BRIEF COMMUNICATIONS

PHENOTYPIC VARIABILITY IN A MEXICAN MESTIZO FAMILY WITH RETINAL VASCULOPATHY WITH CEREBRAL LEUKODYSTROPHY AND TREX1 MUTATION p.V235Gfs*6

NANCY MONROY-JARAMILLO, AURELIO CERÓN, ELIZABETH LEÓN, VERÓNICA RIVAS, ADRIANA OCHOA-MORALES, MARÍA GEORGINA ARTEAGA-ALCARAZ, FAUSTO CARLOS NOCEDAL-RUSTRIAN, CECILIA GALLEGOS, MARÍA ELISA ALONSO-VILATELA, TERESA CORONA, JOSÉ FLORES

1Department of Neurogenetics, 2Department of Neurology and 3Clinical Laboratory of Neurodegenerative Diseases, Instituto Nacional de Neurología y Neurocirugía Manuel Velasco Suárez, Mexico City; 4State Research Coordination, Mexican Social Security Institute (IMSS), Pachuca, Hgo.; 5Hospital Regional #25 Ignacio Zaragoza, Mexican Social Security Institute, Mexico City; 6Department of Pathology, Hospital Ángeles del Pedregal, Mexico City, Mexico

Figure S1. Brain biopsy of Case 2. Images obtained from brain biopsy (objective ×40), stained with hematoxylin and eosin. The presence of brain parenchyma with fibrinoid thrombosis is observed in the specimen.
Figure S2. The TREX1 mutation identified in a Mexican Mestizo genealogy with retinal vasculopathy with cerebral leukodystrophy. At the top of the figure are the electropherograms of the wild-type sequence (WT) and the heterozygous p.V235fs mutation (MUT), indicated by an arrow. At the bottom, the comparison of partial wild-type and mutant nucleotide and protein sequences showing the consequences of the frameshift mutation. An arrow above the sequence of nucleotides indicates the insertion c.703_704insG; p.V235Gfs*6. NM_033629 was used as reference sequence.