Congenital Giant Juvenile Xanthogranuloma in a 3-Month-Old Boy

A 3-month-old boy was seen for evaluation of rapidly growing cutaneous orange-yellowish nodular lesions of variable diameters (from millimeters to few centimeters) on the right wall of the chest that had been present since birth (**Figure 1**, A and B). The patient was otherwise completely healthy and did not have any trauma history. There was no personal or family history of any skin tumors or infectious diseases.

The chest ultrasound examination revealed a well-defined hypoechoic soft tissue mass with polycyclic margins (3 × 2.5 × 1 cm) and polylobulated within the subcutaneous tissue without extension into the underlying muscle (**Figure 2**, A). The lesion was not compressible. On color Doppler evaluation, there was evidence of many intralesional arterial and venous flow signals (**Figure 2**, B and **Video** [available at www.jpeds.com]). The diagnosis of high flow vascular anomaly was unlikely due to poor compressibility and high resistance indices of the lesion on velocimetric analysis. An abdominal ultrasound was negative for similar masses.

We excised the lesion and adjacent normal tissue. Histologic examination showed a poorly demarcated, dense infiltrate of histiocytes, admixed with Touton giant cells, lymphocytes, and eosinophils, involving the dermis and the upper subcutis. On the immunohistochemical study, the histiocytes were CD68+, FXIIIa+, S100-, and CD1a- (**Figure 3**). Given its size (enlarged in <6 weeks) and histopathologic analysis, the lesion was interpreted as a giant juvenile xanthogranuloma (JXG). Spontaneous regression was unlikely, and in view of the rapid growth, we were concerned that waiting would lead to cosmetic and functional problems.[1] Therefore, we performed an extended excision; the tumor did not recur after resection.

JXG, the most common non-Langerhans cell histiocytosis, is a benign self-limited histiocytic proliferative disorder characterized by small yellow-red nodules or papules.[1] The etiopathology is unknown.[2] The average age of onset is 2 years.[3] Classic JXG is divided into 2 main clinical subtypes: dome-shapes papules (<0.5 cm) and single/multiple nodules (<2 cm). A rare variant is referred to as giant (lesions larger than 2 cm). Cutaneous lesions are the most common form among classic JXG (67%-71%). The most common extracutaneous sites are subcutaneous or deep soft tissue (15%-16%); they most commonly involve the eye, followed by lung and liver manifestations (4%-5%).[2]

Classic JXG has been reported as congenital in 15%-20% of patients. Among giant cases, 45%-50% are congenital.[2,4] The course is self-limited over several months to years; it is rare for persistence beyond late childhood.[5]

The most common affected areas are the head, neck, and trunk, followed by the proximal extremities.[1] Histopathologic examination is essential to confirm the diagnosis.

JXG must be distinguished from infantile hemangioma. Hypoechogenicity because of fewer interfaces, lower vascular density, and higher arterial resistance indexes are found.[5]

