Joshua L Deignan

List of Publications by Year
in descending order

36	3,037	394421 19	377865 34
papers	citations	h-index	g-index

37	37	37	6958
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Addendum: Technical standards and guidelines for spinal muscular atrophy testing. Genetics in Medicine, 2021, 23, 2462.	2.4	2
2	Incidental detection of acquired variants in germline genetic and genomic testing: a points to consider statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1179-1184.	2.4	13
3	Clinical Exome Reanalysis: Current Practice and Beyond. Molecular Diagnosis and Therapy, 2021, 25, 529-536.	3.8	27
4	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	2.5	58
5	CFTR variant testing: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1288-1295.	2.4	39
6	Points to consider when assessing relationships (or suspecting misattributed relationships) during family-based clinical genomic testing: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1285-1287.	2.4	9
7	Estimating the relative frequency of leukodystrophies and recommendations for carrier screening in the era of nextâ€generation sequencing. American Journal of Medical Genetics, Part A, 2020, 182, 1906-1912.	1.2	22
8	Points to consider in the reevaluation and reanalysis of genomic test results: a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 1267-1270.	2.4	147
9	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
10	Response to Knoppers et al.. Genetics in Medicine, 2019, 21, 2403.	2.4	0
11	Homozygosity for the A431E mutation in PSEN1 presenting with a relatively aggressive phenotype. Neuroscience Letters, 2019, 699, 195-198.	2.1	8
12	Misattributed parentage as an unanticipated finding during exome/genome sequencing: current clinical laboratory practices and an opportunity for standardization. Genetics in Medicine, 2019, 21, 861-866.	2.4	14
13	Patient re-contact after revision of genomic test results: points to considerâ€”a statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 769-771.	2.4	91
14	Identification of novel PIEZO1 variants using prenatal exome sequencing and correlation to ultrasound and autopsy findings of recurrent hydrops fetalis. American Journal of Medical Genetics, Part A, 2018, 176, 2829-2834.	1.2	29
15	Scaling resolution of variant classification differences in ClinVar between 41 clinical laboratories through an outlier approach. Human Mutation, 2018, 39, 1641-1649.	2.5	50
16	OncoKids. Journal of Molecular Diagnostics, 2018, 20, 765-776.	2.8	58
17	Is it time to retire fragile X testing as a first-tier test for developmental delay, intellectual disability, and autism spectrum disorder?. Genetics in Medicine, 2017, 19, 1380-1381.	2.4	15
18	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	2.4	68

#	Article	IF	Citations
19	Lipoyltransferase 1 Gene Defect Resulting in Fatal Lactic Acidosis in Two Siblings. Case Reports in Obstetrics and Gynecology, 2016, 2016, 1-4.	0.3	8
20	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. Annals of the New York Academy of Sciences, 2016, 1366, 49-60.	3.8	23
21	An infant with <scp>MLH</scp>3 variants, <scp>FOXG</scp>1â€duplication and multiple, benign cranial and spinal tumors: A clinical exome sequencing study. Genes Chromosomes and Cancer, 2016, 55, 131-142.	2.8	3
22	Molecular Diagnosis of Cystic Fibrosis. Current Protocols in Human Genetics, 2016, 88, 9.28.1-9.28.6.	3.5	3
23	Classifying Germline Sequence Variants in the Era of Next-Generation Sequencing. Clinical Chemistry, 2016, 62, 799-806.	3.2	0
24	Early Infantile Epileptic Encephalopathy with a de novo variant in ZEB2 identified by exome sequencing. European Journal of Medical Genetics, 2016, 59, 70-74.	1.3	8
25	De Novo Nonsense Mutations in KAT6A, a Lysine Acetyl-Transferase Gene, Cause a Syndrome Including Microcephaly and Global Developmental Delay. American Journal of Human Genetics, 2015, 96, 498-506.	6.2	115
26	Variation in pre-PCR processing of FFPE samples leads to discrepancies inBRAFandEGFRmutation detection: a diagnostic RING trial. Journal of Clinical Pathology, 2015, 68, 111-118.	2.0	34
27	Instability of a dinucleotide repeat in the 3â€²â€untranslated region (UTR) of the microsomal prostaglandin E synthaseâ€1 (mPGESâ€1) gene in microsatellite instabilityâ€high (MSIâ€H) colorectal carcinoma. Molecular Oncology, 2015, 9, 1252-1258.	4.6	2
28	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	9.0	211
29	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
30	Assessing the necessity of confirmatory testing for exome-sequencing results in a clinical molecular diagnostic laboratory. Genetics in Medicine, 2014, 16, 510-515.	2.4	121
31	Molecular Pathology Curriculum for Medical Laboratory Scientists. Journal of Molecular Diagnostics, 2014, 16, 288-296.	2.8	13
32	ACMG clinical laboratory standards for next-generation sequencing. Genetics in Medicine, 2013, 15, 733-747.	2.4	794
33	Privacy and data management in the era of massively parallel next-generation sequencing. Expert Review of Molecular Diagnostics, 2011, 11, 457-459.	3.1	12
34	Contrasting features of urea cycle disorders in human patients and knockout mouse models. Molecular Genetics and Metabolism, 2008, 93, 7-14.	1.1	55
35	Polyamine homeostasis in arginase knockout mice. American Journal of Physiology - Cell Physiology, 2007, 293, C1296-C1301.	4.6	18
36	Ornithine deficiency in the arginase double knockout mouse. Molecular Genetics and Metabolism, 2006, 89, 87-96.	1.1	64