DISEASE:
COL4A1-related familial vascular leukoencephalopathy

NAME:	COL4A1-related familial vascular leukoencephalopathy
DESCRIPTION:	COL4A1-related familial vascular leukoencephalopathy is a rare, genetic, neurological disease characterized by the presence of fragile small-vessel intracerebral vasculature in various members of a single family, manifesting, clinically, with single or recurrent hemorrhagic and/or ischemic stroke and, frequently, ocular and renal involvement. Neuroimaging reveals diffuse, periventricular leukoencephalopathy associated with dilated perivascular spaces, lacunar infarction and microhemorrhages.
ORPHACODE:	36383
SYNONYMS:	COL4A1-related brain small vessel disease with hemorrhage COL4A1-related retinal arteriolar tortuosity-infantile hemiparesis-autosomal dominant leukoencephalopathy syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	COL4A1
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- collagen type IV alpha 1 chain

Related Gene Panels

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG

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