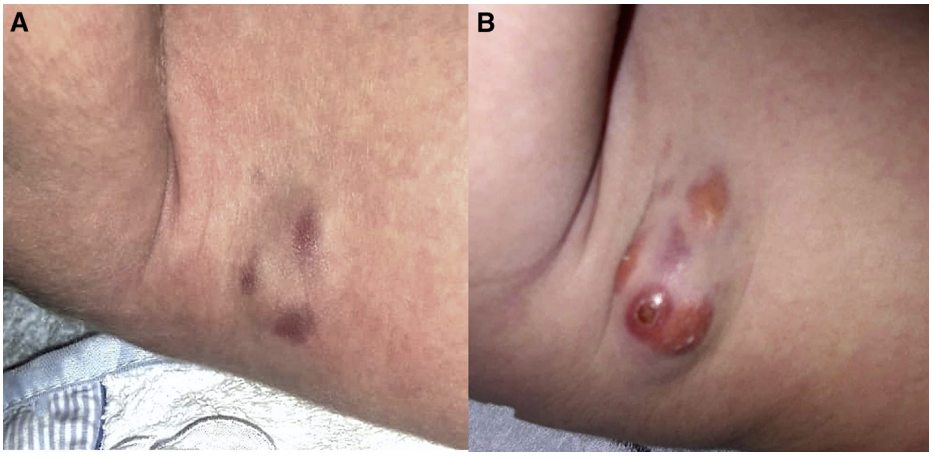

Figure 1. **A,** First clinical examination shows the presence of some millimetric reddish lesions on the right wall of the chest. **B,** Six weeks later, more prominent orange-yellowish nodular lesions with variable diameters and hard-elastic consistency are visible.

The authors declare no conflicts of interest.

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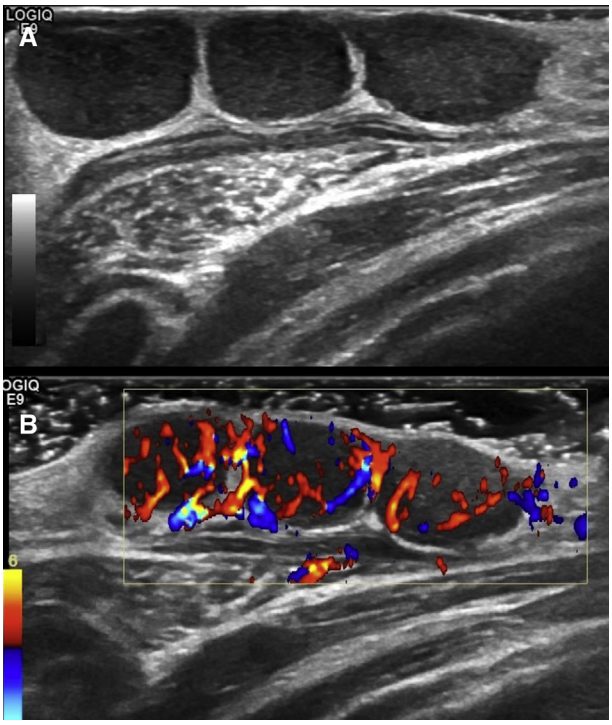

Figure 2. **A,** US examination shows a well-defined polylobulated hypoechoic soft tissue mass with polycyclic margins (3 × 2, 5 × 1 cm in diameter) within the subcutaneous tissue. The lesion is not well compressible. **B,** On color-Doppler evaluation, there was evidence of intralesional arterial and venous flow signals.

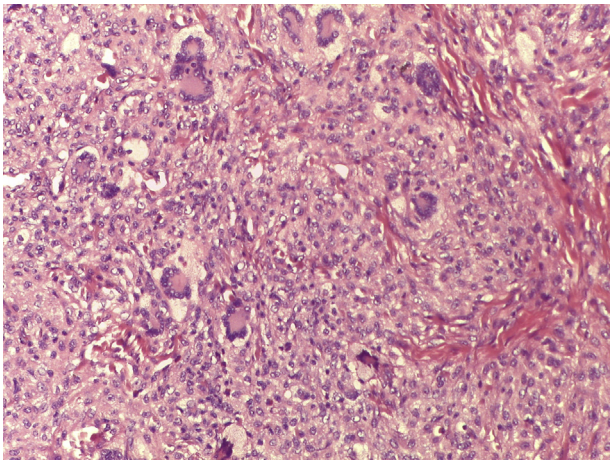

Figure 3. Histiocytes with lightly eosinophilic and foamy cytoplasm, admixed with Touton giant cells. H&E ×250.

Other nodular soft tissue lesions that should be considered are rhabdomyosarcoma, myofibromatosis, leukemia, neuroblastoma metastasis, giant cell fibroblastoma, juvenile nodular fasciitis, dermatofibrosarcoma protuberans, and other soft tissue neoplasms.[6] ■

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