

Steven F Dobrowolski

List of Publications by Year in descending order

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Version: 2024-02-01

33
papers

1,122
citations

471509

17
h-index

395702

33
g-index

37
all docs

37
docs citations

37
times ranked

1689
citing authors

#	ARTICLE	IF	CITATIONS
1	Base-pair neutral homozygotes can be discriminated by calibrated high-resolution melting of small amplicons. <i>Nucleic Acids Research</i> , 2008, 36, 3401-3408.	14.5	99
2	Host conditioning and rejection monitoring in hepatocyte transplantation in humans. <i>Journal of Hepatology</i> , 2017, 66, 987-1000.	3.7	99
3	Identifying sequence variants in the human mitochondrial genome using high-resolution melt (HRM) profiling. <i>Human Mutation</i> , 2009, 30, 891-898.	2.5	81
4	Molecular genetics and impact of residual in vitro phenylalanine hydroxylase activity on tetrahydrobiopterin responsiveness in Turkish PKU population. <i>Molecular Genetics and Metabolism</i> , 2011, 102, 116-121.	1.1	71
5	Newborn Blood Spot Screening Test Using Multiplexed Real-Time PCR to Simultaneously Screen for Spinal Muscular Atrophy and Severe Combined Immunodeficiency. <i>Clinical Chemistry</i> , 2015, 61, 412-419.	3.2	68
6	Unique aspects of sequence variant interpretation for inborn errors of metabolism (IEM): The ClinGen IEM Working Group and the Phenylalanine Hydroxylase Gene. <i>Human Mutation</i> , 2018, 39, 1569-1580.	2.5	50
7	Mutations in the phenylalanine hydroxylase gene identified in 95 patients with phenylketonuria using novel systems of mutation scanning and specific genotyping based upon thermal melt profiles. <i>Molecular Genetics and Metabolism</i> , 2007, 91, 218-227.	1.1	44
8	Newborn Screening for Spinal Muscular Atrophy by Calibrated Short-Amplicon Melt Profiling. <i>Clinical Chemistry</i> , 2012, 58, 1033-1039.	3.2	36
9	Parental attitudes toward newborn screening for Duchenne/Becker muscular dystrophy and spinal muscular atrophy. <i>Muscle and Nerve</i> , 2014, 49, 822-828.	2.2	33
10	Optimization of an Automated DNA Purification Protocol for Neonatal Screening. <i>Archives of Pathology and Laboratory Medicine</i> , 1999, 123, 1154-1160.	2.5	33
11	A porcine model of phenylketonuria generated by CRISPR/Cas9 genome editing. <i>JCI Insight</i> , 2020, 5, .	5.0	29
12	Streamlined assessment of gene variants by high resolution melt profiling utilizing the ornithine transcarbamylase gene as a model system. <i>Human Mutation</i> , 2007, 28, 1133-1140.	2.5	26
13	The phenylalanine hydroxylase c.30C>G synonymous variation (p.G10G) creates a common exonic splicing silencer. <i>Molecular Genetics and Metabolism</i> , 2010, 100, 316-323.	1.1	23
14	Impaired mitochondrial medium-chain fatty acid oxidation drives periportal macrovesicular steatosis in sirtuin-5 knockout mice. <i>Scientific Reports</i> , 2020, 10, 18367.	3.3	21
15	Altered DNA methylation in PAH deficient phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2015, 115, 72-77.	1.1	20
16	Physiological Perspectives on the Use of Triheptanoin as Anaplerotic Therapy for Long Chain Fatty Acid Oxidation Disorders. <i>Frontiers in Genetics</i> , 2020, 11, 598760.	2.3	19
17	A limited spectrum of phenylalanine hydroxylase mutations is observed in phenylketonuria patients in western Poland and implications for treatment with 6R tetrahydrobiopterin. <i>Journal of Human Genetics</i> , 2009, 54, 335-339.	2.3	18
18	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. <i>PLoS Genetics</i> , 2016, 12, e1006039.	3.5	18

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19	A bone mineralization defect in the Pahenu2 model of classical phenylketonuria involves compromised mesenchymal stem cell differentiation. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 193-199.	1.1	18
20	Modeling correlates of low bone mineral density in patients with phenylalanine hydroxylase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 363-372.	3.6	14
21	The high-density lipoprotein receptor Scarb1 is required for normal bone differentiation in vivo and in vitro. <i>Laboratory Investigation</i> , 2019, 99, 1850-1860.	3.7	13
22	Biliary-Atresia-Associated Mannosidase-1-Alpha-2 Gene Regulates Biliary and Ciliary Morphogenesis and Laterality. <i>Frontiers in Physiology</i> , 2020, 11, 538701.	2.8	13
23	Clinical, biochemical, mitochondrial, and metabolomic aspects of methylmalonate semialdehyde dehydrogenase deficiency: Report of a fifth case. <i>Molecular Genetics and Metabolism</i> , 2020, 129, 272-277.	1.1	12
24	Phenylketonuria oxidative stress and energy dysregulation: Emerging pathophysiological elements provide interventional opportunity. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 111-117.	1.1	10
25	Complex patterns of inheritance, including synergistic heterozygosity, in inborn errors of metabolism: Implications for precision medicine driven diagnosis and treatment. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 1-9.	1.1	8
26	Mesenchymal stem cell energy deficit and oxidative stress contribute to osteopenia in the Pahenu2 classical PKU mouse. <i>Molecular Genetics and Metabolism</i> , 2021, 132, 173-179.	1.1	8
27	Phenylalanine hydroxylase genotype-phenotype associations in the United States: A single center study. <i>Molecular Genetics and Metabolism</i> , 2019, 128, 415-421.	1.1	7
28	Mitochondrial respiratory chain disorders in the Old Order Amish population. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 296-303.	1.1	6
29	Acquired deficiency of peroxisomal dicarboxylic acid catabolism is a metabolic vulnerability in hepatoblastoma. <i>Journal of Biological Chemistry</i> , 2021, 296, 100283.	3.4	6
30	Comparative metabolomics in the Pahenu2 classical PKU mouse identifies cerebral energy pathway disruption and oxidative stress. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 38-45.	1.1	5
31	Growth and mineralization of osteoblasts from mesenchymal stem cells on microporous membranes: Epithelial-like growth with transmembrane resistance and pH gradient. <i>Biochemical and Biophysical Research Communications</i> , 2021, 580, 14-19.	2.1	3
32	A New View of Bone Loss in Phenylketonuria. <i>Organogenesis</i> , 2021, , 1-6.	1.2	2
33	An Infant with a Constellation of Biochemical Abnormalities. <i>Clinical Chemistry</i> , 2021, 67, 1035-1036.	3.2	1