Cambridge Elements

Elements in Genetics in Epilepsy
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SCN2A-RELATED DISORDERS

Edited by

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Abstract: *SCN2A* encodes a voltage-gated sodium channel (designated $Na_V1.2$) vital for generating neuronal action potentials (APs). Pathogenic *SCN2A* variants are associated with a diverse array of neurodevelopmental disorders featuring neonatal or infantile onset epilepsy, developmental delay, autism, intellectual disability, and movement disorders. This remarkable clinical heterogeneity is mirrored by extensive allelic heterogeneity and complex genotype–phenotype relationships partially explained by divergent functional consequences of pathogenic variants. Emerging therapeutic strategies targeted to specific patterns of $Na_V1.2$ dysfunction offer hope for improving the lives of individuals affected by *SCN2A*-related disorders. This Element provides a review of the clinical features, genetic basis, pathophysiology, pharmacology, and treatment of these genetic conditions authored by leading experts in the field and accompanied by perspectives shared by affected families. This title is also available as Open Access on Cambridge Core.

Keywords: sodium channel, epilepsy, epileptic encephalopathy, neonatal seizures, epilepsy genetics, autism spectrum disorder, neurodevelopmental disorder, intellectual disability, precision medicine

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