



LECTURE

Genetic causes of cerebral small vessel disease

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Abstract

Hereditary causes of cerebral small vessel disease (SVD) are rare but important to recognize in patients suffering from stroke or vascular dementia. These include various diseases with particular clinical and imaging features which are helpful in driving the clinician to a proper molecular diagnosis. Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is characterized by migraine with aura, ischaemic cerebrovascular events, dementia and a conspicuous pattern of white matter changes. A second type has been described by a mutation in the gene HTRA1. The same gene is responsible for Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL), which presents concomitantly with lumbago and alopecia. More

recently, Cathepsin-A-related arteriopathy with strokes and leukoencephalopathy (CARASAL) has been described. COL4A1/A2-related cerebral SVD causes cerebral lacunar infarcts as well as intracerebral haemorrhage. Autosomal dominant retinal vasculopathy with cerebral leukodystrophy is caused by a mutation in TREX1 and shows a more systemic involvement of kidney and retina. Familial forms of cerebral amyloid angiopathy present more frequently with lobar haemorrhage. Lastly, Fabry Disease is caused by mutation of the gene encoding alfa-galactosidase A and distinguishes itself from the other genetic SVD by the fact that it is X-linked. Common characteristics are painful acroparaesthesia, angiokeratomas, kidney failure and suggestive ophthalmological findings.

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