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List of Publications by Year
in descending order

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45 papers	3,521 citations	279798 23 h-index	223800 46 g-index
47 all docs	47 docs citations	47 times ranked	8057 citing authors

#	Article	IF	Citations
1	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
2	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	21.4	641
3	Genetic control of the CD4/CD8 T-cell ratio in humans. Nature Medicine, 1995, 1, 1279-1283.	30.7	398
4	First-trimester exposure to metformin and risk of birth defects: a systematic review and meta-analysis. Human Reproduction Update, 2014, 20, 656-669.	10.8	114
5	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. European Journal of Human Genetics, 2015, 23, 1068-1071.	2.8	113
6	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
7	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. European Journal of Human Genetics, 2015, 23, 753-760.	2.8	73
8	Prevalence, characteristics, and survival of children with esophageal atresia: A 32â€year populationâ€based study including 1,417,724 consecutive newborns. Birth Defects Research Part A: Clinical and Molecular Teratology, 2016, 106, 542-548.	1.6	66
9	Defective intracellular transport and processing of OA1 is a major cause of ocular albinism type 1. Human Molecular Genetics, 2000, 9, 3011-3018.	2.9	63
10	Neurofibromatosis-1: a maximum likelihood estimation of mutation rate. Human Genetics, 1990, 84, 116-8.	3.8	62
11	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silverâ€“Russell and Beckwithâ€“Wiedemann syndromes. Clinical Epigenetics, 2016, 8, 23.	4.1	54
12	In Vivo Detection of Choroidal Abnormalities Related to NF1: Feasibility and Comparison With Standard NIH Diagnostic Criteria in Pediatric Patients. , 2015, 56, 6036.		46
13	Anesthesiologic problems in Williams syndrome: the CACNL2A locus is not involved. Human Genetics, 1996, 98, 317-320.	3.8	45
14	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. European Journal of Human Genetics, 2015, 23, 1254-1258.	2.8	42
15	Functional connectivity correlates of response inhibition impairment in anorexia nervosa. Psychiatry Research - Neuroimaging, 2016, 247, 9-16.	1.8	40
16	Natural history of optic pathway gliomas in a cohort of unselected patients affected by Neurofibromatosis 1. Journal of Neuro-Oncology, 2017, 134, 279-287.	2.9	39
17	Evidence of a third ADPKD locus is not supported by re-analysis of designated PKD3 families. Kidney International, 2014, 85, 383-392.	5.2	37
18	Advances in the Pathogenesis of Cardiorenal Syndrome Type 3. Oxidative Medicine and Cellular Longevity, 2015, 2015, 1-8.	4.0	32

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19	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. Human Genetics, 2001, 108, 51-54.	3.8	31
20	Association study of AMH and AMHRII polymorphisms with unexplained infertility. Fertility and Sterility, 2010, 94, 1244-1248.	1.0	31
21	RETINAL VASCULAR ABNORMALITIES IN A LARGE COHORT OF PATIENTS AFFECTED BY NEUROFIBROMATOSIS TYPE 1. Retina, 2018, 38, 585-593.	1.7	30
22	Autosomal dominant benign recurrent intrahepatic cholestasis (BRIC) unlinked to 18q21 and 2q24. American Journal of Medical Genetics Part A, 2000, 95, 450-453.	2.4	29
23	Shared genetic risk between eating disorderâ€•and substanceâ€useâ€related phenotypes: Evidence from genomeâ€wide association studies. Addiction Biology, 2021, 26, e12880.	2.6	28
24	The Arg1038Gly missense variant in the NF1 gene causes a mild phenotype without neurofibromas. Molecular Genetics & Genomic Medicine, 2019, 7, e616.	1.2	26
25	Optic Pathway Glioma in Type 1 Neurofibromatosis: Review of Its Pathogenesis, Diagnostic Assessment, and Treatment Recommendations. Cancers, 2019, 11, 1790.	3.7	26
26	Breast cancer in neurofibromatosis 1: survival and risk of contralateral breast cancer in a five country cohort study. Genetics in Medicine, 2020, 22, 398-406.	2.4	26
27	Advising Mothers on the Use of Medications during Breastfeeding. Journal of Human Lactation, 2016, 32, 15-19.	1.6	24
28	Prevalence and survival of patients with anorectal malformations: A population-based study. Journal of Pediatric Surgery, 2019, 54, 1998-2003.	1.6	23
29	Clinical and genetic correlates of decision making in anorexia nervosa. Journal of Clinical and Experimental Neuropsychology, 2016, 38, 327-337.	1.3	22
30	Correlation of peripapillary retinal nerve fibre layer thickness with visual acuity in paediatric patients affected by optic pathway glioma. Acta Ophthalmologica, 2018, 96, e1004-e1009.	1.1	22
31	Human teratogens and genetic phenocopies. Understanding pathogenesis through human genes mutation. European Journal of Medical Genetics, 2017, 60, 22-31.	1.3	21
32	A synonymous splicing mutation in the SF3B4 gene segregates in a family with highly variable Nager syndrome. European Journal of Human Genetics, 2017, 25, 371-375.	2.8	20
33	FBXO28 is a critical gene of the 1q41q42 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1418-1420.	1.2	15
34	Prenatal detection of trisomy 8 mosaicism: Pregnancy outcome and follow up of a series of 17 consecutive cases. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 221, 23-27.	1.1	15
35	Is CFTR 621+3 A>G a cystic fibrosis causing mutation?. Journal of Human Genetics, 2010, 55, 23-26.	2.3	14
36	Neural signatures of the interaction between the 5-HTTLPR genotype and stressful life events in healthy women. Psychiatry Research - Neuroimaging, 2014, 223, 157-163.	1.8	14

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37	Retinal Vascular and Neural Remodeling Secondary to Optic Nerve Axonal Degeneration. Ophthalmology Retina, 2018, 2, 827-835.	2.4	14
38	Validation of CFTR intronic variants identified during cystic fibrosis population screening by a minigene splicing assay. Clinical Chemistry and Laboratory Medicine, 2015, 53, 1719-23.	2.3	12
39	Epilepsy in NF1: Epidemiologic, Genetic, and Clinical Features. A Monocentric Retrospective Study in a Cohort of 784 Patients. Cancers, 2021, 13, 6336.	3.7	10
40	Is there a link between COQ6 and schwannomatosis?. Genetics in Medicine, 2015, 17, 312-313.	2.4	7
41	Hybrid Minigene Assay: An Efficient Tool to Characterize mRNA Splicing Profiles of NF1 Variants. Cancers, 2021, 13, 999.	3.7	7
42	Catecholâ€Oâ€Methyltransferase (COMT) Val158Met Polymorphism and Eating Disorders: Data From a New Biobank and Metaâ€Analysis of Previously Published Studies. European Eating Disorders Review, 2017, 25, 524-532.	4.1	6
43	Ichthyosis and Kallmann syndrome: Not always a contiguous gene syndrome. Journal of Dermatological Science, 2015, 78, 158-160.	1.9	4
44	Regression of gadolinium-enhanced lesions in patients affected by neurofibromatosis type 1. Radiologia Medica, 2016, 121, 214-217.	7.7	4
45	The Role of Cell-Free Plasma DNA in Peritoneal Dialysis Patients with Peritonitis. Peritoneal Dialysis International, 2015, 35, 755-758.	2.3	2