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Severe progressive brain involvement in a patient with TRMT10C mutation

Comprometimento cerebral progressivo grave em paciente com mutação no gene TRMT10C

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A 2-month-old girl presented hypotonia, followed by progressive cognitive and motor deterioration, pyramidal signs, hearing loss, refractory epilepsy, and high serum lactate level. A biceps brachii biopsy presented cytochrome c oxidase negative fibers, and serial brain magnetic resonance imaging (MRI) showed progressive brain involvement (Figure 1). Whole-exome sequencing showed the homozygous pathogenic variant c.542G>T (p.Arg181Leu) in *TRMT10C*. The nuclear gene *TRMTC10C* encodes RNase P protein responsible for mt-tRNA maturation and causes an autosomal recessive mitochondrial disease[1]. To our knowledge, variants in *TRMT10C* have only been described once in two patients presenting hypotonia, lactic acidosis, and sensorineural deafness, but the brain imaging progression pattern was never depicted[2].

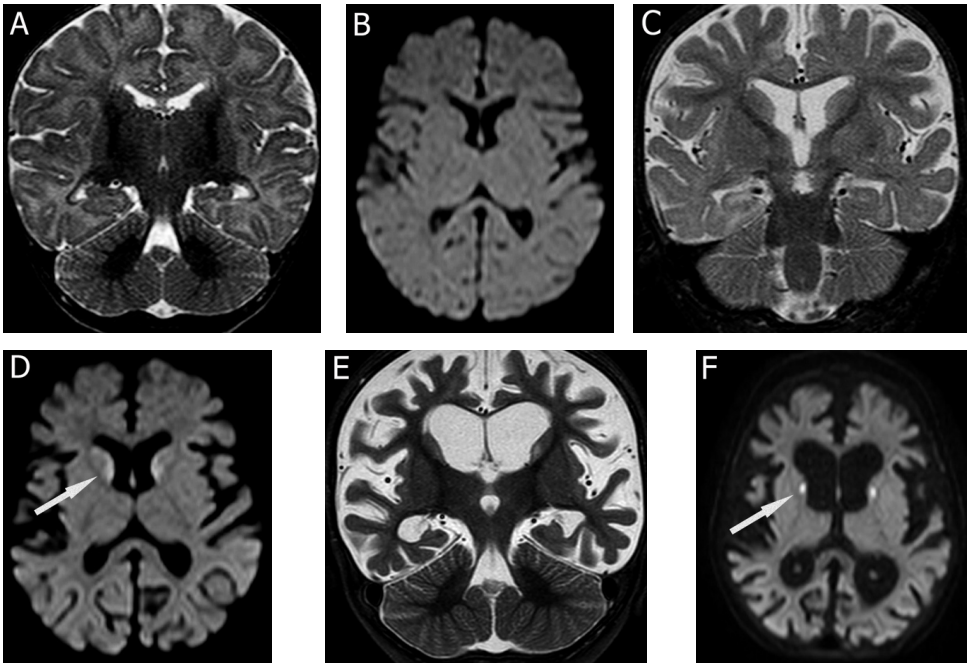

Figure 1. Comparative images for follow-up demonstrating rapidly progressive generalized brain atrophy. Comparative coronal images on fluid attenuated inversion recovery [FLAIR] (A, C and E) and Axial plane images on T2 FSE sequence (B, D, and F) were obtained when the patient was 3, 8, and 13 months of age, respectively, showing predominantly involvement of supratentorial regions, particularly affecting the white matter and superficial cortex with progressive enlargement of sulci and ventricular dilation. Deep gray matter has less evident atrophy with selective abnormal basal ganglia T2/FLAIR hyperintensity (arrows). This striatal abnormality has particularly involved the head of caudate nuclei and anterior parts of putamen, and was more evident in the last examination.

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