VASCULAR DISEASE, VASCULITIS

GENERALIZED NET-LIKE ERYTHEMA IN AN ADOLESCENT: ONLY CLUE TO DIAGNOSIS OF ADA 2 DEFICIENCY

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Background: Deficiency of adenosine deaminase type 2 (DADA2) is a recently described autoinflammatory disorder. It is a rare vasculopathy characterized mainly by widespread livedo racemosa, recurrent low-grade fever, neurological and gastrointestinal symptoms.

Observation: 15-year-old boy presented with a 3-year history of generalized rash. He did not report any associated symptoms. Dermatological examination revealed irregular net-like violaceous erythema on trunk and extremities. His past medical history was insignificant. On the basis of the cutaneous findings, suspicion was raised for a vasculitis or vasculopathy. The patient's WBC count was 7.0 x 109/l, Hgb 13.1 g/dl and the platelet count 327x 109/l. The erythrocyte sedimentation rate (ESR) level was 37 mm/h (normal 0-15).
Antinuclear antibodies, anti double-stranded DNA antibody, anticardiolipin antibodies, complement C3 and C4 levels were all unremarkable. Ultrasound revealed no hepatosplenomegaly.
Histopathological examination revealed leukocytoclastic vasculitis involving small vessels. Diagnosis of DADA2 was confirmed by CECR1 gene sequencing.
Treatment with adalimumab was initiated. Clinical and laboratory follow-up of the patient did not show any abnormalities since 1 year.

Key message: Dermatologists should be able to recognize DADA2 disease before systemic manifestations become obvious. Early diagnosis, thus prompt treatment initiation is crucial in DADA2 to decrease mortality. Delays in diagnosis may lead to high morbidity and potentially fatal consequences such as neurologic events.