

Tommaso Pippucci

List of Publications by Year in descending order

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Version: 2024-02-01

85
papers

3,645
citations

136950

32
h-index

155660

55
g-index

91
all docs

91
docs citations

91
times ranked

8252
citing authors

#	ARTICLE	IF	CITATIONS
1	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. <i>American Journal of Human Genetics</i> , 2019, 105, 267-282.	6.2	237
2	EXCAVATOR: detecting copy number variants from whole-exome sequencing data. <i>Genome Biology</i> , 2013, 14, R120.	9.6	213
3	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. <i>European Journal of Human Genetics</i> , 2020, 28, 1602-1614.	2.8	208
4	Mutations in the 5' UTR of ANKRD26, the Ankirin Repeat Domain 26 Gene, Cause an Autosomal-Dominant Form of Inherited Thrombocytopenia, THC2. <i>American Journal of Human Genetics</i> , 2011, 88, 115-120.	6.2	200
5	Mutations in the mammalian target of rapamycin pathway regulators <i>NPRL2</i> and <i>NPRL3</i> cause focal epilepsy. <i>Annals of Neurology</i> , 2016, 79, 120-131.	5.3	190
6	Recurrent De Novo and Biallelic Variation of ATAD3A, Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	6.2	146
7	The landscape of epilepsy-related GATOR1 variants. <i>Genetics in Medicine</i> , 2019, 21, 398-408.	2.4	137
8	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. <i>Nature Communications</i> , 2019, 10, 4920.	12.8	99
9	Clinical and pathogenic features of <i>ETV6</i> -related thrombocytopenia with predisposition to acute lymphoblastic leukemia. <i>Haematologica</i> , 2016, 101, 1333-1342.	3.5	92
10	<i>H</i> - <i>M</i> : detection of runs of homozygosity from whole-exome sequencing data. <i>Bioinformatics</i> , 2014, 30, 2852-2859.	4.1	88
11	Enhanced copy number variants detection from whole-exome sequencing data using EXCAVATOR2. <i>Nucleic Acids Research</i> , 2016, 44, gkw695.	14.5	75
12	Mutations in RAD21 Disrupt Regulation of APOB in Patients With Chronic Intestinal Pseudo-Obstruction. <i>Gastroenterology</i> , 2015, 148, 771-782.e11.	1.3	71
13	Read count approach for DNA copy number variants detection. <i>Bioinformatics</i> , 2012, 28, 470-478.	4.1	67
14	A Novel Null Homozygous Mutation Confirms CACNA2D2 as a Gene Mutated in Epileptic Encephalopathy. <i>PLoS ONE</i> , 2013, 8, e82154.	2.5	67
15	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. <i>Journal of Clinical Investigation</i> , 2019, 130, 108-125.	8.2	65
16	Analysis of 339 pregnancies in 181 women with 13 different forms of inherited thrombocytopenia. <i>Haematologica</i> , 2014, 99, 1387-1394.	3.5	63
17	GATOR1 complex: the common genetic actor in focal epilepsies. <i>Journal of Medical Genetics</i> , 2016, 53, 503-510.	3.2	58
18	ACTN1-related thrombocytopenia: identification of novel families for phenotypic characterization. <i>Blood</i> , 2015, 125, 869-872.	1.4	57

