Hyperpigmented nodule overlying the right Achilles tendon

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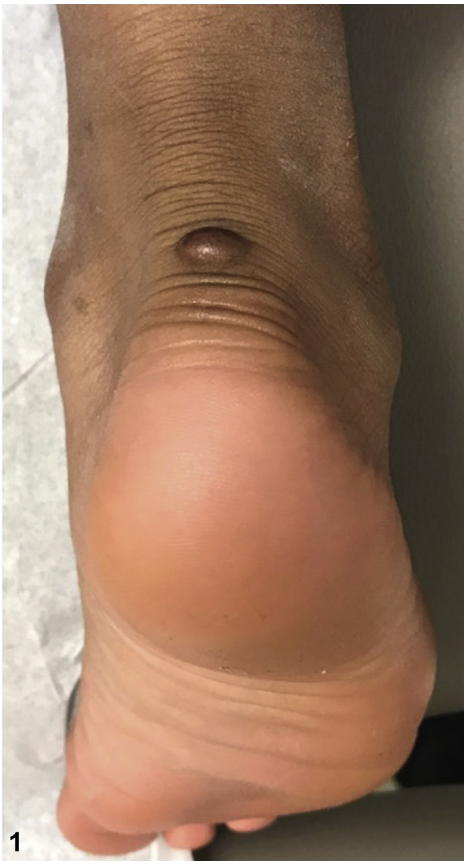

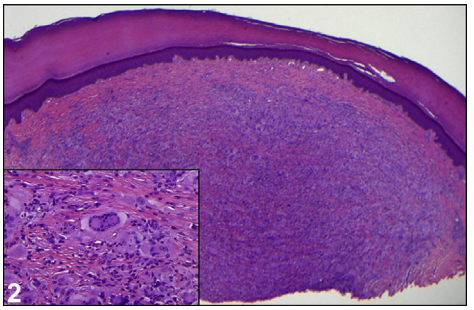

A 7-year-old otherwise healthy African-American girl with no significant medical history presented with a 6-month history of a slow-growing, nontender, nonpruritic, firm, hyperpigmented nodule overlying the right Achilles tendon (Fig 1). Stroking of the lesion did not produce any localized skin changes. There were other similar lesions noted on examination. Conjunctival examination and visual acuity were normal. The patient was developmentally normal without any focal neurologic deficits. Axillary and inguinal freckling were absent.

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Routine laboratory tests were within normal limits. There is no family history of hypercholesterolemia or early cardiac death. A shave biopsy of the entire lesion was performed (Fig 2) without recurrence.

Question 1: Which of the following is the most likely diagnosis?

A. Congenital self-healing reticulohistiocytoma (CSHR)

B. Erythema elevatum diutinum (EED)

C. Tendinous xanthoma

D. Cutaneous mastocytoma

E. Juvenile xanthogranuloma (JXG)

Answers:

A. CSHR — Incorrect. Although individual lesions of CSHR may present similarly to JXG, the histopathology helps clearly differentiate these two entities. On histopathology, CSHR displays a proliferation of Langerhans cells with minimal Touton giant cells. Clinically these lesions resolve spontaneously.

B. EED — Incorrect. EED is a chronic small vessel vasculitis that manifests as multiple papules, plaques, or nodules of varying color, most often on extensor surfaces. It is associated with infections, malignancies, and autoimmune disorders. On histopathology, there is often a grenz zone and a mixed inflammatory infiltrate with lymphocytes, neutrophils, eosinophils, and plasma cells. The presence of leukocytoclastic vasculitis is also common in this diagnosis.

C. Tendinous xanthoma — Incorrect. Tendinous xanthomas are smooth, firm, yellowish nodules overlying tendons and are associated with familial hypercholesterolemia syndromes. On histopathology, there is an abundance of foam cells with a mild inflammatory infiltrate.

