Devin Oglesbee

List of Publications by Year
in descending order

74 papers	4,086 citations	117625 34 h-index	118850 62 g-index
79 all docs	79 docs citations	79 times ranked	6145 citing authors

#	Article	IF	Citations
1	Investigating Mitochondrial Redox Potential with Redox-sensitive Green Fluorescent Protein Indicators. Journal of Biological Chemistry, 2004, 279, 13044-13053.	3.4	846
2	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. Genetics in Medicine, 2011, 13, 230-254.	2.4	308
3	Reduction of the falseâ€positive rate in newborn screening by implementation of MS/MSâ€based secondâ€tier tests: The Mayo Clinic experience (2004â€“2007). Journal of Inherited Metabolic Disease, 2007, 30, 585-592.	3.6	189
4	Combined Newborn Screening for Succinylacetone, Amino Acids, and Acylcarnitines in Dried Blood Spots. Clinical Chemistry, 2008, 54, 657-664.	3.2	138
5	Determination of Total Homocysteine, Methylmalonic Acid, and 2-Methylcitric Acid in Dried Blood Spots by Tandem Mass Spectrometry. Clinical Chemistry, 2010, 56, 1686-1695.	3.2	121
6	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117
7	Second-Tier Test for Quantification of Alloisoleucine and Branched-Chain Amino Acids in Dried Blood Spots to Improve Newborn Screening for Maple Syrup Urine Disease (MSUD). Clinical Chemistry, 2008, 54, 542-549.	3.2	114
8	Disease-Causing Mitochondrial Heteroplasmy Segregated Within Induced Pluripotent Stem Cell Clones Derived from a Patient with MELAS. Stem Cells, 2013, 31, 1298-1308.	3.2	112
9	Confirming Variants in Next-Generation Sequencing Panel Testing by Sanger Sequencing. Journal of Molecular Diagnostics, 2015, 17, 456-461.	2.8	109
10	Precision newborn screening for lysosomal disorders. Genetics in Medicine, 2018, 20, 847-854.	2.4	99
11	Circulating markers of NADH-reductive stress correlate with mitochondrial disease severity. Journal of Clinical Investigation, 2021, 131, .	8.2	95
12	The Subunit Composition of the Human NADH Dehydrogenase Obtained by Rapid One-step Immunopurification. Journal of Biological Chemistry, 2003, 278, 13619-13622.	3.4	93
13	Newborn screening for lysosomal storage disorders. Seminars in Perinatology, 2015, 39, 206-216.	2.5	91
14	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic. Mayo Clinic Proceedings, 2016, 91, 297-307.	3.0	83
15	Natural underlying mt<scp>DNA</scp> heteroplasmy as a potential source of intraâ€person hi<scp>PSC</scp> variability. EMBO Journal, 2016, 35, 1979-1990.	7.8	71
16	Composition of single-step media used for human embryo culture. Fertility and Sterility, 2017, 107, 1055-1060.e1.	1.0	61
17	Measurement of psychosine in dried blood spots â€” a possible improvement to newborn screening programs for Krabbe disease. Journal of Inherited Metabolic Disease, 2015, 38, 923-929.	3.6	59
18	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59

