Mingchu Xu

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/10641198/publications.pdf

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28	968	516710 16	610901 24
papers	citations	h-index	g-index

29	29	29	1716
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Identification of autosomal recessive novel genes and retinal phenotypes in members of the solute carrier (SLC) superfamily. Genetics in Medicine, 2022, 24, 1523-1535.	2.4	5
2	PHENOTYPIC VARIABILITY OF RECESSIVE RDH12-ASSOCIATED RETINAL DYSTROPHY. Retina, 2019, 39, 2040-2052.	1.7	18
3	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	1.2	10
4	Investigating the disease association of USH2A p.C759F variant by leveraging large retinitis pigmentosa cohort data. Ophthalmic Genetics, 2018, 39, 291-292.	1.2	6
5	GRIPT: a novel case-control analysis method for Mendelian disease gene discovery. Genome Biology, 2018, 19, 203.	8.8	3
6	Whole-exome sequencing revealed HKDC1 as a candidate gene associated with autosomal-recessive retinitis pigmentosa. Human Molecular Genetics, 2018, 27, 4157-4168.	2.9	14
7	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
8	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
9	CEP78is mutated in a distinct type of Usher syndrome. Journal of Medical Genetics, 2017, 54, 190-195.	3.2	42
10	SeqCNV: a novel method for identification of copy number variations in targeted next-generation sequencing data. BMC Bioinformatics, 2017, 18, 147.	2.6	54
11	REEP6 deficiency leads to retinal degeneration through disruption of ER homeostasis and protein trafficking. Human Molecular Genetics, 2017, 26, 2667-2677.	2.9	39
12	Leveraging spliceâ€affecting variant predictors and a minigene validation system to identify Mendelian diseaseâ€causing variants among exonâ€captured variants of uncertain significance. Human Mutation, 2017, 38, 1521-1533.	2.5	27
13	The phenotypic variability of HK1-associated retinal dystrophy. Scientific Reports, 2017, 7, 7051.	3.3	21
14	A Novel Dominant Mutation in SAG, the Arrestin-1 Gene, Is a Common Cause of Retinitis Pigmentosa in Hispanic Families in the Southwestern United States. , 2017, 58, 2774.		31
15	Next-generation sequencing-based molecular diagnosis of 35 Hispanic retinitis pigmentosa probands. Scientific Reports, 2016, 6, 32792.	3.3	45
16	Next-generation sequencing-based molecular diagnosis of 12 inherited retinal disease probands of Uyghur ethnicity. Scientific Reports, 2016, 6, 21384.	3.3	17
17	ADIPOR1Is Mutated in Syndromic Retinitis Pigmentosa. Human Mutation, 2016, 37, 246-249.	2.5	41
18	Mutations inPOMGNT1cause non-syndromic retinitis pigmentosa. Human Molecular Genetics, 2016, 25, 1479-1488.	2.9	42

#	Article	IF	Citations
19	Isolated and Syndromic Retinal Dystrophy Caused by Biallelic Mutations in RCBTB1 , a Gene Implicated in Ubiquitination. American Journal of Human Genetics, 2016, 99, 470-480.	6.2	39
20	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
21	Diagnosis of a mild peroxisomal phenotype with next-generation sequencing. Molecular Genetics and Metabolism Reports, 2016, 9, 75-78.	1.1	29
22	Improved Diagnosis of Inherited Retinal Dystrophies by High-Fidelity PCR of ORF15 followed by Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 817-824.	2.8	21
23	Integrative subcellular proteomic analysis allows accurate prediction of human disease-causing genes. Genome Research, 2016, 26, 660-669.	5.5	22
24	ATF6Is Mutated in Early Onset Photoreceptor Degeneration With Macular Involvement. , 2015, 56, 3889.		53
25	Next-Generation Sequencing and Novel Variant Determination in a Cohort of 92 Familial Exudative Vitreoretinopathy Patients. , 2015, 56, 1937.		84
26	A Homozygous Missense Mutation in NEUROD1 Is Associated With Nonsyndromic Autosomal Recessive Retinitis Pigmentosa. Investigative Ophthalmology and Visual Science, 2015, 56, 150-155.	3.3	25
27	Mutations in human IFT140 cause non-syndromic retinal degeneration. Human Genetics, 2015, 134, 1069-1078.	3.8	62
28	A Missense Mutation inHK1Leads to Autosomal Dominant Retinitis Pigmentosa. , 2014, 55, 7159.		28