

Matteo Cassina

List of Publications by Year in descending order

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Version: 2024-02-01

64
papers

2,704
citations

279487

23
h-index

205818

48
g-index

71
all docs

71
docs citations

71
times ranked

5652
citing authors

#	ARTICLE	IF	CITATIONS
1	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. <i>Nature Genetics</i> , 2019, 51, 1207-1214.	9.4	641
2	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. <i>American Journal of Psychiatry</i> , 2017, 174, 850-858.	4.0	410
3	Treatment of Hyperthyroidism in Pregnancy and Birth Defects. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2010, 95, E337-E341.	1.8	139
4	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.	5.2	114
5	Pregnancy outcome in women exposed to leflunomide before or during pregnancy. <i>Arthritis and Rheumatism</i> , 2012, 64, 2085-2094.	6.7	110
6	Genetics of Coenzyme Q10 Deficiency. <i>Molecular Syndromology</i> , 2014, 5, 156-162.	0.3	102
7	Haploinsufficiency of <i>COQ4</i> causes coenzyme Q ₁₀ deficiency. <i>Journal of Medical Genetics</i> , 2012, 49, 187-191.	1.5	95
8	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
9	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.	1.8	54
10	The <i>COQ2</i> genotype predicts the severity of coenzyme Q ₁₀ deficiency. <i>Human Molecular Genetics</i> , 2016, 25, 4256-4265.	1.4	53
11	Mutations in <i>COQ8B</i> (<i>ADCK4</i>) found in patients with steroid-resistant nephrotic syndrome alter <i>COQ8B</i> function. <i>Human Mutation</i> , 2018, 39, 406-414.	1.1	43
12	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. <i>Biological Psychiatry</i> , 2019, 86, 577-586.	0.7	43
13	Recurrent HERV-H-Mediated 3q13.2-q13.31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. <i>Human Mutation</i> , 2013, 34, 1415-1423.	1.1	40
14	Functional connectivity correlates of response inhibition impairment in anorexia nervosa. <i>Psychiatry Research - Neuroimaging</i> , 2016, 247, 9-16.	0.9	40
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.	1.4	39
16	Pregnancy outcomes in women on metformin for diabetes or other indications among those seeking teratology information services. <i>British Journal of Clinical Pharmacology</i> , 2018, 84, 568-578.	1.1	35
17	RETINAL VASCULAR ABNORMALITIES IN A LARGE COHORT OF PATIENTS AFFECTED BY NEUROFIBROMATOSIS TYPE 1. <i>Retina</i> , 2018, 38, 585-593.	1.0	30
18	Copper and bezafibrate cooperate to rescue cytochrome c oxidase deficiency in cells of patients with <i>sco2</i> mutations. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 21.	1.2	29

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19	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. <i>Neurogenetics</i> , 2016, 17, 65-70.	0.7	29
20	Shared genetic risk between eating disorder and substance use related phenotypes: Evidence from genome-wide association studies. <i>Addiction Biology</i> , 2021, 26, e12880.	1.4	28
21	First trimester diclofenac exposure and pregnancy outcome. <i>Reproductive Toxicology</i> , 2010, 30, 401-404.	1.3	27
22	Genetic susceptibility to teratogens: State of the art. <i>Reproductive Toxicology</i> , 2012, 34, 186-191.	1.3	27
23	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.	0.6	26
24	Optic Pathway Glioma in Type 1 Neurofibromatosis: Review of Its Pathogenesis, Diagnostic Assessment, and Treatment Recommendations. <i>Cancers</i> , 2019, 11, 1790.	1.7	26
25	Serotonin transporter gene polymorphism in eating disorders: Data from a new biobank and META-analysis of previous studies. <i>World Journal of Biological Psychiatry</i> , 2016, 17, 244-257.	1.3	24
26	Migraine therapy during pregnancy and lactation. <i>Expert Opinion on Drug Safety</i> , 2010, 9, 937-948.	1.0	23
27	Prevalence and survival of patients with anorectal malformations: A population-based study. <i>Journal of Pediatric Surgery</i> , 2019, 54, 1998-2003.	0.8	23
28	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 103.	1.2	23
29	Clinical and genetic correlates of decision making in anorexia nervosa. <i>Journal of Clinical and Experimental Neuropsychology</i> , 2016, 38, 327-337.	0.8	22
30	Infantile epilepsy associated with mosaic 2q24 duplication including SCN2A and SCN3A. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2011, 20, 813-816.	0.9	21
31	Eating-induced epileptic spasms in a boy with <i>MECP2</i> duplication syndrome: insights into pathogenesis of genetic epilepsies. <i>Epileptic Disorders</i> , 2012, 14, 414-417.	0.7	21
32	Pregnancy outcome in women exposed to antiepileptic drugs: Teratogenic role of maternal epilepsy and its pharmacologic treatment. <i>Reproductive Toxicology</i> , 2013, 39, 50-57.	1.3	21
33	Human teratogens and genetic phenocopies. Understanding pathogenesis through human genes mutation. <i>European Journal of Medical Genetics</i> , 2017, 60, 22-31.	0.7	21
34	A synonymous splicing mutation in the <i>SF3B4</i> gene segregates in a family with highly variable Nager syndrome. <i>European Journal of Human Genetics</i> , 2017, 25, 371-375.	1.4	20
35	Recessive Spondylocarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in <i>MYH3</i> . <i>American Journal of Human Genetics</i> , 2018, 102, 1115-1125.	2.6	18
36	Pharmacologic treatment of hyperthyroidism during pregnancy. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 612-619.	1.6	17