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19	Allelic diversity in MCAD deficiency: The biochemical classification of 54 variants identified during 5years of ACADM sequencing. Molecular Genetics and Metabolism, 2010, 100, 241-250.	1.1	58
20	m.3243A > G-Induced Mitochondrial Dysfunction Impairs Human Neuronal Development and Reduces Neuronal Network Activity and Synchronicity. Cell Reports, 2020, 31, 107538.	6.4	56
21	Streamlined determination of lysophosphatidylcholines in dried blood spots for newborn screening of X-linked adrenoleukodystrophy. Molecular Genetics and Metabolism, 2015, 114, 46-50.	1.1	54
22	Automated Spectrophotometric Analysis of Mitochondrial Respiratory Chain Complex Enzyme Activities in Cultured Skin Fibroblasts. Clinical Chemistry, 2005, 51, 2110-2116.	3.2	51
23	Development of a newborn screening follow-up algorithm for the diagnosis of isobutyryl-CoA dehydrogenase deficiency. Genetics in Medicine, 2007, 9, 108-116.	2.4	50
24	Multiplex Droplet Digital PCR Method Applicable to Newborn Screening, Carrier Status, and Assessment of Spinal Muscular Atrophy. Clinical Chemistry, 2018, 64, 1753-1761.	3.2	45
25	Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. ELife, 2020, 9, .	6.0	45
26	Moonlighting newborn screening markers: the incidental discovery of a second-tier test for Pompe disease. Genetics in Medicine, 2018, 20, 840-846.	2.4	44
27	Two-Tier Approach to the Newborn Screening of Methylenetetrahydrofolate Reductase Deficiency and Other Remethylation Disorders with Tandem Mass Spectrometry. Journal of Pediatrics, 2010, 157, 271-275.	1.8	43
28	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
29	Bile Acid Profiles in Primary Sclerosing Cholangitis and Their Ability to Predict Hepatic Decompensation. Hepatology, 2021, 74, 281-295.	7.3	40
30	The critical role of psychosine in screening, diagnosis, and monitoring of Krabbe disease. Genetics in Medicine, 2020, 22, 1108-1118.	2.4	39
31	Newborn screening for lysosomal storage disorders and other neuronopathic conditions. Developmental Disabilities Research Reviews, 2013, 17, 247-253.	2.9	38
32	Simultaneous Testing for 6 Lysosomal Storage Disorders and X-Adrenoleukodystrophy in Dried Blood Spots by Tandem Mass Spectrometry. Clinical Chemistry, 2016, 62, 1248-1254.	3.2	37
33	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
34	Design and Analytical Validation of Clinical DNA Sequencing Assays. Archives of Pathology and Laboratory Medicine, 2012, 136, 41-46.	2.5	35
35	Aripiprazole and trazodone cause elevations of 7-dehydrocholesterol in the absence of Smithâ€“Lemliâ€“Opitz Syndrome. Molecular Genetics and Metabolism, 2013, 110, 176-178.	1.1	34
36	An adult onset case of alphaâ€methylâ€acylâ€CoA racemase deficiency. Journal of Inherited Metabolic Disease, 2010, 33, 349-353.	3.6	32

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37	Incorporation of Second-Tier Biomarker Testing Improves the Specificity of Newborn Screening for Mucopolysaccharidosis Type I. International Journal of Neonatal Screening, 2020, 6, 10.	3.2	32
38	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
39	Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features. JIMD Reports, 2017, 40, 63-69.	1.5	27
40	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. PLoS ONE, 2019, 14, e0223337.	2.5	27
41	A replicating module as the unit of mitochondrial structure and functioning. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1555, 192-195.	1.0	24
42	Laboratory screening and diagnosis of open neural tube defects, 2019 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 462-474.	2.4	23
43	A Comparative Effectiveness Study of Newborn Screening Methods for Four Lysosomal Storage Disorders. International Journal of Neonatal Screening, 2020, 6, 44.	3.2	23
44	Clinical characterization of primary hyperoxaluria type 3 in comparison with types 1 and 2. Nephrology Dialysis Transplantation, 2022, 37, 869-875.	0.7	23
45	Sorbitol Is a Severity Biomarker for <scp>PMM2â€CDG</scp> with Therapeutic Implications. Annals of Neurology, 2021, 90, 887-900.	5.3	22
46	Silent Tyrosinemia Type I Without Elevated Tyrosine or Succinylacetone Associated with Liver Cirrhosis and Hepatocellular Carcinoma. Human Mutation, 2016, 37, 1097-1105.	2.5	21
47	Outcomes of four patients with homocysteine remethylation disorders detected by newborn screening. Genetics in Medicine, 2016, 18, 162-167.	2.4	21
48	A missense variant in FTCD is associated with arsenic metabolism and toxicity phenotypes in Bangladesh. PLoS Genetics, 2019, 15, e1007984.	3.5	19
49	The Combined Impact of CLIR Post-Analytical Tools and Second Tier Testing on the Performance of Newborn Screening for Disorders of Propionate, Methionine, and Cobalamin Metabolism. International Journal of Neonatal Screening, 2020, 6, 33.	3.2	19
50	Laboratory analysis of acylcarnitines, 2020 update: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 249-258.	2.4	19
51	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	2.4	18
52	Normal Muscle Respiratory Chain Enzymes Can Complicate Mitochondrial Disease Diagnosis. Pediatric Neurology, 2006, 35, 289-292.	2.1	16
53	Mitochondrial DNA sequence data reveals association of haplogroup U with psychosis in bipolar disorder. Journal of Psychiatric Research, 2017, 84, 221-226.	3.1	15
54	Usefulness of frataxin immunoassays for the diagnosis of Friedreich ataxia. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 994-1002.	1.9	14

