Matteo Cassina

List of Publications by Year
in descending order

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	Genotypeâ€“phenotype correlation in Gordonâ€™s syndrome: report of two cases carrying novel heterozygous mutations. Journal of Nephrology, 2022, 35, 859-862.	0.9	5
2	Handle with care â€” interpretation, synthesis and dissemination of data on paracetamol in pregnancy. Nature Reviews Endocrinology, 2022, 18, 191-191.	4.3	8
3	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
4	Ondansetron should never be used in pregnancy. BJOG: an International Journal of Obstetrics and Gynaecology, 2021, 128, 111-112.	1.1	3
5	Ondansetron in pregnancy revisited: Assessment and pregnancy labelling by the European Medicines Agency (EMA) & Pharmacovigilance Risk Assessment Committee (PRAC). Basic and Clinical Pharmacology and Toxicology, 2021, 128, 579-582.	1.2	4
6	Hybrid Minigene Assay: An Efficient Tool to Characterize mRNA Splicing Profiles of NF1 Variants. Cancers, 2021, 13, 999.	1.7	7
7	Craniosynostosis is a feature of <scp><i>CHD7</i></scp>â€•related <scp>CHARGE</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2160-2163.	0.7	2
8	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
9	DFNA20/26 and Other ACTG1-Associated Phenotypes: A Case Report and Review of the Literature. Audiology Research, 2021, 11, 582-593.	0.8	5
10	Victor A. McKusick, the â€œFather of Medical Geneticsâ€• Audiology Research, 2021, 11, 636-638.	0.8	1
11	Genetics & Epigenetics of Hereditary Deafness: An Historical Overview. Audiology Research, 2021, 11, 629-635.	0.8	3
12	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
13	Dysfunctional coping is related to impaired skinâ€•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
14	Biallelic mutations in the TOGARAM1 gene cause a novel primary ciliopathy. Journal of Medical Genetics, 2020, 58, jmedgenet-2020-106833.	1.5	12
15	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. Orphanet Journal of Rare Diseases, 2020, 15, 103.	1.2	23
16	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
17	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. Biological Psychiatry, 2019, 86, 577-586.	0.7	43
18	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

#	ARTICLE	IF	CITATIONS
19	The Arg1038Gly missense variant in the <i>NF1</i> gene causes a mild phenotype without neurofibromas. Molecular Genetics & Genomic Medicine, 2019, 7, e616.	0.6	26
20	Prevalence and survival of patients with anorectal malformations: A population-based study. Journal of Pediatric Surgery, 2019, 54, 1998-2003.	0.8	23
21	Optic Pathway Glioma in Type 1 Neurofibromatosis: Review of Its Pathogenesis, Diagnostic Assessment, and Treatment Recommendations. Cancers, 2019, 11, 1790.	1.7	26
22	Pregnancy outcomes in women on metformin for diabetes or other indications among those seeking teratology information services. British Journal of Clinical Pharmacology, 2018, 84, 568-578.	1.1	35
23	RETINAL VASCULAR ABNORMALITIES IN A LARGE COHORT OF PATIENTS AFFECTED BY NEUROFIBROMATOSIS TYPE 1. Retina, 2018, 38, 585-593.	1.0	30
24	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
25	Mutations in COQ8B (ADCK4) found in patients with steroid-resistant nephrotic syndrome alter COQ8B function. Human Mutation, 2018, 39, 406-414.	1.1	43
26	Recessive Spondylocarpotarsal Synostosis Syndrome Due to Compound Heterozygosity for Variants in MYH3. American Journal of Human Genetics, 2018, 102, 1115-1125.	2.6	18
27	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. American Journal of Psychiatry, 2017, 174, 850-858.	4.0	410
28	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
29	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
30	Catecholâ€<i>O</i>â€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
31	Human teratogens and genetic phenocopies. Understanding pathogenesis through human genes mutation. European Journal of Medical Genetics, 2017, 60, 22-31.	0.7	21
32	Congenital Anomalies in Contaminated Sites: A Multisite Study in Italy. International Journal of Environmental Research and Public Health, 2017, 14, 292.	1.2	8
33	Response to: Papetti et al., â€œThe crucial role of FBXO28 in the pathogenesis of the 1q41q42 microdeletion syndromeâ€. American Journal of Medical Genetics, Part A, 2016, 170, 3054-3054.	0.7	0
34	The<i>COQ2</i>genotype predicts the severity of coenzyme Q₁₀deficiency. Human Molecular Genetics, 2016, 25, 4256-4265.	1.4	53
35	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
36	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

