Joseph J Shen

List of Publications by Year
in descending order

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Version: 2024-02-01

32 papers	3,206 citations	331670 21 h-index	414414 32 g-index
35 all docs	35 docs citations	35 times ranked	5573 citing authors

#	Article	IF	Citations
1	The role of clinical response to treatment in determining pathogenicity of genomic variants. Genetics in Medicine, 2021, 23, 581-585.	2.4	18
2	Response to Biesecker et al.. Genetics in Medicine, 2021, 23, 793-794.	2.4	0
3	CNKSR2-related neurodevelopmental and epilepsy disorder: a cohort of 13 new families and literature review indicating a predominance of loss of function pathogenic variants. BMC Medical Genomics, 2021, 14, 186.	1.5	10
4	A Case of Lysosomal Acid Lipase Deficiency Confirmed by Response to Sebelipase Alfa Therapy. Journal of Pediatric Gastroenterology and Nutrition, 2020, 71, 726-730.	1.8	3
5	A Recurrent Missense Variant in AP2M1 Impairs Clathrin-Mediated Endocytosis and Causes Developmental and Epileptic Encephalopathy. American Journal of Human Genetics, 2019, 104, 1060-1072.	6.2	78
6	Identifying and Addressing Genetic Counseling Challenges among Indigenous People of Oaxacaâ€"One Center's Experience with Two Immigrant Farmworker Families in the Central Valley of California. Journal of Genetic Counseling, 2018, 27, 996-1004.	1.6	7
7	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	8.1	95
8	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
9	BRAT1â€related diseaseâ€"identification of a patient without early lethality. American Journal of Medical Genetics, Part A, 2016, 170, 699-702.	1.2	24
10	TBX6 Null Variants and a Common Hypomorphic Allele in Congenital Scoliosis. New England Journal of Medicine, 2015, 372, 341-350.	27.0	239
11	Two cases of Templeâ€"Baraitser syndrome. Clinical Dysmorphology, 2015, 24, 55-60.	0.3	6
12	Regions of homozygosity identified by oligonucleotide SNP arrays: evaluating the incidence and clinical utility. European Journal of Human Genetics, 2015, 23, 663-671.	2.8	54
13	Mutations in the voltage-gated potassium channel gene KCNH1 cause Temple-Baraitser syndrome and epilepsy. Nature Genetics, 2015, 47, 73-77.	21.4	130
14	Jaffeâ€"Campanacci syndrome, revisited: detailed clinical and molecular analyses determine whether patients have neurofibromatosis type 1, coincidental manifestations, or a distinct disorder. Genetics in Medicine, 2014, 16, 448-459.	2.4	33
15	Scoliosis and vertebral anomalies: Additional abnormal phenotypes associated with chromosome 16p11.2 rearrangement. American Journal of Medical Genetics, Part A, 2014, 164, 1118-1126.	1.2	38
16	Evidence for disease penetrance relating to CNV size: Pelizaeusâ€"Merzbacher disease and manifesting carriers with a familial 11 Mb duplication at Xq22. Clinical Genetics, 2012, 81, 532-541.	2.0	27
17	Clinical spectrum associated with recurrent genomic rearrangements in chromosome 17q12. European Journal of Human Genetics, 2010, 18, 278-284.	2.8	114
18	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	3.2	447

#	Article	IF	Citations
19	Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. Genetics in Medicine, 2010, 12, 19-24.	2.4	91
20	Activin Signaling: Effects on Body Composition and Mitochondrial Energy Metabolism. Endocrinology, 2009, 150, 3521-3529.	2.8	43
21	Deficiency of Growth Differentiation Factor 3 Protects against Diet-Induced Obesity by Selectively Acting on White Adipose. Molecular Endocrinology, 2009, 23, 113-123.	3.7	52
22	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	21.4	535
23	Phenotypic consequences of genetic variation at hemizygous alleles: Sotos syndrome is a contiguous gene syndrome incorporating coagulation factor twelve (FXII) deficiency. Genetics in Medicine, 2005, 7, 479-483.	2.4	44
24	Terminal Deletion of Chromosome 15q26.1: Case Report and Brief Literature Review. Journal of Perinatology, 2005, 25, 429-432.	2.0	30
25	Low factor XII level in an individual with Sotos syndrome. Pediatric Blood and Cancer, 2005, 44, 187-189.	1.5	6
26	Mandibuloacral dysplasia caused by homozygosity for the R527H mutation in lamin A/C. Journal of Medical Genetics, 2003, 40, 854-857.	3.2	57
27	Partial deletions of the long arm of chromosome 13 associated with holoprosencephaly and the Dandy-Walker malformation. American Journal of Medical Genetics Part A, 2002, 112, 384-389.	2.4	33
28	Centromeric genotyping and direct analysis of nondisjunction in humans: Down syndrome. Chromosoma, 1998, 107, 166-172.	2.2	15
29	Characterization of susceptible chiasma configurations that increase the risk for maternal nondisjunction of chromosome 21. Human Molecular Genetics, 1997, 6, 1391-1399.	2.9	185
30	Susceptible chiasmate configurations of chromosome 21 predispose to nonâ€“disjunction in both maternal meiosis I and meiosis II. Nature Genetics, 1996, 14, 400-405.	21.4	362
31	Human aneuploidy: Incidence, origin, and etiology. , 1996, 28, 167.		1
32	Human aneuploidy: Incidence, origin, and etiology. Environmental and Molecular Mutagenesis, 1996, 28, 167-175.	2.2	5