D. Cutaneous mastocytoma — Incorrect. Mastocytomas typically occur in childhood and many spontaneously involute. Darier sign may be present with stroking of the lesion. On histopathology, there are numerous monotonous mast cells with fried egg appearance and occasional eosinophils in the papillary dermis.

E. JXG — Correct. JXG is a benign non-Langerhans cell histiocytosis that develops in early childhood. The most common manifestation is a solitary cutaneous nodule, but multiple lesions and extracutaneous involvement may be present. On histopathology, there are foamy histiocytes and Touton giant cells (Fig 2).

Question 2: What is the next best step in the management of this patient?

A. Ophthalmology referral

B. Reassurance

C. Bone marrow biopsy

D. Fasting lipid panel

E. Brain magnetic resonance imaging (MRI)

Answers:

A. Ophthalmology referral — Incorrect. Ophthalmologic screening for ocular involvement in a patient with a solitary cutaneous JXG is unnecessary. Survey studies have found that 0.3% of all patients with cutaneous JXG will have intraocular manifestations, and there is no current evidence that routine screening in children with cutaneous JXG will result in earlier diagnosis of ocular JXG. Ophthalmology referrals should be reserved for patients with risk factors for ocular JXG such as those with multiple skin lesions, those younger than 2 years of age, and those who are experiencing visual symptoms.[1]

B. Reassurance — Correct. Solitary JXG lesions are found to occur in up to 89% of cases with no other systemic manifestations. In this case, it is important to reassure the patient and the family that the lesion is benign.[1]

C. Bone marrow biopsy — Incorrect. A bone marrow biopsy may be indicated for workup of a suspected hematologic malignancy. This patient has no other concerning signs or symptoms for leukemia or lymphoma, and the complete blood count was normal.

D. Fasting lipid panel — Incorrect. JXG has no link to hypercholesterolemia, and a lipid panel would not be appropriate in this case.

E. Brain MRI — Incorrect. Although JXG can involve the central nervous system, this patient displays no focal neurologic deficits, motor or speech delays, or other neurologic sequelae that would warrant an MRI.[2]

Question 3: The additional presence of axillary and inguinal freckling would convey an increased risk for which of the following conditions?

A. Juvenile myelomonocytic leukemia (JMML)

B. Mast cell leukemia

C. Adrenal crisis

D. Cardiovascular disease

E. Vestibular schwannomas

Answers:

A. JMML — Correct. There is a triple association between JXG, neurofibromatosis (NF)-1 and JMML. The presence of axillary and inguinal freckling may suggest NF-1, which would give this patient a 20 to 32 times higher risk for JMML development, which portends a poor prognosis with a mean survival of about 4 years. Suspected patients should be referred to oncology for consideration of a bone marrow biopsy. Bone marrow transplantation is the only potentially curative therapy.[3]

B. Mast cell leukemia — Incorrect. Mast cell leukemia is an aggressive variant of systemic mastocytosis and is not usually associated with NF-1, which would be the suggested diagnosis if the patient had axillary and inguinal freckling.

C. Adrenal crisis — Incorrect. Increased pigmentation can be a sign of Addison disease or other causes of adrenal insufficiency, which can be complicated by adrenal crisis. The increased pigmentation in these conditions is usually more generalized with accentuation in the palmar creases along with potential mucosal involvement.

D. Cardiovascular disease — Incorrect. Cardiovascular disease would be more likely to occur in children with familial hypercholesterolemia and other risk factors such as diabetes and chronic kidney disease.

E. Vestibular schwannomas — Incorrect. NF-2 is associated with multiple central nervous system tumors, most commonly bilateral vestibular schwannomas that can lead to hearing loss. Axillary and inguinal freckling are more suggestive of NF-1.

Abbreviations used:

CSHR: congenital self-healing reticulohistiocytoma
EED: erythema elevatum diutinum
JMML: juvenile myelomonocytic leukemia
JXG: juvenile xanthogranuloma
MRI: magnetic resonance imaging
NF: neurofibromatosis

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