

IonNeuroNet publication list

Benign Infantile Seizures and Paroxysmal Dyskinesia Caused by an SCN8A Mutation

Ann Neurol. 2016 Mar; 79(3):428-36.

Elena Gardella, MD, PhD, Felicitas Becker, MD, Rikke S. Møller, PhD, Julian Schubert, PhD, Johannes R. Lemke, MD, Line H. G. Larsen, MSc, Hans Eiberg, PhD, Michael Nothnagel, PhD, Holger Thiele, MD, Janine Altmüller, PhD, Steffen Syrbe, MD, Andreas Merkenschlager, MD, Thomas Bast, MD, Bernhard Steinhoff, MD, Peter Nürnberg, MD, Yuan Mang, MSc, Louise Bakke Möller, MSc, Pia Gellert, MD, Sarah E. Heron, PhD, Leanne M. Dibbens, PhD, Sarah Weckhuysen, MD, PhD, Hans Atli Dahl, PhD, Saskia Biskup, MD, PhD, Niels Tommerup, PhD, Helle Hjalgrim, MD, PhD, Holger Lerche, MD, Sándor Beniczky, MD, PhD, and Yvonne G. Weber, MD

De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy

Nat Genet. 2015 Apr;47(4):393-9. Epub 2015 Mar

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Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes

Nat Genet. 2014 Dec;46(12):1327-32. doi: 10.1038/ng.3130. Epub 2014 Nov 2

Schubert J, Siekierska A, Langlois M, May P, Huneau C, Becker F, Muhle H, Suls A, Lemke JR, de Kovel CG, Thiele H, Konrad K, Kawalia A, Toliat MR, Sander T, Rüschendorf F, Caliebe A, Nagel I, Kohl B, Kecskés A, Jacmin M, Hardies K, Weckhuysen S, Riesch E, Dorn T, Brilstra EH, Baulac S, Møller RS, Hjalgrim H, Koeleman BP; EuroEPINOMICS RES Consortium, Jurkat-Rott K, Lehman-Horn F, Roach JC, Glusman G, Hood L, Galas DJ, Martin B, de Witte PA, Biskup S, De Jonghe P, Helbig I, Balling R, Nürnberg P, Crawford AD, Esguerra CV, Weber YG, Lerche H.

Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes

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Potassium channel genes and benign familial neonatal epilepsy

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