Steven F Dobrowolski

List of Publications by Year
in descending order

33	1,122	471509 17	395702 33
papers	citations	h-index	g-index

37	37	37	1689
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Comparative metabolomics in the Pahenu2 classical PKU mouse identifies cerebral energy pathway disruption and oxidative stress. Molecular Genetics and Metabolism, 2022, 136, 38-45.	1.1	5
2	Phenylketonuria oxidative stress and energy dysregulation: Emerging pathophysiological elements provide interventional opportunity. Molecular Genetics and Metabolism, 2022, 136, 111-117.	1.1	10
3	Acquired deficiency of peroxisomal dicarboxylic acid catabolism is a metabolic vulnerability in hepatoblastoma. Journal of Biological Chemistry, 2021, 296, 100283.	3.4	6
4	Mesenchymal stem cell energy deficit and oxidative stress contribute to osteopenia in the Pahenu2 classical PKU mouse. Molecular Genetics and Metabolism, 2021, 132, 173-179.	1.1	8
5	An Infant with a Constellation of Biochemical Abnormalities. Clinical Chemistry, 2021, 67, 1035-1036.	3.2	1
6	A New View of Bone Loss in Phenylketonuria. Organogenesis, 2021, , 1-6.	1.2	2
7	Growth and mineralization of osteoblasts from mesenchymal stem cells on microporous membranes: Epithelial-like growth with transmembrane resistance and pH gradient. Biochemical and Biophysical Research Communications, 2021, 580, 14-19.	2.1	3
8	Biliary-Atresia-Associated Mannosidase-1-Alpha-2 Gene Regulates Biliary and Ciliary Morphogenesis and Laterality. Frontiers in Physiology, 2020, 11, 538701.	2.8	13
9	Clinical, biochemical, mitochondrial, and metabolomic aspects of methylmalonate semialdehyde dehydrogenase deficiency: Report of a fifth case. Molecular Genetics and Metabolism, 2020, 129, 272-277.	1.1	12
10	Impaired mitochondrial medium-chain fatty acid oxidation drives periportal macrovesicular steatosis in sirtuin-5 knockout mice. Scientific Reports, 2020, 10, 18367.	3.3	21
11	Physiological Perspectives on the Use of Triheptanoin as Anaplerotic Therapy for Long Chain Fatty Acid Oxidation Disorders. Frontiers in Genetics, 2020, 11, 598760.	2.3	19
12	A porcine model of phenylketonuria generated by CRISPR/Cas9 genome editing. JCI Insight, 2020, 5, .	5.0	29
13	Complex patterns of inheritance, including synergistic heterozygosity, in inborn errors of metabolism: Implications for precision medicine driven diagnosis and treatment. Molecular Genetics and Metabolism, 2019, 128, 1-9.	1.1	8
14	Phenylalanine hydroxylase genotype-phenotype associations in the United States: A single center study. Molecular Genetics and Metabolism, 2019, 128, 415-421.	1.1	7
15	The high-density lipoprotein receptor Scarb1 is required for normal bone differentiation in vivo and in vitro. Laboratory Investigation, 2019, 99, 1850-1860.	3.7	13
16	Unique aspects of sequence variant interpretation for inborn errors of metabolism (IEM): The ClinGen IEM Working Group and the Phenylalanine Hydroxylase Gene. Human Mutation, 2018, 39, 1569-1580.	2.5	50
17	A bone mineralization defect in the Pahenu2 model of classical phenylketonuria involves compromised mesenchymal stem cell differentiation. Molecular Genetics and Metabolism, 2018, 125, 193-199.	1.1	18
18	Host conditioning and rejection monitoring in hepatocyte transplantation in humans. Journal of Hepatology, 2017, 66, 987-1000.	3.7	99

#	Article	IF	Citations
19	The Splicing Efficiency of Activating HRAS Mutations Can Determine Costello Syndrome Phenotype and Frequency in Cancer. PLoS Genetics, 2016, 12, e1006039.	3.5	18
20	Modeling correlates of low bone mineral density in patients with phenylalanine hydroxylase deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 363-372.	3.6	14
21	Mitochondrial respiratory chain disorders in the Old Order Amish population. Molecular Genetics and Metabolism, 2016, 118, 296-303.	1.1	6
22	Altered DNA methylation in PAH deficient phenylketonuria. Molecular Genetics and Metabolism, 2015, 115, 72-77.	1.1	20
23	Newborn Blood Spot Screening Test Using Multiplexed Real-Time PCR to Simultaneously Screen for Spinal Muscular Atrophy and Severe Combined Immunodeficiency. Clinical Chemistry, 2015, 61, 412-419.	3.2	68
24	Parental attitudes toward newborn screening for Duchenne/Becker muscular dystrophy and spinal muscular atrophy. Muscle and Nerve, 2014, 49, 822-828.	2.2	33
25	Newborn Screening for Spinal Muscular Atrophy by Calibrated Short-Amplicon Melt Profiling. Clinical Chemistry, 2012, 58, 1033-1039.	3.2	36
26	Molecular genetics and impact of residual in vitro phenylalanine hydroxylase activity on tetrahydrobiopterin responsiveness in Turkish PKU population. Molecular Genetics and Metabolism, 2011, 102, 116-121.	1.1	71
27	The phenylalanine hydroxylase c.30C>G synonymous variation (p.G10G) creates a common exonic splicing silencer. Molecular Genetics and Metabolism, 2010, 100, 316-323.	1.1	23
28	Identifying sequence variants in the human mitochondrial genome using high-resolution melt (HRM) profiling. Human Mutation, 2009, 30, 891-898.	2.5	81
29	A limited spectrum of phenylalanine hydroxylase mutations is observed in phenylketonuria patients in western Poland and implications for treatment with 6R tetrahydrobiopterin. Journal of Human Genetics, 2009, 54, 335-339.	2.3	18
30	Base-pair neutral homozygotes can be discriminated by calibrated high-resolution melting of small amplicons. Nucleic Acids Research, 2008, 36, 3401-3408.	14.5	99
31	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. Molecular Genetics and Metabolism, 2007, 91, 218-227.	1.1	44
32	Streamlined assessment of gene variants by high resolution melt profiling utilizing the ornithine transcarbamylase gene as a model system. Human Mutation, 2007, 28, 1133-1140.	2.5	26
33	Optimization of an Automated DNA Purification Protocol for Neonatal Screening. Archives of Pathology and Laboratory Medicine, 1999, 123, 1154-1160.	2.5	33