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37	<i>FBXO28</i> is a critical gene of the 1q41q42 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1418-1420.	0.7	15
38	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
39	14q12 duplication including FOXP1: Is there a common age-dependent epileptic phenotype?. Brain and Development, 2014, 36, 402-407.	0.6	14
40	Forkhead Box G1 Gene Haploinsufficiency: An Emerging Cause of Dyskinetic Encephalopathy of Infancy. Neuropediatrics, 2015, 46, 056-064.	0.3	14
41	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
42	Mutational spectrum and clinical signatures in 114 families with hereditary multiple osteochondromas: insights into molecular properties of selected exostosin variants. Human Molecular Genetics, 2019, 28, 2133-2142.	1.4	12
43	Biallelic mutations in the TOGARAM1 gene cause a novel primary ciliopathy. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-106833.	1.5	12
44	Therapy of inflammatory bowel diseases in pregnancy and lactation. Expert Opinion on Drug Safety, 2009, 8, 695-707.	1.0	10
45	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
46	A Hunter Patient with a Severe Phenotype Reveals Two Large Deletions and Two Duplications Extending 1.2 Mb Distally to IDS Locus. JIMD Reports, 2014, 17, 13-21.	0.7	9
47	Congenital Anomalies in Contaminated Sites: A Multisite Study in Italy. International Journal of Environmental Research and Public Health, 2017, 14, 292.	1.2	8
48	Dysfunctional coping is related to impaired skin-related quality of life and psychological distress in patients with neurofibromatosis type 1 with major skin involvement. British Journal of Dermatology, 2020, 182, 1449-1457.	1.4	8
49	Handle with care – interpretation, synthesis and dissemination of data on paracetamol in pregnancy. Nature Reviews Endocrinology, 2022, 18, 191-191.	4.3	8
50	Hybrid Minigene Assay: An Efficient Tool to Characterize mRNA Splicing Profiles of NF1 Variants. Cancers, 2021, 13, 999.	1.7	7
51	Catechol-O-methyltransferase (COMT) Val158Met Polymorphism and Eating Disorders: Data From a New Biobank and Meta-analysis of Previously Published Studies. European Eating Disorders Review, 2017, 25, 524-532.	2.3	6
52	Congenital Pulmonary Airway Malformation (CPAM) [Congenital Cystic Adenomatoid Malformation] Associated with Tracheoesophageal Fistula and Agenesis of the Corpus Callosum. Fetal and Pediatric Pathology, 2012, 31, 169-175.	0.4	5
53	Genotype-phenotype correlation in Gordon's syndrome: report of two cases carrying novel heterozygous mutations. Journal of Nephrology, 2022, 35, 859-862.	0.9	5
54	Auditory Outcome after Cochlear Implantation in Children with DFNB7/11 Caused by Pathogenic Variants in <i>TMC1</i> Gene. Audiology and Neuro-Otology, 2021, 26, 157-163.	0.6	5

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55	DFNA20/26 and Other ACTG1-Associated Phenotypes: A Case Report and Review of the Literature. <i>Audiology Research</i> , 2021, 11, 582-593.	0.8	5
56	Ondansetron in pregnancy revisited: Assessment and pregnancy labelling by the European Medicines Agency (EMA) & Pharmacovigilance Risk Assessment Committee (PRAC). <i>Basic and Clinical Pharmacology and Toxicology</i> , 2021, 128, 579-582.	1.2	4
57	Ondansetron should never be used in pregnancy. <i>BJOG: an International Journal of Obstetrics and Gynaecology</i> , 2021, 128, 111-112.	1.1	3
58	Genetics & Epigenetics of Hereditary Deafness: An Historical Overview. <i>Audiology Research</i> , 2021, 11, 629-635.	0.8	3
59	Craniosynostosis is a feature of <i><sc><i>CHD7</i></sc></i> â€related <i><sc>CHARGE</sc></i> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2160-2163.	0.7	2
60	Medications in Pregnancy and Lactation. <i>Obstetrics and Gynecology</i> , 2009, 114, 166.	1.2	1
61	Victor A. McKusick, the â€œFather of Medical Geneticsâ€; <i>Audiology Research</i> , 2021, 11, 636-638.	0.8	1
62	Treatment of Hyperthyroidism in Pregnancy and Birth Defects. <i>Obstetrical and Gynecological Survey</i> , 2011, 66, 135-137.	0.2	0
63	Response to: Papetti et al., â€œThe crucial role of FBXO28 in the pathogenesis of the 1q41q42 microdeletion syndromeâ€; <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3054-3054.	0.7	0
64	Analysis of p53 polymorphisms in individuals with multiple melanocytic nevi. <i>European Journal of Dermatology</i> , 2013, 23, 280-281.	0.3	0