Lessons learned from 40 novel PIGA patients and

Epilepsia
61, 1142-1155
DOI: 10.1111/epi.16545

Citation Report

#	Article	IF	Citations
1	Investigating Developmental and Epileptic Encephalopathy Using Drosophila melanogaster. International Journal of Molecular Sciences, 2020, 21, 6442.	1.8	14
2	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	0.8	15
3	Early-Onset Developmental and Epileptic Encephalopathies of Infancy: An Overview of the Genetic Basis and Clinical Features. Pediatric Neurology, 2021, 116, 85-94.	1.0	28
4	Congenital Disorders of Glycosylation from a Neurological Perspective. Brain Sciences, 2021, 11, 88.	1.1	53
5	Deep-Phenotyping the Less Severe Spectrum of PIGT Deficiency and Linking the Gene to Myoclonic Atonic Seizures. Frontiers in Genetics, 2021, 12, 663643.	1.1	6
6	Migrating Focal Seizures and Myoclonic Status in ARV1- Related Encephalopathy. Neurology: Genetics, 2021, 7, e593.	0.9	6
7	Diaphragmatic Hernia as a Prenatal Feature of Glycosylphosphatidylinositol Biosynthesis Defects and the Overlap With Fryns Syndrome â€“ Literature Review. Frontiers in Genetics, 2021, 12, 674722.	1.1	4
8	PIGQ-Related Glycophosphatidylinositol Deficiency Associated with Nonprogressive Congenital Ataxia. Cerebellum, 2022, 21, 525-530.	1.4	2
9	Liver Involvement in Congenital Disorders of Glycosylation: A Systematic Review. Journal of Pediatric Gastroenterology and Nutrition, 2021, 73, 444-454.	0.9	2
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11	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. Genes, 2021, 12, 1051.	1.0	36
12	Congenital Disorders of Glycosylation: What Clinicians Need to Know?. Frontiers in Pediatrics, 2021, 9, 715151.	0.9	21
13	Spectrum of Neurological Symptoms in Glycosylphosphatidylinositol Biosynthesis Defects: Systematic Review. Frontiers in Neurology, 2021, 12, 758899.	1.1	2
14	Pyridoxine or pyridoxalâ€5â€phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study. Developmental Medicine and Child Neurology, 2022, 64, 789-798.	1.1	6
15	Constitutional PIGA mutations cause a novel subtype of hemochromatosis in patients with neurologic dysfunction. Blood, 2022, 139, 1418-1422.	0.6	8
17	PRENATAL ULTRASOUND FINDINGS ASSOCIATED WITH PIGW VARIANTS:: ONE MORE PIECE IN THE FRYNS SYNDROME PUZZLE? PIGW â€related prenatal findings. Prenatal Diagnosis, 0, , .	1.1	1
18	The correlation between multiple congenital anomalies hypotonia seizures syndrome 2 and PIGA: a case of novel PIGA germline variant and literature review. Molecular Biology Reports, 2022, 49, 10469-10477.	1.0	1
19	Case report: Functional characterization of a de novo c.145G>A p.Val49Met pathogenic variant in a case of PIGA-CDG with megacolon. Frontiers in Genetics, 0, 13, .	1.1	1

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20	Biallelic variants in PIGN cause Fryns syndrome, multiple congenital anomalies-hypotonia-seizures syndrome, and neurologic phenotypes: AÂgenotypeâ€“phenotype correlation study. Genetics in Medicine, 2023, 25, 37-48.	1.1	3
21	Epidemiology of congenital disorders of glycosylation (CDG)â€”overview and perspectives. , 2022, 1, .		6
22	Cannabidiol Add-On in Glycosylphosphatidylinositol-Related Drug-Resistant Epilepsy. Cannabis and Cannabinoid Research, 0, , .	1.5	2