Rui Xiao

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9125955/publications.pdf

Version: 2024-02-01

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	Resolution of Disease Phenotypes Resulting from Multilocus Genomic Variation. New England Journal of Medicine, 2017, 376, 21-31.	27.0	565
2	Nutrient-sensing nuclear receptors coordinate autophagy. Nature, 2014, 516, 112-115.	27.8	412
3	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
4	Mutations in the nuclear bile acid receptor FXR cause progressive familial intrahepatic cholestasis. Nature Communications, 2016, 7, 10713.	12.8	227
5	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
6	Clinical exome sequencing for fetuses with ultrasound abnormalities and a suspected Mendelian disorder. Genome Medicine, 2018, 10, 74.	8.2	105
7	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
8	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	8.2	55
9	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
10	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
11	FXR-dependent Rubicon induction impairs autophagy in models of human cholestasis. Journal of Hepatology, 2020, 72, 1122-1131.	3.7	47
12	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	2.4	36
13	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
14	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
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	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
17	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
18	Liver receptor homologâ€1 is a critical determinant of methylâ€pool metabolism. Hepatology, 2016, 63, 95-106.	7.3	24

#	Article	IF	Citations
19	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
20	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
21	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
22	DamIP: A novel method to identify DNA binding sites in vivo. Nuclear Receptor Signaling, 2010, 8, nrs.08003.	1.0	14
23	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
24	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
25	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
26	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7
27	Research Resource: The Estrogen Receptor Î± Cistrome Defined by DamIP. Molecular Endocrinology, 2012, 26, 349-357.	3.7	5
28	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
29	<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
30	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