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List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3022015/publications.pdf

Version: 2024-02-01

45	3,521	279701	223716
papers	citations	23 h-index	46 g-index

47	47	47	8057
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Shared genetic risk between eating disorderâ€•and substanceâ€•useâ€•related phenotypes: Evidence from genomeâ€•wide association studies. Addiction Biology, 2021, 26, e12880.	1.4	28
2	Hybrid Minigene Assay: An Efficient Tool to Characterize mRNA Splicing Profiles of NF1 Variants. Cancers, 2021, 13, 999.	1.7	7
3	Epilepsy in NF1: Epidemiologic, Genetic, and Clinical Features. A Monocentric Retrospective Study in a Cohort of 784 Patients. Cancers, 2021, 13, 6336.	1.7	10
4	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.	1.1	26
5	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.	1.1	80
6	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. Nature Genetics, 2019, 51, 1207-1214.	9.4	641
7	The Arg1038Gly missense variant in the NF1 gene causes a mild phenotype without neurofibromas. Molecular Genetics & Genomic Medicine, 2019, 7, e616.	0.6	26
8	Prevalence and survival of patients with anorectal malformations: A population-based study. Journal of Pediatric Surgery, 2019, 54, 1998-2003.	0.8	23
9	Optic Pathway Glioma in Type 1 Neurofibromatosis: Review of Its Pathogenesis, Diagnostic Assessment, and Treatment Recommendations. Cancers, 2019, 11, 1790.	1.7	26
10	Retinal Vascular and Neural Remodeling Secondary to Optic Nerve Axonal Degeneration. Ophthalmology Retina, 2018, 2, 827-835.	1.2	14
11	RETINAL VASCULAR ABNORMALITIES IN A LARGE COHORT OF PATIENTS AFFECTED BY NEUROFIBROMATOSIS TYPE 1. Retina, 2018, 38, 585-593.	1.0	30
12	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.	0.5	15
13	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.	0.6	22
14	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
15	Natural history of optic pathway gliomas in a cohort of unselected patients affected by Neurofibromatosis 1. Journal of Neuro-Oncology, 2017, 134, 279-287.	1.4	39
16	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.	1.4	20
17	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.	2.3	6
18	Human teratogens and genetic phenocopies. Understanding pathogenesis through human genes mutation. European Journal of Medical Genetics, 2017, 60, 22-31.	0.7	21

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19	Regression of gadolinium-enhanced lesions in patients affected by neurofibromatosis type 1. Radiologia Medica, 2016, 121, 214-217.	4.7	4
20	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
21	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.	1.8	54
22	Functional connectivity correlates of response inhibition impairment in anorexia nervosa. Psychiatry Research - Neuroimaging, 2016, 247, 9-16.	0.9	40
23	Clinical and genetic correlates of decision making in anorexia nervosa. Journal of Clinical and Experimental Neuropsychology, 2016, 38, 327-337.	0.8	22
24	Advising Mothers on the Use of Medications during Breastfeeding. Journal of Human Lactation, 2016, 32, 15-19.	0.8	24
25	FBXO28 is a critical gene of the 1q41q42 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1418-1420.	0.7	15
26	In Vivo Detection of Choroidal Abnormalities Related to NF1: Feasibility and Comparison With Standard NIH Diagnostic Criteria in Pediatric Patients. , 2015, 56, 6036.		46
27	Advances in the Pathogenesis of Cardiorenal Syndrome Type 3. Oxidative Medicine and Cellular Longevity, 2015, 2015, 1-8.	1.9	32
28	Is there a link between COQ6 and schwannomatosis?. Genetics in Medicine, 2015, 17, 312-313.	1.1	7
29	The Role of Cell-Free Plasma DNA in Peritoneal Dialysis Patients with Peritonitis. Peritoneal Dialysis International, 2015, 35, 755-758.	1.1	2
30	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.	1.4	73
31	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. European Journal of Human Genetics, 2015, 23, 1254-1258.	1.4	42
32	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.	1.4	12
33	Ichthyosis and Kallmann syndrome: Not always a contiguous gene syndrome. Journal of Dermatological Science, 2015, 78, 158-160.	1.0	4
34	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. European Journal of Human Genetics, 2015, 23, 1068-1071.	1.4	113
35	Evidence of a third ADPKD locus is not supported by re-analysis of designated PKD3 families. Kidney International, 2014, 85, 383-392.	2.6	37
36	First-trimester exposure to metformin and risk of birth defects: a systematic review and meta-analysis. Human Reproduction Update, 2014, 20, 656-669.	5.2	114

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37	Neural signatures of the interaction between the 5-HTTLPR genotype and stressful life events in healthy women. Psychiatry Research - Neuroimaging, 2014, 223, 157-163.	0.9	14
38	Is CFTR 621+3 A>G a cystic fibrosis causing mutation?. Journal of Human Genetics, 2010, 55, 23-26.	1.1	14
39	Association study of AMH and AMHRII polymorphisms with unexplained infertility. Fertility and Sterility, 2010, 94, 1244-1248.	0.5	31
40	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.	1.8	31
41	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
42	Defective intracellular transport and processing of OA1 is a major cause of ocular albinism type 1. Human Molecular Genetics, 2000, 9, 3011-3018.	1.4	63
43	Anesthesiologic problems in Williams syndrome: the CACNL2A locus is not involved. Human Genetics, 1996, 98, 317-320.	1.8	45
44	Genetic control of the CD4/CD8 T-cell ratio in humans. Nature Medicine, 1995, 1, 1279-1283.	15.2	398
45	Neurofibromatosis-1: a maximum likelihood estimation of mutation rate. Human Genetics, 1990, 84, 116-8.	1.8	62