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Journal articles

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**Title:** Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia.

**Citation:** American journal of human genetics, Apr 2016, vol. 98, no. 4, p. 615-626, 1537-6605 (April 7, 2016)


**Abstract:** Glycosylphosphatidylinositol (GPI) is a glycolipid that anchors >150 various proteins to the cell surface. At least 27 genes are involved in biosynthesis and transport of GPI-anchored proteins (GPI-APs). To date, mutations in 13 of these genes are known to cause inherited GPI deficiencies (IGDs), and all are inherited as recessive traits. IGDs mainly
manifest as intellectual disability, epilepsy, coarse facial features, and multiple organ anomalies. These symptoms are caused by the decreased surface expression of GPI-APs or by structural abnormalities of GPI. Here, we present five affected individuals (from two consanguineous families from Egypt and Pakistan and one non-consanguineous family from Japan) who show intellectual disability, hypotonia, and early-onset seizures. We identified pathogenic variants in PIGG, a gene in the GPI pathway. In the consanguineous families, homozygous variants c.928C>T (p.Gln310(*)) and c.2261+1G>C were found, whereas the Japanese individual was compound heterozygous for c.2005C>T (p.Arg669Cys) and a 2.4 Mb deletion involving PIGG. PIGG is the enzyme that modifies the second mannose with ethanolamine phosphate, which is removed soon after GPI is attached to the protein. Physiological significance of this transient modification has been unclear. Using B lymphoblasts from affected individuals of the Egyptian and Japanese families, we revealed that PIGG activity was almost completely abolished; however, the GPI-APs had normal surface levels and normal structure, indicating that the pathogenesis of PIGG deficiency is not yet fully understood. The discovery of pathogenic variants in PIGG expands the spectrum of IGDs and further enhances our understanding of this etiopathogenic class of intellectual disability. Copyright 2016 The American Society of Human Genetics. Published by Elsevier Inc. All rights reserved.

Source: Medline

Title: Implementation of an evidence-based seizure algorithm in intellectual disability nursing: A pilot study

Citation: Journal of Intellectual Disabilities, March 2016, vol./is. 20/1(55-64), 1744-6295;1744-6309 (March 2016)

Author(s): Auberry K., Cullen D.

Language: English

Abstract: Based on the results of the Surrogate Decision-Making Self Efficacy Scale (Lopez, 2009a), this study sought to determine whether nurses working in the field of intellectual disability (ID) experience increased confidence when they implemented the American Association of Neuroscience Nurses (AANN) Seizure Algorithm during telephone triage. The
results of the study indicated using the AANN Seizure Algorithm increased self-confidence for many of the nurses in guiding care decisions during telephone triage. The treatment effect was statistically significant \(-3.169(p < 0.01)\) for a small sample of study participants. This increase in confidence is clinically essential for two reasons. Many individuals with ID and epilepsy reside within community-based settings. ID nurses provide seizure guidance to this population living in community-based settings via telephone triage. Evidenced-based training tools provide a valuable mechanism by guiding nurses via best practices. Nurses may need to be formally trained for seizure management due to high epilepsy rates in this population.

**Publication Type:** Journal: Article

**Source:** EMBASE

**Full Text:**
Available from Highwire Press in *Journal of Intellectual Disabilities*

**Title:** Effectiveness of antiepileptic therapy in patients with PCDH19 mutations

**Citation:** Seizure, February 2016, vol./is. 35/(106–110), 1059-1311;1532-2688 (01 Feb 2016)


**Language:** English

**Abstract:** Purpose PCDH19 mutations cause epilepsy and mental retardation limited to females (EFMR) or Dravet-like syndromes. Especially in the first years of life, epilepsy is known to be highly pharmacoresistant. The aim of our study was to evaluate the effectiveness of antiepileptic therapy in patients with PCDH19 mutations. Methods We report a retrospective multicenter study of antiepileptic therapy in 58 female patients with PCDH19 mutations and epilepsy aged 2-27 years (mean age 10.6 years). Results The most effective
drugs after 3 months were clobazam and bromide, with a responder rate of 68% and 67%, respectively, where response was defined as seizure reduction of at least 50%. Defining long-term response as the proportion of responders after 12 months of treatment with a given drug in relation to the number of patients treated for at least 3 months, the most effective drugs after 12 months were again bromide and clobazam, with a long-term response of 50% and 43%, respectively. Seventy-four percent of the patients became seizure-free for at least 3 months, 47% for at least one year. Significance The most effective drugs in patients with PCDH19 mutations were bromide and clobazam. Although epilepsy in PCDH19 mutations is often pharmaco-resistant, three quarters of the patients became seizure-free for at least for 3 months and half of them for at least one year. However, assessing the effectiveness of the drugs is difficult because a possible age-dependent spontaneous seizure remission must be considered.

**Publication Type:** Journal: Article

**Source:** EMBASE