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19	Epilepsy with auditory features. <i>Neurology: Genetics</i> , 2015, 1, e5.	1.9	55
20	A novel missense mutation in ANO5/TMEM16E is causative for gnathodiaphyseal dysplasia in a large Italian pedigree. <i>European Journal of Human Genetics</i> , 2013, 21, 613-619.	2.8	53
21	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. <i>Epilepsia</i> , 2019, 60, 797-806.	5.1	52
22	Homozygous <i>NOTCH3</i> null mutation and impaired <i>NOTCH3</i> signaling in recessive early-onset arteriopathy and cavitating leukoencephalopathy. <i>EMBO Molecular Medicine</i> , 2015, 7, 848-858.	6.9	48
23	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogyposis. <i>American Journal of Human Genetics</i> , 2019, 105, 689-705.	6.2	48
24	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. <i>Human Molecular Genetics</i> , 2015, 24, 3335-3347.	2.9	47
25	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogyposis. <i>American Journal of Human Genetics</i> , 2018, 102, 116-132.	6.2	46
26	Advances in genetic testing and optimization of clinical management in children and adults with epilepsy. <i>Expert Review of Neurotherapeutics</i> , 2020, 20, 251-269.	2.8	45
27	The homozygosity index (HI) approach reveals high allele frequency for Wilson disease in the Sardinian population. <i>European Journal of Human Genetics</i> , 2013, 21, 1308-1311.	2.8	43
28	<i>ALDH18A1</i> gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. <i>Brain</i> , 2016, 139, e3-e3.	7.6	42
29	Loss-of-function mutations in PTPRJ cause a new form of inherited thrombocytopenia. <i>Blood</i> , 2019, 133, 1346-1357.	1.4	40
30	De Novo SOX4 Variants Cause a Neurodevelopmental Disease Associated with Mild Dysmorphism. <i>American Journal of Human Genetics</i> , 2019, 104, 246-259.	6.2	40
31	FA2H-related disorders: a novel c.270+3A>T splice-site mutation leads to a complex neurodegenerative phenotype. <i>Developmental Medicine and Child Neurology</i> , 2011, 53, 958-961.	2.1	39
32	Bi-allelic mutations in <i>TRAPPC2L</i> result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. <i>Journal of Medical Genetics</i> , 2018, 55, 753-764.	3.2	39
33	From Whole Gene Deletion to Point Mutations of <i>EP300</i> -Positive Rubinstein-Taybi Patients: New Insights into the Mutational Spectrum and Peculiar Clinical Hallmarks. <i>Human Mutation</i> , 2016, 37, 175-183.	2.5	36
34	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. <i>Brain</i> , 2020, 143, 2380-2387.	7.6	34
35	A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype. <i>Human Molecular Genetics</i> , 2014, 23, 3607-3617.	2.9	33
36	5'UTR point substitutions and N-terminal truncating mutations of ANKRD26 in acute myeloid leukemia. <i>Journal of Hematology and Oncology</i> , 2017, 10, 18.	17.0	33

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37	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. <i>Human Genetics</i> , 2016, 135, 1117-1125.	3.8	29
38	XCAVATOR: accurate detection and genotyping of copy number variants from second and third generation whole-genome sequencing experiments. <i>BMC Genomics</i> , 2017, 18, 747.	2.8	29
39	SLFN14-related thrombocytopenia: identification within a large series of patients with inherited thrombocytopenia. <i>Thrombosis and Haemostasis</i> , 2016, 115, 1076-1079.	3.4	28
40	EX-HOM (EXome HOMozygosity): A Proof of Principle. <i>Human Heredity</i> , 2011, 72, 45-53.	0.8	27
41	Syndromic intellectual disability: A new phenotype caused by an aromatic amino acid decarboxylase gene (DDC) variant. <i>Gene</i> , 2015, 559, 144-148.	2.2	27
42	Somatic APC mosaicism and oligogenic inheritance in genetically unsolved colorectal adenomatous polyposis patients. <i>European Journal of Human Genetics</i> , 2018, 26, 387-395.	2.8	26
43	Is Focal Cortical Dysplasia/Epilepsy Caused by Somatic <i>MTOR</i> Mutations Always a Unilateral Disorder?. <i>Neurology: Genetics</i> , 2021, 7, e540.	1.9	26
44	Exploring by whole exome sequencing patients with initial diagnosis of Rubinsteinâ€“Taybi syndrome: the interconnections of epigenetic machinery disorders. <i>Human Genetics</i> , 2019, 138, 257-269.	3.8	25
45	Sleep-related hypermotor epilepsy (SHE): Contribution of known genes in 103 patients. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2020, 74, 60-64.	2.0	25
46	Wholeâ€“exome sequencing in adult patients with developmental and epileptic encephalopathy: It is never too late. <i>Clinical Genetics</i> , 2020, 98, 477-485.	2.0	25
47	Characterization and identification of hidden rare variants in the human genome. <i>BMC Genomics</i> , 2015, 16, 340.	2.8	24
48	Spontaneous remission in a Diamondâ€“Blackfan anaemia patient due to a revertant uniparental disomy ablating a <i>de novo RPS19</i> mutation. <i>British Journal of Haematology</i> , 2019, 185, 994-998.	2.5	24
49	A novel pedigree with familial cortical myoclonic tremor and epilepsy (<i>FCMTE</i>): Clinical characterization, refinement of the <i>FCMTE2</i> locus, and confirmation of a founder haplotype. <i>Epilepsia</i> , 2013, 54, 1298-1306.	5.1	23
50	Detection of Runs of Homozygosity from Whole Exome Sequencing Data: State of the Art and Perspectives for Clinical, Population and Epidemiological Studies. <i>Human Heredity</i> , 2014, 77, 63-72.	0.8	21
51	<i>PRIMA1</i> mutation: a new cause of nocturnal frontal lobe epilepsy. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 821-830.	3.7	21
52	A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. <i>Journal of Human Genetics</i> , 2017, 62, 259-264.	2.3	21
53	A new form of inherited thrombocytopenia due to monoallelic loss of function mutation in the thrombopoietin gene. <i>British Journal of Haematology</i> , 2018, 181, 698-701.	2.5	21
54	Whole Exome Sequencing allows the identification of two novel groups of Xeroderma pigmentosum in Tunisia, XP-D and XP-E: Impact on molecular diagnosis. <i>Journal of Dermatological Science</i> , 2018, 89, 172-180.	1.9	20