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55	A Droplet Digital PCR Method for Severe Combined Immunodeficiency Newborn Screening. Journal of Molecular Diagnostics, 2017, 19, 755-765.	2.8	14
56	Allelic spectrum of formiminotransferaseâ€cyclodeaminase gene variants in individuals with formiminoglutamic aciduria. Molecular Genetics & Genomic Medicine, 2017, 5, 795-799.	1.2	14
57	Homogentisic acid interference in routine urine creatinine determination. Molecular Genetics and Metabolism, 2010, 100, 103-104.	1.1	13
58	An overview of peroxisomal biogenesis disorders. Molecular Genetics and Metabolism, 2005, 84, 299-301.	1.1	11
59	High-Throughput Immunoassay for the Biochemical Diagnosis of Friedreich Ataxia in Dried Blood Spots and Whole Blood. Clinical Chemistry, 2013, 59, 1461-1469.	3.2	10
60	Multiplex testing for the screening of lysosomal storage disease in urine: Sulfatides and glycosaminoglycan profiles in 40 cases of sulfatiduria. Molecular Genetics and Metabolism, 2020, 129, 106-110.	1.1	10
61	A new <scp>Dâ€galactose</scp> treatment monitoring index for <scp>PGM1â€CDG</scp>. Journal of Inherited Metabolic Disease, 2021, 44, 1263-1271.	3.6	10
62	Co-morbidity of Sanfilippo Syndrome type C and d-2-hydroxyglutaric aciduria. Journal of Neurology, 2011, 258, 1564-1565.	3.6	7
63	CAP/ACMG proficiency testing for biochemical genetics laboratories: a summary of performance. Genetics in Medicine, 2018, 20, 83-90.	2.4	7
64	Unexpected Fabry Disease in a Renal Allograft Kidney: An Underrecognized Cause of Poor Allograft Function. Ultrastructural Pathology, 2011, 35, 92-96.	0.9	6
65	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
66	Immune dysfunction in MGAT2â€CDG : A clinical report and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 213-218.	1.2	5
67	Laboratory monitoring of patients with hereditary tyrosinemia type I. Molecular Genetics and Metabolism, 2020, 130, 247-254.	1.1	4
68	The low excretor phenotype of glutaric acidemia type I is a source of false negative newborn screening results and challenging diagnoses. JIMD Reports, 2021, 60, 67-74.	1.5	4
69	Cerebellar and multi-system metabolic reprogramming associated with trauma exposure and post-traumatic stress disorder (PTSD)-like behavior in mice. Neurobiology of Stress, 2021, 14, 100300.	4.0	4
70	An activating germline IDH1 variant associated with a tumor entity characterized by unilateral and bilateral chondrosarcoma of the mastoid. Human Genetics and Genomics Advances, 2020, 1, 100006.	1.7	3
71	High-resolution mass spectrometric analysis of cardiolipin profiles in Barth syndrome. Mitochondrion, 2021, 60, 27-32.	3.4	2
72	Laboratory Diagnosis of Inborn Errors of Metabolism. , 0, , 531-544.		1

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73	Human Genetic Disorders. , 2016, , 595-663.		1
74	Risk categorization for oversight of laboratory-developed tests for inherited conditions: an updated position statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 983-985.	2.4	0