

Maurizio Clementi

List of Publications by Year in descending order

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Version: 2024-02-01

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	Shared genetic risk between eating disorder and substance use-related phenotypes: Evidence from genome-wide association studies. <i>Addiction Biology</i> , 2021, 26, e12880.	2.6	28
2	Hybrid Minigene Assay: An Efficient Tool to Characterize mRNA Splicing Profiles of NF1 Variants. <i>Cancers</i> , 2021, 13, 999.	3.7	7
3	Epilepsy in NF1: Epidemiologic, Genetic, and Clinical Features. A Monocentric Retrospective Study in a Cohort of 784 Patients. <i>Cancers</i> , 2021, 13, 6336.	3.7	10
4	Breast cancer in neurofibromatosis 1: survival and risk of contralateral breast cancer in a five country cohort study. <i>Genetics in Medicine</i> , 2020, 22, 398-406.	2.4	26
5	Clinical spectrum of individuals with pathogenic <i>NF1</i> missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. <i>Human Mutation</i> , 2020, 41, 299-315.	2.5	80
6	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. <i>Nature Genetics</i> , 2019, 51, 1207-1214.	21.4	641
7	The Arg1038Gly missense variant in the <i>NF1</i> gene causes a mild phenotype without neurofibromas. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e616.	1.2	26
8	Prevalence and survival of patients with anorectal malformations: A population-based study. <i>Journal of Pediatric Surgery</i> , 2019, 54, 1998-2003.	1.6	23
9	Optic Pathway Glioma in Type 1 Neurofibromatosis: Review of Its Pathogenesis, Diagnostic Assessment, and Treatment Recommendations. <i>Cancers</i> , 2019, 11, 1790.	3.7	26
10	Retinal Vascular and Neural Remodeling Secondary to Optic Nerve Axonal Degeneration. <i>Ophthalmology Retina</i> , 2018, 2, 827-835.	2.4	14
11	RETINAL VASCULAR ABNORMALITIES IN A LARGE COHORT OF PATIENTS AFFECTED BY NEUROFIBROMATOSIS TYPE 1. <i>Retina</i> , 2018, 38, 585-593.	1.7	30
12	Prenatal detection of trisomy 8 mosaicism: Pregnancy outcome and follow up of a series of 17 consecutive cases. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2018, 221, 23-27.	1.1	15
13	Correlation of peripapillary retinal nerve fibre layer thickness with visual acuity in paediatric patients affected by optic pathway glioma. <i>Acta Ophthalmologica</i> , 2018, 96, e1004-e1009.	1.1	22
14	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	12.6	1,085
15	Natural history of optic pathway gliomas in a cohort of unselected patients affected by Neurofibromatosis 1. <i>Journal of Neuro-Oncology</i> , 2017, 134, 279-287.	2.9	39
16	A synonymous splicing mutation in the SF3B4 gene segregates in a family with highly variable Nager syndrome. <i>European Journal of Human Genetics</i> , 2017, 25, 371-375.	2.8	20
17	Catechol-O-Methyltransferase (COMT) Val158Met Polymorphism and Eating Disorders: Data From a New Biobank and Meta-Analysis of Previously Published Studies. <i>European Eating Disorders Review</i> , 2017, 25, 524-532.	4.1	6
18	Human teratogens and genetic phenocopies. Understanding pathogenesis through human genes mutation. <i>European Journal of Medical Genetics</i> , 2017, 60, 22-31.	1.3	21

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19	Regression of gadolinium-enhanced lesions in patients affected by neurofibromatosis type 1. <i>Radiologia Medica</i> , 2016, 121, 214-217.	7.7	4
20	Prevalence, characteristics, and survival of children with esophageal atresia: A 32-year population-based study including 1,417,724 consecutive newborns. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2016, 106, 542-548.	1.6	66
21	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silver-Russell and Beckwith-Wiedemann syndromes. <i>Clinical Epigenetics</i> , 2016, 8, 23.	4.1	54
22	Functional connectivity correlates of response inhibition impairment in anorexia nervosa. <i>Psychiatry Research - Neuroimaging</i> , 2016, 247, 9-16.	1.8	40
23	Clinical and genetic correlates of decision making in anorexia nervosa. <i>Journal of Clinical and Experimental Neuropsychology</i> , 2016, 38, 327-337.	1.3	22
24	Advising Mothers on the Use of Medications during Breastfeeding. <i>Journal of Human Lactation</i> , 2016, 32, 15-19.	1.6	24
25	<i>FBXO28</i> is a critical gene of the 1q41q42 microdeletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1418-1420.	1.2	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. <i>Oxidative Medicine and Cellular Longevity</i> , 2015, 2015, 1-8.	4.0	32
28	Is there a link between COQ6 and schwannomatosis?. <i>Genetics in Medicine</i> , 2015, 17, 312-313.	2.4	7
29	The Role of Cell-Free Plasma DNA in Peritoneal Dialysis Patients with Peritonitis. <i>Peritoneal Dialysis International</i> , 2015, 35, 755-758.	2.3	2
30	Loss-of-function variants of SETD5 cause intellectual disability and the core phenotype of microdeletion 3p25.3 syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 753-760.	2.8	73
31	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. <i>European Journal of Human Genetics</i> , 2015, 23, 1254-1258.	2.8	42
32	Validation of CFTR intronic variants identified during cystic fibrosis population screening by a minigene splicing assay. <i>Clinical Chemistry and Laboratory Medicine</i> , 2015, 53, 1719-23.	2.3	12
33	Ichthyosis and Kallmann syndrome: Not always a contiguous gene syndrome. <i>Journal of Dermatological Science</i> , 2015, 78, 158-160.	1.9	4
34	p.Arg1809Cys substitution in neurofibromin is associated with a distinctive NF1 phenotype without neurofibromas. <i>European Journal of Human Genetics</i> , 2015, 23, 1068-1071.	2.8	113
35	Evidence of a third ADPKD locus is not supported by re-analysis of designated PKD3 families. <i>Kidney International</i> , 2014, 85, 383-392.	5.2	37
36	First-trimester exposure to metformin and risk of birth defects: a systematic review and meta-analysis. <i>Human Reproduction Update</i> , 2014, 20, 656-669.	10.8	114

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37	Neural signatures of the interaction between the 5-HTTLPR genotype and stressful life events in healthy women. <i>Psychiatry Research - Neuroimaging</i> , 2014, 223, 157-163.	1.8	14
38	Is CFTR 621+3 A>G a cystic fibrosis causing mutation?. <i>Journal of Human Genetics</i> , 2010, 55, 23-26.	2.3	14
39	Association study of AMH and AMHRII polymorphisms with unexplained infertility. <i>Fertility and Sterility</i> , 2010, 94, 1244-1248.	1.0	31
40	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. <i>Human Genetics</i> , 2001, 108, 51-54.	3.8	31
41	Autosomal dominant benign recurrent intrahepatic cholestasis (BRIC) unlinked to 18q21 and 2q24. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 450-453.	2.4	29
42	Defective intracellular transport and processing of OA1 is a major cause of ocular albinism type 1. <i>Human Molecular Genetics</i> , 2000, 9, 3011-3018.	2.9	63
43	Anesthesiologic problems in Williams syndrome: the CACNL2A locus is not involved. <i>Human Genetics</i> , 1996, 98, 317-320.	3.8	45
44	Genetic control of the CD4/CD8 T-cell ratio in humans. <i>Nature Medicine</i> , 1995, 1, 1279-1283.	30.7	398
45	Neurofibromatosis-1: a maximum likelihood estimation of mutation rate. <i>Human Genetics</i> , 1990, 84, 116-8.	3.8	62