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	Clinical Exome Sequencing for Genetic Identification of Rare Mendelian Disorders. JAMA - Journal of the American Medical Association, 2014, 312, 1880.	7.4	842
2	ACMG clinical laboratory standards for next-generation sequencing. Genetics in Medicine, 2013, 15, 733-747.	2.4	794
3	Exome Sequencing in the Clinical Diagnosis of Sporadic or Familial Cerebellar Ataxia. JAMA Neurology, 2014, 71, 1237.	9.0	211
4	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
5	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
6	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
7	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
8	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
9	Ornithine deficiency in the arginase double knockout mouse. Molecular Genetics and Metabolism, 2006, 89, 87-96.	1.1	64
10	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
11	OncoKids. Journal of Molecular Diagnostics, 2018, 20, 765-776.	2.8	58
12	A diagnostic ceiling for exome sequencing in cerebellar ataxia and related neurological disorders. Human Mutation, 2020, 41, 487-501.	2.5	58
13	Contrasting features of urea cycle disorders in human patients and knockout mouse models. Molecular Genetics and Metabolism, 2008, 93, 7-14.	1.1	55
14	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
15	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
16	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
17	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
18	Clinical Exome Reanalysis: Current Practice and Beyond. Molecular Diagnosis and Therapy, 2021, 25, 529-536.	3.8	27

#	Article	IF	Citations
19	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. Annals of the New York Academy of Sciences, 2016, 1366, 49-60.	3.8	23
20	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
21	Polyamine homeostasis in arginase knockout mice. American Journal of Physiology - Cell Physiology, 2007, 293, C1296-C1301.	4.6	18
22	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
23	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
24	Molecular Pathology Curriculum for Medical Laboratory Scientists. Journal of Molecular Diagnostics, 2014, 16, 288-296.	2.8	13
25	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
26	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
27	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
28	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
29	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
30	Homozygosity for the A431E mutation in PSEN1 presenting with a relatively aggressive phenotype. Neuroscience Letters, 2019, 699, 195-198.	2.1	8
31	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
32	Molecular Diagnosis of Cystic Fibrosis. Current Protocols in Human Genetics, 2016, 88, 9.28.1-9.28.6.	3.5	3
33	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
34	Addendum: Technical standards and guidelines for spinal muscular atrophy testing. Genetics in Medicine, 2021, 23, 2462.	2.4	2
35	Classifying Germline Sequence Variants in the Era of Next-Generation Sequencing. Clinical Chemistry, 2016, 62, 799-806.	3.2	0
36	Response to Knoppers et al.. Genetics in Medicine, 2019, 21, 2403.	2.4	0