Teaching NeuroImage: New Pattern of Periventricular Nodular Heterotopia in Twins With a Pathogenic Variant in the MAP1B Gene

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Prior reports of variants associated with the MAP1B gene in periventricular nodular heterotopia (PVNH) patients have described frontally predominant PVNH and perisylvian polymicrogyria, with clinical manifestations of seizures and cognitive impairment.\textsuperscript{1}

We report a family with PVNH related to a variant in the MAP1B gene. The 8-year-old index girl was admitted to our hospital for intractable epilepsy presenting as unresponsiveness, behavioral arrest, blank staring, and oral automatisms lasting for about 30 seconds. She had age-appropriate motor skills but mild intellectual disability (ID).

Exome analysis revealed a heterozygous missense variant in MAP1B (NM_005909), c.6230G>A (p.R2077H) in the index patient, her twin sister, and their mother.

Unlike the imaging pattern previously reported, the MRI showed gray matter heterotopia along the trigones and occipito-temporal horns. Additional unique features of this case included bilateral frontal dominant pachygyria, alongside subcortical band heterotopia belonging to the Lissencephaly spectrum\textsuperscript{2} seen in both twins (figure.1).

The seizure type, ID, and neuroimaging phenotype were identical in her twin sister at the age of 9. Their mother had the same mutation without seizures. Their grandmother had both the
epileptic phenotype and mild ID since childhood (figure 2).


Figure 1. Brain magnetic resonance imaging (FLAIR sequence) of the index patient (A-C) and her twin sister (D-F). The images showed bilateral periventricular heterotopia (white arrows), pachygyria (white arrowheads) and subcortical band heterotopia (SBH) (yellow arrows).
**Figure 2. Pedigree diagram:** epilepsy (vertical line), MAP1B mutation (NM_005909), c.6230G>A (p.R2077H) (horizontal lines)
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