A 16-Year-Old Girl with Polyuria, Polydipsia, and a New-Onset Rash Around Her Elbows and Knees

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A 16-year-old girl presented with shortness of breath for 1 day, polyuria and polydipsia for 1 week, and a new-onset rash around her elbows and knees for 2 weeks. Clinical physical examination on presentation was significant for altered mental status, kussmaul pattern breathing, and presence of yellow papular lesions on the extensor surfaces of upper and lower extremities (Figure 1). Laboratory analysis showed acidosis (pH of 6.9), hyperglycemia (serum glucose of 450 mg/dL), elevated hemoglobin A1c (>14%), serum amylase of 612 u/L, serum lipase of 5,387 u/L, and serum triglycerides of 930 mg/dL. A biopsy of skin lesions showed dermal collection of foamy histiocytes with an infiltrate of neutrophils and lymphocytes, confirming that the lesions were xanthomas.

Figure 1. Yellowish, well-defined linear papular skin eruption seen on the extensor surfaces of (A) right knee and (B) left elbow, consistent with eruptive xanthomas.

For diagnosis, see page e318

Editor’s note: Each month, this department features a discussion of an unusual diagnosis. A description and images are presented, followed by the diagnosis and an explanation of how the diagnosis was determined. As always, your comments are welcome via email at pedann@Healio.com.
Diagnosis:
Eruptive Xanthoma as a Presenting Sign of New Onset Diabetes

Based on her clinical presentation, physical examination, and laboratory findings, a diagnosis of eruptive xanthomas, severe hypertriglyceridemia, diabetic ketoacidosis (DKA), and acute pancreatitis in the setting of new onset diabetes was made. Her hospital course was significant for initial worsening of serum triglycerides, which peaked at 2,515 mg/dL on day 2 of admission. Triglycerides then improved to 614 mg/dL with the insulin drip. She was discharged home on a low-fat diet and subcutaneous insulin (1 unit/kg per day). Her blood glucose control and triglyceride levels improved considerably with the intensive insulin regimen at home. The patient’s fasting triglyceride level was in the normal range (170 mg/dL) at the 1-month follow-up, and her xanthomas had resolved completely by the 6-month follow-up visit (Figure 2) without lipid-lowering medications.

DISCUSSION
Eruptive xanthomas are a cutaneous sign of excessive serum levels of triglyceride-rich, very low-density lipoproteins and chylomicrons. Uncontrolled diabetes mellitus (both type 1 and type 2) has been associated with eruptive xanthomas caused by insulin resistance and impaired clearance of triglycerides by lipoprotein lipase.1,2 Diet along with insulin therapy was sufficient for treating hypertriglyceridemia and resolution of xanthomas, avoiding the need for triglyceride-lowering medications such as fibrates, omega-3-fatty acids, or nicotinic acid.

REFERENCES

Figure 2. Residual mild hyperpigmentation with resolution of xanthomas on extensor surface of (A) right knee and (B) left elbow at the 6-month follow-up visit.