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	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
2	Next-Generation Sequencing and Novel Variant Determination in a Cohort of 92 Familial Exudative Vitreoretinopathy Patients. , 2015, 56, 1937.		84
3	Mutations in human IFT140 cause non-syndromic retinal degeneration. Human Genetics, 2015, 134, 1069-1078.	3.8	62
4	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
5	SeqCNV: a novel method for identification of copy number variations in targeted next-generation sequencing data. BMC Bioinformatics, 2017, 18, 147.	2.6	54
6	ATF6Is Mutated in Early Onset Photoreceptor Degeneration With Macular Involvement. , 2015, 56, 3889.		53
7	Next-generation sequencing-based molecular diagnosis of 35 Hispanic retinitis pigmentosa probands. Scientific Reports, 2016, 6, 32792.	3.3	45
8	Mutations inPOMGNT1cause non-syndromic retinitis pigmentosa. Human Molecular Genetics, 2016, 25, 1479-1488.	2.9	42
9	CEP78is mutated in a distinct type of Usher syndrome. Journal of Medical Genetics, 2017, 54, 190-195.	3.2	42
10	ADIPOR1Is Mutated in Syndromic Retinitis Pigmentosa. Human Mutation, 2016, 37, 246-249.	2.5	41
11	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
12	REEP6 deficiency leads to retinal degeneration through disruption of ER homeostasis and protein trafficking. Human Molecular Genetics, 2017, 26, 2667-2677.	2.9	39
13	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
14	Diagnosis of a mild peroxisomal phenotype with next-generation sequencing. Molecular Genetics and Metabolism Reports, 2016, 9, 75-78.	1.1	29
15	A Missense Mutation inHK1Leads to Autosomal Dominant Retinitis Pigmentosa. , 2014, 55, 7159.		28
16	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
17	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
18	Integrative subcellular proteomic analysis allows accurate prediction of human disease-causing genes. Genome Research, 2016, 26, 660-669.	5.5	22

#	Article	IF	Citations
19	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
20	The phenotypic variability of HK1-associated retinal dystrophy. Scientific Reports, 2017, 7, 7051.	3.3	21
21	PHENOTYPIC VARIABILITY OF RECESSIVE RDH12-ASSOCIATED RETINAL DYSTROPHY. Retina, 2019, 39, 2040-2052.	1.7	18
22	Next-generation sequencing-based molecular diagnosis of 12 inherited retinal disease probands of Uyghur ethnicity. Scientific Reports, 2016, 6, 21384.	3.3	17
23	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
24	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	1.2	10
25	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
26	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
27	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
28	GRIPT: a novel case-control analysis method for Mendelian disease gene discovery. Genome Biology, 2018, 19, 203.	8.8	3