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	Clinical characterization of primary hyperoxaluria type 3 in comparison with types 1 and 2. Nephrology Dialysis Transplantation, 2022, 37, 869-875.	0.7	23
2	Bile Acid Profiles in Primary Sclerosing Cholangitis and Their Ability to Predict Hepatic Decompensation. Hepatology, 2021, 74, 281-295.	7.3	40
3	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
4	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
5	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	2.4	18
6	Circulating markers of NADH-reductive stress correlate with mitochondrial disease severity. Journal of Clinical Investigation, 2021, 131, .	8.2	95
7	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
8	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
9	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
10	High-resolution mass spectrometric analysis of cardiolipin profiles in Barth syndrome. Mitochondrion, 2021, 60, 27-32.	3.4	2
11	Sorbitol Is a Severity Biomarker for <scp>PMM2â€CDG</scp> with Therapeutic Implications. Annals of Neurology, 2021, 90, 887-900.	5.3	22
12	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
13	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
14	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
15	Laboratory monitoring of patients with hereditary tyrosinemia type I. Molecular Genetics and Metabolism, 2020, 130, 247-254.	1.1	4
16	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
17	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
18	A Comparative Effectiveness Study of Newborn Screening Methods for Four Lysosomal Storage Disorders. International Journal of Neonatal Screening, 2020, 6, 44.	3.2	23

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19	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
20	The critical role of psychosine in screening, diagnosis, and monitoring of Krabbe disease. Genetics in Medicine, 2020, 22, 1108-1118.	2.4	39
21	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
22	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
23	Characterising a healthy adult with a rare HAO1 knockout to support a therapeutic strategy for primary hyperoxaluria. ELife, 2020, 9, .	6.0	45
24	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
25	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6
26	A tailored approach to fusion transcript identification increases diagnosis of rare inherited disease. PLoS ONE, 2019, 14, e0223337.	2.5	27
27	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
28	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
29	A missense variant in FTCD is associated with arsenic metabolism and toxicity phenotypes in Bangladesh. PLoS Genetics, 2019, 15, e1007984.	3.5	19
30	CAP/ACMG proficiency testing for biochemical genetics laboratories: a summary of performance. Genetics in Medicine, 2018, 20, 83-90.	2.4	7
31	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
32	Precision newborn screening for lysosomal disorders. Genetics in Medicine, 2018, 20, 847-854.	2.4	99
33	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
34	Composition of single-step media used for human embryo culture. Fertility and Sterility, 2017, 107, 1055-1060.e1.	1.0	61
35	A Droplet Digital PCR Method for Severe Combined Immunodeficiency Newborn Screening. Journal of Molecular Diagnostics, 2017, 19, 755-765.	2.8	14
36	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

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37	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
38	Allelic spectrum of formiminotransferaseâ€cyclodeaminase gene variants in individuals with formiminoglutamic aciduria. Molecular Genetics & Genomic Medicine, 2017, 5, 795-799.	1.2	14
39	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
40	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
41	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
42	Outcome of Whole Exome Sequencing for Diagnostic Odyssey Cases of an Individualized Medicine Clinic. Mayo Clinic Proceedings, 2016, 91, 297-307.	3.0	83
43	Outcomes of four patients with homocysteine remethylation disorders detected by newborn screening. Genetics in Medicine, 2016, 18, 162-167.	2.4	21
44	Human Genetic Disorders. , 2016, , 595-663.		1
45	Newborn screening for lysosomal storage disorders. Seminars in Perinatology, 2015, 39, 206-216.	2.5	91
46	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
47	Confirming Variants in Next-Generation Sequencing Panel Testing by Sanger Sequencing. Journal of Molecular Diagnostics, 2015, 17, 456-461.	2.8	109
48	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
49	Usefulness of frataxin immunoassays for the diagnosis of Friedreich ataxia. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 994-1002.	1.9	14
50	Newborn screening for lysosomal storage disorders and other neuronopathic conditions. Developmental Disabilities Research Reviews, 2013, 17, 247-253.	2.9	38
51	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
52	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
53	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
54	Enhanced interpretation of newborn screening results without analyte cutoff values. Genetics in Medicine, 2012, 14, 648-655.	2.4	117

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55	Design and Analytical Validation of Clinical DNA Sequencing Assays. Archives of Pathology and Laboratory Medicine, 2012, 136, 41-46.	2.5	35
56	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
57	Co-morbidity of Sanfilippo Syndrome type C and d-2-hydroxyglutaric aciduria. Journal of Neurology, 2011, 258, 1564-1565.	3.6	7
58	Unexpected Fabry Disease in a Renal Allograft Kidney: An Underrecognized Cause of Poor Allograft Function. Ultrastructural Pathology, 2011, 35, 92-96.	0.9	6
59	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
60	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
61	Homogentisic acid interference in routine urine creatinine determination. Molecular Genetics and Metabolism, 2010, 100, 103-104.	1.1	13
62	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
63	An adult onset case of alphaâ€methylâ€acylâ€CoA racemase deficiency. Journal of Inherited Metabolic Disease, 2010, 33, 349-353.	3.6	32
64	Combined Newborn Screening for Succinylacetone, Amino Acids, and Acylcarnitines in Dried Blood Spots. Clinical Chemistry, 2008, 54, 657-664.	3.2	138
65	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
66	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
67	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
68	Normal Muscle Respiratory Chain Enzymes Can Complicate Mitochondrial Disease Diagnosis. Pediatric Neurology, 2006, 35, 289-292.	2.1	16
69	Automated Spectrophotometric Analysis of Mitochondrial Respiratory Chain Complex Enzyme Activities in Cultured Skin Fibroblasts. Clinical Chemistry, 2005, 51, 2110-2116.	3.2	51
70	An overview of peroxisomal biogenesis disorders. Molecular Genetics and Metabolism, 2005, 84, 299-301.	1.1	11
71	Investigating Mitochondrial Redox Potential with Redox-sensitive Green Fluorescent Protein Indicators. Journal of Biological Chemistry, 2004, 279, 13044-13053.	3.4	846
72	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

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73	A replicating module as the unit of mitochondrial structure and functioning. Biochimica Et Biophysica Acta - Bioenergetics, 2002, 1555, 192-195.	1.0	24
74	Laboratory Diagnosis of Inborn Errors of Metabolism. , 0, , 531-544.		1