

Jirair K Bedoyan

List of Publications by Year in descending order

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Version: 2024-02-01

37
papers

982
citations

471509

17
h-index

434195

31
g-index

38
all docs

38
docs citations

38
times ranked

1436
citing authors

#	ARTICLE	IF	CITATIONS
1	Solvent accessibility of E1 α and E1 β residues with known missense mutations causing pyruvate dehydrogenase complex (<sc>PDC</sc>) deficiency: Impact on <sc>PDC</sc> structure and function. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 557-570.	3.6	6
2	Novel multilocus imprinting disturbances in a child with expressive language delay and intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2022, , .	1.2	2
3	Simulations of Pathogenic E1 α Variants: Allosteric and Impact on Pyruvate Dehydrogenase Complex-E1 Structure and Function. <i>Journal of Chemical Information and Modeling</i> , 2022, 62, 3463-3475.	5.4	1
4	A Novel Homozygous Missense Mutation in the <i>YARS</i> Gene: Expanding the Phenotype of <i>YARS</i> Multisystem Disease. <i>Journal of the Endocrine Society</i> , 2021, 5, bvaa196.	0.2	6
5	Enantiomer-specific pharmacokinetics of D,L- β -hydroxybutyrate: Implications for the treatment of multiple <sc>acyl</sc>CoA deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 926-938.	3.6	10
6	Novel presentations associated with a PDHA1 variant –“ Alternating hemiplegia in Hemizygote proband and Guillain Barre Syndrome in Heterozygote mother. <i>European Journal of Paediatric Neurology</i> , 2021, 31, 27-30.	1.6	2
7	Utility of specific amino acid ratios in screening for pyruvate dehydrogenase complex deficiencies and other disorders associated with lactic acidosis. <i>Molecular Genetics and Metabolism</i> , 2021, 132, S6-S7.	1.1	0
8	Utility of specific amino acid ratios in screening for pyruvate dehydrogenase complex deficiencies and other mitochondrial disorders associated with congenital lactic acidosis and newborn screening prospects. <i>JIMD Reports</i> , 2020, 56, 70-81.	1.5	8
9	The E273del variant of uncertain significance of the ornithine transcarbamylase gene - a case for reclassification. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 23, 100598.	1.1	1
10	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. <i>Human Mutation</i> , 2020, 41, 946-960.	2.5	14
11	Consensus guidelines for management of hyperammonaemia in paediatric patients receiving continuous kidney replacement therapy. <i>Nature Reviews Nephrology</i> , 2020, 16, 471-482.	9.6	52
12	Mitochondrial diseases in North America. <i>Neurology: Genetics</i> , 2020, 6, e402.	1.9	38
13	A novel null mutation in the pyruvate dehydrogenase phosphatase catalytic subunit gene (PDP1) causing pyruvate dehydrogenase complex deficiency. <i>JIMD Reports</i> , 2019, 48, 26-35.	1.5	10
14	Early prediction of phenotypic severity in Citrullinemia Type 1. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 1858-1871.	3.7	26
15	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. <i>Annals of Neurology</i> , 2019, 86, 116-128.	5.3	42
16	Pearson Syndrome: A Rare Cause of Failure to Thrive in Infants. <i>Clinical Pediatrics</i> , 2019, 58, 819-824.	0.8	7
17	Life-threatening presentations of propionic acidemia due to the Amish PCCB founder variant. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 21, 100537.	1.1	4
18	Lethal neonatal case and review of primary short-chain enoyl-CoA hydratase (SCEH) deficiency associated with secondary lymphocyte pyruvate dehydrogenase complex (PDC) deficiency. <i>Molecular Genetics and Metabolism</i> , 2017, 120, 342-349.	1.1	31

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19	Succinyl-CoA synthetase (SUCLA2) deficiency in two siblings with impaired activity of other mitochondrial oxidative enzymes in skeletal muscle without mitochondrial DNA depletion. <i>Molecular Genetics and Metabolism</i> , 2017, 120, 213-222.	1.1	24
20	Enzymatic testing sensitivity, variability and practical diagnostic algorithm for pyruvate dehydrogenase complex (PDC) deficiency. <i>Molecular Genetics and Metabolism</i> , 2017, 122, 61-66.	1.1	16
21	The M405V allele of the glutaryl-CoA dehydrogenase gene is an important marker for glutaric aciduria type I (GA-I) low excretors. <i>Molecular Genetics and Metabolism</i> , 2016, 119, 50-56.	1.1	12
22	The Value of Comprehensive Thyroid Function Testing and Family History for Early Diagnosis of MCT8 Deficiency. <i>Clinical Pediatrics</i> , 2016, 55, 286-289.	0.8	4
23	Clinical and biochemical characterization of four patients with mutations in ECHS1. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 79.	2.7	68
24	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 388-396.	1.1	76
25	Novel SMC1A frameshift mutations in children with developmental delay and epilepsy. <i>European Journal of Medical Genetics</i> , 2015, 58, 562-568.	1.3	26
26	Age-related effect of serotonin transporter genotype on amygdala and prefrontal cortex function in adolescence. <i>Human Brain Mapping</i> , 2014, 35, 646-658.	3.6	18
27	Somatic mosaicism for a novel PDHA1 mutation in a male with severe pyruvate dehydrogenase complex deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 362-367.	1.1	6
28	Leigh Syndrome in a Girl With a Novel DLD Mutation Causing E3 Deficiency. <i>Pediatric Neurology</i> , 2013, 48, 67-72.	2.1	38
29	The impact of serotonin transporter (5-HTTLPR) genotype on the development of resting-state functional connectivity in children and adolescents: A preliminary report. <i>NeuroImage</i> , 2012, 59, 2760-2770.	4.2	55
30	Microarray oligonucleotide probe designer: a Web service. <i>Open Access Bioinformatics</i> , 2010, 2, 145.	0.9	5
31	First case of deletion of the faciogenital dysplasia 1 (FGD1) gene in a patient with Aarskog-Scott syndrome. <i>European Journal of Medical Genetics</i> , 2009, 52, 262-264.	1.3	20
32	Congenital diaphragmatic hernia: associated anomalies and antenatal diagnosis. <i>Pediatric Surgery International</i> , 2004, 20, 170-176.	1.4	55
33	Surgical Outcomes after Breast Cancer Surgery: Measuring Acute Lymphedema. <i>Journal of Surgical Research</i> , 2001, 95, 147-151.	1.6	52
34	Condensation of Rat Telomere-specific Nucleosomal Arrays Containing Unusually Short DNA Repeats and Histone H1. <i>Journal of Biological Chemistry</i> , 1996, 271, 18485-18493.	3.4	30
35	Nucleosomal organization of telomere-specific chromatin in rat. <i>Cell</i> , 1993, 73, 775-787.	28.9	168
36	Effect of excess dietary glucose on growth and immune response of <i>Manduca sexta</i> . <i>Journal of Insect Physiology</i> , 1992, 38, 525-532.	2.0	14

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37	DNA repair in a small yeast plasmid folded into chromatin. <i>Nucleic Acids Research</i> , 1990, 18, 2045-2051.	14.5	35