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Variable clinical expression in patients with mosaicism for KCNQ2 mutations

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Titre	Variable clinical expression in patients with mosaicism for KCNQ2 mutations
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Auteur	Milh, Mathieu [1], Lacoste, Caroline [2], Cacciagli, Pierre [3], Abidi, Affef [4], Sutera-Sardo, Julie [5], Tzelepis, Ilias [6], Colin, Estelle [7], Badens, Catherine [8], Afenjar, Alexandra [9], Coeslier, Anne Dieux [10], Dailland, Thomas [11], Lesca, Gaetan [12], Philip, Nicole [13], Villard, Laurent [14]
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Résumé en anglais	Mutations in the KCNQ2 gene, encoding a potassium channel subunit, were reported in patients presenting epileptic phenotypes of varying severity. Patients affected by benign familial neonatal epilepsy (BFNE) are at the milder end of the spectrum, they are affected by early onset epilepsy but their subsequent neurological development is usually normal. Mutations causing BFNE are often inherited from affected parents. Early infantile epileptic encephalopathy type 7 (EIEE7) is at the other end of the severity spectrum and, although EIEE7 patients have early onset epilepsy too, their neurological development is impaired and they will present motor and intellectual deficiency. EIEE7 mutations occur de novo. Electrophysiological experiments suggested a correlation between the type of mutation and the severity of the disease but intra and interfamilial heterogeneity exist. Here, we describe the identification of KCNQ2 mutation carriers who had children affected with a severe epileptic phenotype, and found that these individuals were mosaic for the KCNQ2 mutation. These findings have important consequences for genetic counseling and indicate that neurological development can be normal in the presence of somatic mosaicism for a KCNQ2 mutation.
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