Figure 5. Foamy histiocytes and inflammatory cells (H&E 400x).

DISCUSSION

JXG is a benign fibrohistiocytic lesion accompanied by lipid deposits, first described by Adamson in 1905.[1] It is originally thought to be derived from dermal dendrocytes. Solitary or multiple red-brown papulonodules, 1-10 mm in diameter or larger are found on the head and neck, upper part of the trunk and proximal parts of the limbs.[2]

In the present case, the lesion was in dorsum of foot. Cutaneous presentation, being most common, may involve soft tissues as well as other organs.[3] In our case, there was no organ involvement. JXG has been documented in visceral organs such as lungs, bone, genitalia, gastrointestinal tract (GIT), heart, eyes and oral cavity.[3,4] JXG is a disease of young age with median age of onset within 6-9 months, but lesion may be present at birth in 5-35% of children. Children below 6 months present with multiple nodules in

head and neck region with male preponderance, which is higher in young infants.[1] Severe congenital systemic JXG has been reported in monozygotic twins.[2] JXG is associated with diseases like neurofibromatosis (NF-1), juvenile chronic myelogenous leukemia (CML), other hematological malignancies, Niemann-Pick disease and urticaria pigmentosa. This entity should be separated clinically by various differentials like hemangioma and NF (firm lesion, and with *café-au lait* spot).[3,5] Histopathological diagnosis is established on the basis of the nodule showing poorly demarcated dense histiocytic infiltrate involving the dermis and up to 85% cases infiltrating deep tissue in subcutis and sometimes skeletal muscles with variable number of Touton giant cells. In the present case, the lesion was excised from the subcutaneous plane. These plump foamy histiocytes are large, polygonal or spindle-shaped, with eosinophilic cytoplasm and positive for CD68, HAM 56, vimentin, lysozymes, α_1-antichymotrypsin, but negative for smooth muscle actin, CD34 and S100 protein.[6,7] Staining for these markers may be more intense at the periphery of the lesion. Electron microscopy (EM) shows lipid vacuole, lysosome, cholesterol clefts and myeloid bodies but no Birbeck granules. Other histological variants like spindle-cell xanthogranuloma with spindle-shaped histiocytes arranged in a storiform pattern, scalloped-cell xanthogranuloma with scalloped histiocytes with homogenous cytoplasm are known.[2]

JXG, although presents early in age, has good prognosis as it shows spontaneous regression in fair number of cases.[1,8,9] Pathogenesis being unknown, various theories like infection and physical stimulation have been implicated.[10-12] Although the etiology of JXG is unknown, cholesterol has been found to be the main lipid associated with it. Despite spontaneous regression, excision is required for an esthetic and diagnostic reason as was in the present case. Recurrences are uncommon.[6,12]

CONCLUSION

JXG, an uncommon entity of young children, can occur as early as at birth with nodule or plaque like presentation of any site. Biopsy confirmation and surgical excision should be done if no functional compromise has been assessed before. It is mostly cutaneous and prognostically favorable histiocytic tumor of infancy.

REFERENCES

1. Cypel TK, Zuker RM. Juvenile xanthogranuloma: case report and review of the literature. Can J Plast Surg. 2008;16(3):175-7.
2. Weedon D (Ed.). Weedon's Skin Pathology. 3rd edition, Churchill Livingstone Elsevier: USA. 2010:p.953-4.
3. Dehner LP. Juvenile xanthogranulomas in the first two decades of life: a clinicopathologic study of 174 cases with cutaneous and extracutaneous manifestations. Am J Surg Pathol. 2003;27(5):579-93.
4. Freyer DR, Kennedy R, Bostrom BC, Kohut G, Dehner LP. Juvenile xanthogranuloma: forms of systemic disease and their clinical implications. J Pediatr. 1996;129(2):227-37.
5. Kesavan TM, Sreedevi PK. Juvenile xanthogranuloma. Indian Pediatr. 2005;42(9):950.
6. Sonoda T, Hashimoto H, Enjoji M. Juvenile xanthogranuloma. Clinicopathologic analysis and immunohistochemical study of 57 patients. Cancer. 1985;56(9):2280-6.
7. Zelger B, Cerio R, Orchard G, Wilson-Jones E. Juvenile and adult xanthogranuloma. A histological and immunohistochemical comparison. Am J Surg Pathol. 1994;18(2):126-35.
8. Jung T, Emmert S, Günzl HJ, Neumann C, Rünger TM. Congenital manifestations of juvenile xanthogranuloma (large nodular form). Hautarzt. 2000;51(6):423-6.
9. Margulis A, Melin-Aldana H, Bauer BS. Juvenile xanthogranuloma invading the muscles in the head and neck: clinicopathological case report. Ann Plast Surg. 2003;50(4):425-8.
10. Chang MW. Update on juvenile xanthogranuloma: unusual cutaneous and systemic variants. Semin Cutan Med Surg. 1999;18(3):195-205.
11. Zvulunov A, Barak Y, Metzker A. Juvenile xanthogranuloma, neurofibromatosis, and juvenile chronic myelogenous leukemia. World statistical analysis. Arch Dermatol. 1995;131(8):904-8.
12. Cohen BA, Hood A. Xanthogranuloma: report on clinical and histologic findings in 64 patients. Pediatr Dermatol. 1989;6(4):262-6.