Evolution of brain lesions in a patient with *TREX1* cerebroretinal vasculopathy

Figure 1 An irregular peripherally enhancing mass in the right frontal lobe with extensive vasogenic edema (2008)

A 44-year-old woman with a history of retinal vasculopathy presented with headaches and hemiparesis. MRI brain showed a right frontal peripherally enhancing mass with surrounding edema (figure 1) thought to represent a malignant glioma; however, pathology showed necrosis with reactive gliosis but no evidence of neoplasm (figure 2). Family history was significant for multiple relatives with similar lesions on head MRIs suspicious for neoplasm but excluded by biopsy. An underlying familial leukoencephalopathy was suspected, and a novel heterozygous nonsense mutation c.1359G>T; p.Glu285* was identified in *TREX1*, in which C-terminal frameshift mutations cause autosomal dominant retinal vasculopathy with cerebral leukodystrophy.¹

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Figure 2 Serial MRIs over 6 years (2008–2014) show increasing confluent fluid-attenuated inversion recovery hyperintensity in bifrontal and right periatrinal white matter

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1. Richards A, van den Maagdenberg AM, Jen JC, et al. C-terminal truncations in human 3'-5' DNA exonuclease TREX1 cause autosomal dominant retinal vasculopathy with cerebral leukodystrophy. *Nat Genet* 2007;39:1068–1070.

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