DISEASE:
Retinal arterial tortuosity

NAME:	Retinal arterial tortuosity
DESCRIPTION:	A rare genetic cerebral small vessel disease characterized by isolated marked tortuosity of second-order and third-order retinal arteries with normal first-order arteries and venous system, typically located in the macular and peripapillary area and developing during childhood or early adulthood. The disease may be asymptomatic, although most patients present variable degrees of transient vision loss due to retinal hemorrhage following physical exertion or minor trauma.
ORPHACODE:	75326
SYNONYMS:	Familial isolated retinal arterial tortuosity Retinal arteriolar tortuosity Retinal hemorrhage with vascular tortuosity Tortuosity of retinal arteries
XREF(S):	Orphanet
ANALYTE(S):	COL4A1
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