#	Article	IF	Citations
37	Functional connectivity correlates of response inhibition impairment in anorexia nervosa. Psychiatry Research - Neuroimaging, 2016, 247, 9-16.	0.9	40
38	Serotonin transporter gene polymorphism in eating disorders: Data from a new biobank and META-analysis of previous studies. World Journal of Biological Psychiatry, 2016, 17, 244-257.	1.3	24
39	Clinical and genetic correlates of decision making in anorexia nervosa. Journal of Clinical and Experimental Neuropsychology, 2016, 38, 327-337.	0.8	22
40	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. Neurogenetics, 2016, 17, 65-70.	0.7	29
41	<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
42	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
43	Forkhead Box G1 Gene Haploinsufficiency: An Emerging Cause of Dyskinetic Encephalopathy of Infancy. Neuropediatrics, 2015, 46, 056-064.	0.3	14
44	Genetics of Coenzyme Q10 Deficiency. Molecular Syndromology, 2014, 5, 156-162.	0.3	102
45	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
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	14q12 duplication including FOXG1: Is there a common age-dependent epileptic phenotype?. Brain and Development, 2014, 36, 402-407.	0.6	14
48	Pregnancy outcome in women exposed to antiepileptic drugs: Teratogenic role of maternal epilepsy and its pharmacologic treatment. Reproductive Toxicology, 2013, 39, 50-57.	1.3	21
49	Recurrent HERV-H-Mediated 3q13.2-q13.31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. Human Mutation, 2013, 34, 1415-1423.	1.1	40
50	Analysis of p53 polymorphisms in individuals with multiple melanocytic nevi. European Journal of Dermatology, 2013, 23, 280-281.	0.3	0
51	Haploinsufficiency of<i>COQ4</i>causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	1.5	95
52	Congenital Pulmonary Airway Malformation (CPAM) [Congenital Cystic Adenomatoid Malformation] Associated with Tracheoesophageal Fistula and Agensesis of the Corpus Callosum. Fetal and Pediatric Pathology, 2012, 31, 169-175.	0.4	5
53	Copper and bezafibrate cooperate to rescue cytochrome c oxidase deficiency in cells of patients with sco2 mutations. Orphanet Journal of Rare Diseases, 2012, 7, 21.	1.2	29
54	Eatingâ€induced epileptic spasms in a boy with <i>MECP2</i> duplication syndrome: insights into pathogenesis of genetic epilepsies. Epileptic Disorders, 2012, 14, 414-417.	0.7	21

#	Article	IF	Citations
55	Pregnancy outcome in women exposed to leflunomide before or during pregnancy. Arthritis and Rheumatism, 2012, 64, 2085-2094.	6.7	110
56	Pharmacologic treatment of hyperthyroidism during pregnancy. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 612-619.	1.6	17
57	Genetic susceptibility to teratogens: State of the art. Reproductive Toxicology, 2012, 34, 186-191.	1.3	27
58	Infantile epilepsy associated with mosaic 2q24 duplication including SCN2A and SCN3A. Seizure: the Journal of the British Epilepsy Association, 2011, 20, 813-816.	0.9	21
59	Treatment of Hyperthyroidism in Pregnancy and Birth Defects. Obstetrical and Gynecological Survey, 2011, 66, 135-137.	0.2	0
60	First trimester diclofenac exposure and pregnancy outcome. Reproductive Toxicology, 2010, 30, 401-404.	1.3	27
61	Migraine therapy during pregnancy and lactation. Expert Opinion on Drug Safety, 2010, 9, 937-948.	1.0	23
62	Treatment of Hyperthyroidism in Pregnancy and Birth Defects. Journal of Clinical Endocrinology and Metabolism, 2010, 95, E337-E341.	1.8	139
63	Therapy of inflammatory bowel diseases in pregnancy and lactation. Expert Opinion on Drug Safety, 2009, 8, 695-707.	1.0	10
64	Medications in Pregnancy and Lactation. Obstetrics and Gynecology, 2009, 114, 166.	1.2	1