

Figure e-1: (A) The family pedigree. Genetic sequencing results are provided for parents, index case and siblings. (B and C) Axial T2-weighted MRI scans through the lateral ventricles and the top of the lateral ventricles at presentation show a largely cortical acute left ACA and MCA territory infarct (arrows) and a mature right MCA infarction involving the cortex, white matter and basal ganglia. (D) The ADC map corresponding to the slice in (A) confirms the left sided infarct is acute – dark on ADC map (arrows). (E), (F) Axial T2-weighted MRI 6 years later demonstrate maturation of the left MCA infarct and extension of the right MCA infarct (arrows). ACA = anterior cerebral artery, MCA = middle cerebral artery, ADC = apparent diffusion coefficient.

