

Seven Year Old Boy with Abnormal Behavior - Methylmalonic Aciduria and Homocystinuria

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Mutations in the methylmalonic aciduria and homocystinuria type c.394C> T (p.R 132X) can cause a defect in B₁₂ metabolism, which could lead to neuropsychiatric disorder.

We report a seven year old boy with abnormal behavior. Brain MRI showed T2 and FLAIR hyperintensities in the cerebral white matter.

The patient methylmalonic aciduria and homocystinuria type c gene was positive for c.394C> T (p.R 132X). The clinical symptoms improved after Cobalamin replacement therapy.

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