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List of Publications by Year
in descending order

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30 papers	2,380 citations	430874 18 h-index	395702 33 g-index
34 all docs	34 docs citations	34 times ranked	6964 citing authors

#	Article	IF	Citations
1	BBS1 branchpoint variant is associated with non-syndromic retinitis pigmentosa. Journal of Medical Genetics, 2022, 59, 438-444.	3.2	13
2	Diagnostic yield of next-generation sequencing in 87 families with neurodevelopmental disorders. Orphanet Journal of Rare Diseases, 2022, 17, 60.	2.7	21
3	Clinical exome sequencingâ€"Mistakes and caveats. Human Mutation, 2022, 43, 1041-1055.	2.5	20
4	Scrutinizing pathogenicity of the USH2A c.2276â€‰Gâ€‰>â€‰T; p.(Cys759Phe) variant. Npj Genomic Medicine, 2022, 7, .	3.8	5
5	Novel Compound Heterozygous Mutation in TRAPPC9 Gene: The Relevance of Whole Genome Sequencing. Genes, 2021, 12, 557.	2.4	14
6	Systematic analysis of short tandem repeats in 38,095 exomes provides an additional diagnostic yield. Genetics in Medicine, 2021, 23, 1569-1573.	2.4	21
7	Whole genome sequencing and in vitro splice assays reveal genetic causes for inherited retinal diseases. Npj Genomic Medicine, 2021, 6, 97.	3.8	27
8	Identification of ADHD risk genes in extended pedigrees by combining linkage analysis and whole-exome sequencing. Molecular Psychiatry, 2020, 25, 2047-2057.	7.9	17
9	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
10	Presence of Genetic Variants Among Young Men With Severe COVID-19. JAMA - Journal of the American Medical Association, 2020, 324, 663.	7.4	626
11	Sudden death in epilepsy and ectopic neurohypophysis in Joubert syndrome 23 diagnosed using SNVs/indels and structural variants pipelines on WGS data: a case report. BMC Medical Genetics, 2020, 21, 96.	2.1	5
12	Family-based exome sequencing identifies rare coding variants in age-related macular degeneration. Human Molecular Genetics, 2020, 29, 2022-2034.	2.9	26
13	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	2.5	4
14	TBCK Encephaloneuropathy With Abnormal Lysosomal Storage: Use of a Structural Variant Bioinformatics Pipeline on Whole-Genome Sequencing Data Unravels a 20-Year-Old Clinical Mystery. Pediatric Neurology, 2019, 96, 74-75.	2.1	11
15	Genome-Wide Association Study Reveals Variants in CFH and CFHR4 Associated with Systemic Complement Activation. Ophthalmology, 2018, 125, 1064-1074.	5.2	55
16	Whole-Exome Sequencing in Age-Related Macular Degeneration Identifies Rare Variants in COL8A1, a Component of Bruchâ€™s Membrane. Ophthalmology, 2018, 125, 1433-1443.	5.2	35
17	Genetic screening for macular dystrophies in patients clinically diagnosed with dry ageâ€related macular degeneration. Clinical Genetics, 2018, 94, 569-574.	2.0	18
18	Association of Genetic Variants With Response to Antiâ€“Vascular Endothelial Growth Factor Therapy in Age-Related Macular Degeneration. JAMA Ophthalmology, 2018, 136, 875.	2.5	30

#	Article	IF	Citations
19	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. Nature Genetics, 2018, 50, 26-41.	21.4	286
20	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	27.8	544
21	Integration of liver gene co-expression networks and eGWAs analyses highlighted candidate regulators implicated in lipid metabolism in pigs. Scientific Reports, 2017, 7, 46539.	3.3	30
22	The Functional Effect of Rare Variants in Complement Genes on C3b Degradation in Patients With Age-Related Macular Degeneration. JAMA Ophthalmology, 2017, 135, 39.	2.5	48
23	Epigenetic regulation of the ELOVL6 gene is associated with a major QTL effect on fatty acid composition in pigs. Genetics Selection Evolution, 2015, 47, 20.	3.0	35
24	New insight into the SSC8 genetic determination of fatty acid composition in pigs. Genetics Selection Evolution, 2014, 46, 28.	3.0	16
25	Differences in Muscle Transcriptome among Pigs Phenotypically Extreme for Fatty Acid Composition. PLoS ONE, 2014, 9, e99720.	2.5	66
26	A Co-Association Network Analysis of the Genetic Determination of Pig Conformation, Growth and Fatness. PLoS ONE, 2014, 9, e114862.	2.5	36
27	Analysis of porcine adipose tissue transcriptome reveals differences in de novo fatty acid synthesis in pigs with divergent muscle fatty acid composition. BMC Genomics, 2013, 14, 843.	2.8	98
28	Polymorphism in the ELOVL6 Gene Is Associated with a Major QTL Effect on Fatty Acid Composition in Pigs. PLoS ONE, 2013, 8, e53687.	2.5	52
29	Liver transcriptome profile in pigs with extreme phenotypes of intramuscular fatty acid composition. BMC Genomics, 2012, 13, 547.	2.8	118
30	Survey of SSC12 Regions Affecting Fatty Acid Composition of Intramuscular Fat Using High-Density SNP Data. Frontiers in Genetics, 2011, 2, 101.	2.3	12