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Confirming the recessive inheritance of SCN1B mutations in developmental epileptic encephalopathy

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Dominant SCN1B (Sodium channel subunit beta-1) mutations are known to cause several epilepsy syndromes in humans. Only two epilepsy patients to date have been reported to have recessive mutations in SCN1B as the likely cause of their phenotype. Here, we confirm the recessive inheritance of two novel SCN1B mutations in five children from three families with developmental epileptic encephalopathy. The "negative" clinical exome in one of these families highlight the need to consider recessive mutations in the interpretation of variants in typically dominant genes.

Biography

Wafaa Ramadan has completed her MBBS this June from AlFaisal University, Riyadh, KSA.

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