Rui Xiao

List of Publications by Year
in descending order

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30 papers	2,452 citations	430874 18 h-index	477307 29 g-index
31 all docs	31 docs citations	31 times ranked	7936 citing authors

#	Article	IF	Citations
1	Retrospective analysis of a clinical exome sequencing cohort reveals the mutational spectrum and identifies candidate diseaseâ€“associated loci for BAFopathies. Genetics in Medicine, 2022, 24, 364-373.	2.4	12
2	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
3	Deletion of Nuclear Receptor Constitutive Androstane Receptor CAR Increases Anxiety and Lowers Androgen Levels. Journal of the Endocrine Society, 2021, 5, A807-A807.	0.2	0
4	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7
5	<scp>Genotypeâ€phenotype</scp> study and expansion of <scp>ARL6IP1â€related</scp> complicated hereditary spastic paraplegia. Clinical Genetics, 2021, 99, 477-480.	2.0	3
6	Bi-allelic Mutations in NADSYN1 Cause Multiple Organ Defects and Expand the Genotypic Spectrum of Congenital NAD Deficiency Disorders. American Journal of Human Genetics, 2020, 106, 129-136.	6.2	27
7	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	2.4	36
8	GARSâ€related disease in infantile spinal muscular atrophy: Implications for diagnosis and treatment. American Journal of Medical Genetics, Part A, 2020, 182, 1167-1176.	1.2	15
9	FXR-dependent Rubicon induction impairs autophagy in models of human cholestasis. Journal of Hepatology, 2020, 72, 1122-1131.	3.7	47
10	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	8.2	55
11	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
12	Variants in MED12L, encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. Genetics in Medicine, 2019, 21, 2713-2722.	2.4	28
13	Case report and novel treatment of an autosomal recessive Leigh syndrome caused by shortâ€chain enoylâ€CoA hydratase deficiency. American Journal of Medical Genetics, Part A, 2019, 179, 803-807.	1.2	18
14	First case of neutropenia and thrombocytopenia in the setting of cerebral cavernous malformation 3. International Journal of Hematology, 2019, 110, 95-101.	1.6	3
15	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
16	De Novo Variants Disrupting the HX Repeat Motif of ATN1 Cause a Recognizable Non-Progressive Neurocognitive Syndrome. American Journal of Human Genetics, 2019, 104, 542-552.	6.2	19
17	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
18	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	8.2	105

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19	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
20	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	27.0	565
21	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
22	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
23	Clinical and molecular characterization of de novo loss of function variants in HNRNPU. American Journal of Medical Genetics, Part A, 2017, 173, 2680-2689.	1.2	34
24	Liver receptor homologâ€1 is a critical determinant of methylâ€pool metabolism. Hepatology, 2016, 63, 95-106.	7.3	24
25	KIF5A mutations cause an infantile onset phenotype including severe myoclonus with evidence of mitochondrial dysfunction. Annals of Neurology, 2016, 80, 633-637.	5.3	47
26	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	12.8	227
27	Nutrient-sensing nuclear receptors coordinate autophagy. Nature, 2014, 516, 112-115.	27.8	412
28	Research Resource: The Estrogen Receptor Î± Cistrome Defined by DamIP. Molecular Endocrinology, 2012, 26, 349-357.	3.7	5
29	DamIP: Using Mutant DNA Adenine Methyltransferase to Study DNAâ€Protein Interactions In Vivo. Current Protocols in Molecular Biology, 2011, 94, Unit21.21.	2.9	9
30	DamIP: A novel method to identify DNA binding sites in vivo. Nuclear Receptor Signaling, 2010, 8, nrs.08003.	1.0	14