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Whole exome sequencing identifies a heterozygous missense variant in the GABRB3 gene in a patient with Dravet syndrome

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Background: Dravet syndrome is a rare and severe type of epilepsy in infants. Approximately 70-80% of DS cases are caused by mutations in *SCN1A*, the gene encoding the alpha-1 subunit of the sodium channel, while some proradic cases would have variants in several other genes including but not limited to *PCDH19, GABRG2, SCN1B, SCN9A* and *CHD2.*

Purpose & Methods: We performed whole-exome sequencing in 6 SCN1A-negative patients with Dravet syndrome in order to identify other related genes for this disorder. The exome sequencing libraries of 14 individuals, including 4 parent–proband trios and 2 unrelated probands were prepared using the SureSelectXT Library Prep Kit and the obtained libraries were sequenced on an Illumina HiSeq 4000. The candidate variants were interpreted and classified according to the American College of Medical Genetics and Genomics (ACMG) standards and guidelines.

Results: In one affected individual, we detected a novel de novo heterozygous missense mutation p.Arg232Gln in *GABRB3*, the gene encoding the β3-subunit of the gamma-aminobutyric acid type A (GABAA) receptor, which mediates inhibitory signaling within the central nervous system. Furthermore, a heterogeneous *SCN1A* variant p.Arg393His that had been undetected by previous Sanger sequencing was revealed in another patient, whose father was mosaic to the variant.

Conclusion: Our result extended the genetic basis of Dravet syndrome and confirmed the utility of whole-exome sequencing in genetic diagnosis.

Biography

Do Thi Thu Hang has obtained her PhD in Pharmacy in Sungkyunkwan University, South Korea in 2009 and did Postdoctoral training in Molecular and Cellular Immunology at QIA, South Korea and then at Deakin University, Australia from 2009-2012. From 2005-2009, her research focused on molecular mechanisms underlying pathogenesis of Alzheimer's disease and H1N1 influenza virus infection. She has returned to Vietnam in 2012 and started her new research on genetics of severe epilepsy syndromes, especially of Dravet syndrome. She is interested in applying next-generation sequencing for research and clinical molecular diagnostics.

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