Jirair K Bedoyan

List of Publications by Year
in descending order

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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	Nucleosomal organization of telomere-specific chromatin in rat. Cell, 1993, 73, 775-787.	28.9	168
2	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. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
3	Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79.	2.7	68
4	Congenital diaphragmatic hernia: associated anomalies and antenatal diagnosis. Pediatric Surgery International, 2004, 20, 170-176.	1.4	55
5	The impact of serotonin transporter (5-HTTLPR) genotype on the development of resting-state functional connectivity in children and adolescents: A preliminary report. NeuroImage, 2012, 59, 2760-2770.	4.2	55
6	Surgical Outcomes after Breast Cancer Surgery: Measuring Acute Lymphedema. Journal of Surgical Research, 2001, 95, 147-151.	1.6	52
7	Consensus guidelines for management of hyperammonaemia in paediatric patients receiving continuous kidney replacement therapy. Nature Reviews Nephrology, 2020, 16, 471-482.	9.6	52
8	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
9	Leigh Syndrome in a Girl With a Novel DLD Mutation Causing E3 Deficiency. Pediatric Neurology, 2013, 48, 67-72.	2.1	38
10	Mitochondrial diseases in North America. Neurology: Genetics, 2020, 6, e402.	1.9	38
11	DNA repair in a small yeast plasmid folded into chromatin. Nucleic Acids Research, 1990, 18, 2045-2051.	14.5	35
12	Lethal neonatal case and review of primary short-chain enoyl-CoA hydratase (SCEH) deficiency associated with secondary lymphocyte pyruvate dehydrogenase complex (PDC) deficiency. Molecular Genetics and Metabolism, 2017, 120, 342-349.	1.1	31
13	Condensation of Rat Telomere-specific Nucleosomal Arrays Containing Unusually Short DNA Repeats and Histone H1. Journal of Biological Chemistry, 1996, 271, 18485-18493.	3.4	30
14	Novel SMC1A frameshift mutations in children with developmental delay and epilepsy. European Journal of Medical Genetics, 2015, 58, 562-568.	1.3	26
15	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26
16	Succinyl-CoA synthetase (SUCLA2) deficiency in two siblings with impaired activity of other mitochondrial oxidative enzymes in skeletal muscle without mitochondrial DNA depletion. Molecular Genetics and Metabolism, 2017, 120, 213-222.	1.1	24
17	First case of deletion of the faciogenital dysplasia 1 (FGD1) gene in a patient with Aarskogâ€“Scott syndrome. European Journal of Medical Genetics, 2009, 52, 262-264.	1.3	20
18	Ageâ€related effect of serotonin transporter genotype on amygdala and prefrontal cortex function in adolescence. Human Brain Mapping, 2014, 35, 646-658.	3.6	18

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19	Enzymatic testing sensitivity, variability and practical diagnostic algorithm for pyruvate dehydrogenase complex (PDC) deficiency. Molecular Genetics and Metabolism, 2017, 122, 61-66.	1.1	16
20	Effect of excess dietary glucose on growth and immune response of Manduca sexta. Journal of Insect Physiology, 1992, 38, 525-532.	2.0	14
21	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	2.5	14
22	The M405V allele of the glutaryl-CoA dehydrogenase gene is an important marker for glutaric aciduria type I (GA-I) low excretors. Molecular Genetics and Metabolism, 2016, 119, 50-56.	1.1	12
23	A novel null mutation in the pyruvate dehydrogenase phosphatase catalytic subunit gene (PDP1) causing pyruvate dehydrogenase complex deficiency. JIMD Reports, 2019, 48, 26-35.	1.5	10
24	Enantiomerâ€specific pharmacokinetics of D,Lâ€3â€hydroxybutyrate: Implications for the treatment of multiple <scp>acylâ€CoA</scp> dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 926-938.	3.6	10
25	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. JIMD Reports, 2020, 56, 70-81.	1.5	8
26	Pearson Syndrome: A Rare Cause of Failure to Thrive in Infants. Clinical Pediatrics, 2019, 58, 819-824.	0.8	7
27	Somatic mosaicism for a novel PDHA1 mutation in a male with severe pyruvate dehydrogenase complex deficiency. Molecular Genetics and Metabolism Reports, 2014, 1, 362-367.	1.1	6
28	A Novel Homozygous Missense Mutation in the YARS Gene: Expanding the Phenotype of YARS Multisystem Disease. Journal of the Endocrine Society, 2021, 5, bvaa196.	0.2	6
29	Solvent accessibility of E1Î± and E1Î² residues with known missense mutations causing pyruvate dehydrogenase complex (<scp>PDC</scp>) deficiency: Impact on <scp>PDCâ€E1</scp> structure and function. Journal of Inherited Metabolic Disease, 2022, 45, 557-570.	3.6	6
30	Microarray oligonucleotide probe designer: a Web service. Open Access Bioinformatics, 2010, 2, 145.	0.9	5
31	The Value of Comprehensive Thyroid Function Testing and Family History for Early Diagnosis of MCT8 Deficiency. Clinical Pediatrics, 2016, 55, 286-289.	0.8	4
32	Life-threatening presentations of propionic acidemia due to the Amish PCCB founder variant. Molecular Genetics and Metabolism Reports, 2019, 21, 100537.	1.1	4
33	Novel presentations associated with a PDHA1 variant â€“ Alternating hemiplegia in Hemizygote proband and Guillain Barre Syndrome in Heterozygote mother. European Journal of Paediatric Neurology, 2021, 31, 27-30.	1.6	2
34	Novel multilocus imprinting disturbances in a child with expressive language delay and intellectual disability. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
35	The E273del variant of uncertain significance of the ornithine transcarbamylase gene - a case for reclassification. Molecular Genetics and Metabolism Reports, 2020, 23, 100598.	1.1	1
36	Simulations of Pathogenic E1Î± Variants: Allostery and Impact on Pyruvate Dehydrogenase Complex-E1 Structure and Function. Journal of Chemical Information and Modeling, 2022, 62, 3463-3475.	5.4	1

#	Article	IF	Citations
37	Utility of specific amino acid ratios in screening for pyruvate dehydrogenase complex deficiencies and other disorders associated with lactic acidosis. Molecular Genetics and Metabolism, 2021, 132, S6-S7.	1.1	0