A rare case of an inherited vasculopathy: CADASIL in a skin biopsy

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Abstract

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is a rare inherited vascular disorder thought to be caused by pathogenic mutations in the NOTCH3 gene, which is critical for vascular smooth muscle cell differentiation and normal vascular development. Abnormal accumulation of a portion of the NOTCH3 gene product in the vessel wall lead to structural and functional abnormalities, causing damage in affected organs. Although the clinical symptoms of CADASIL are primarily neurological, resulting from white matter infarcts, it is a systemic disease that affect small arteries and arterioles. Definitive diagnosis requires clinicopathologic correlation and genetic testing, but electron microscopy can identify characteristic ultrastructural changes in vessel walls. We describe a case of CADASIL identified on a skin biopsy.