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55	Estimating the Allele Frequency of Autosomal Recessive Disorders through Mutational Records and Consanguinity: The Homozygosity Index (HI). <i>Annals of Human Genetics</i> , 2012, 76, 159-167.	0.8	18
56	Guideline recommendations for diagnosis and clinical management of Ring14 syndrome—first report of an ad hoc task force. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 69.	2.7	18
57	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. <i>American Journal of Human Genetics</i> , 2021, 108, 1330-1341.	6.2	18
58	<i>ACTN1</i> mutations lead to a benign form of platelet macrocytosis not always associated with thrombocytopenia. <i>British Journal of Haematology</i> , 2018, 183, 276-288.	2.5	16
59	A novel mutation in SPART gene causes a severe neurodevelopmental delay due to mitochondrial dysfunction with complex I impairments and altered pyruvate metabolism. <i>FASEB Journal</i> , 2019, 33, 11284-11302.	0.5	15
60	Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 475-485.	3.7	15
61	Prenatal diagnosis of Simpson-Golabi-Behmel syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3258-3264.	1.2	14
62	Profile of neuropsychological impairment in Sleep-related Hypermotor Epilepsy. <i>Sleep Medicine</i> , 2018, 48, 8-15.	1.6	13
63	A stereo EEG study in a patient with sleep-related hypermotor epilepsy due to DEPDC5 mutation. <i>Seizure: the Journal of the British Epilepsy Association</i> , 2017, 53, 51-54.	2.0	11
64	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. <i>American Journal of Human Genetics</i> , 2019, 105, 987-995.	6.2	11
65	Autosomal dominant partial epilepsy with auditory features: A new locus on chromosome 19q13.11-q13.31. <i>Epilepsia</i> , 2014, 55, 841-848.	5.1	10
66	BRCA1 p.His1673del is a pathogenic mutation associated with a predominant ovarian cancer phenotype. <i>Oncotarget</i> , 2017, 8, 22640-22648.	1.8	10
67	Defective lipid signalling caused by mutations in <i>PIK3C2B</i> underlies focal epilepsy. <i>Brain</i> , 2022, 145, 2313-2331.	7.6	10
68	Functional analysis of <i>TLK2</i> variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. <i>Journal of Medical Genetics</i> , 2022, 59, 170-179.	3.2	9
69	Epilepsy with auditory features: Long-term outcome and predictors of terminal remission. <i>Epilepsia</i> , 2018, 59, 834-843.	5.1	8
70	Phenotype variability of GLUT1 deficiency syndrome: Description of a case series with novel SLC2A1 gene mutations. <i>Epilepsy and Behavior</i> , 2018, 79, 169-173.	1.7	8
71	Autozygosity-driven genetic diagnosis in consanguineous families from Italy and the Greater Middle East. <i>Human Genetics</i> , 2020, 139, 1429-1441.	3.8	8
72	Distal renal tubular acidosis in a Libyan patient: Evidence for digenic inheritance. <i>European Journal of Medical Genetics</i> , 2018, 61, 1-7.	1.3	7

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73	<i>DEPDC5</i> mutations in epilepsy with auditory features. <i>Epilepsia</i> , 2016, 57, 335-335.	5.1	6
74	Loss of function mutations in <i>CCDC32</i> cause a congenital syndrome characterized by craniofacial, cardiac and neurodevelopmental anomalies. <i>Human Molecular Genetics</i> , 2020, 29, 1489-1497.	2.9	6
75	Expanding the clinical phenotype of the ultra-rare <i>Skraban-Deardorff</i> syndrome: Two novel individuals with <i>WDR26</i> loss-of-function variants and a literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1712-1720.	1.2	6
76	Accurate Detection of Hot-Spot MTOR Somatic Mutations in Archival Surgical Specimens of Focal Cortical Dysplasia by Molecular Inversion Probes. <i>Molecular Diagnosis and Therapy</i> , 2020, 24, 571-577.	3.8	5
77	A New Homozygous <i>CACNB2</i> Mutation has Functional Relevance and Supports a Role for Calcium Channels in Autism Spectrum Disorder. <i>Journal of Autism and Developmental Disorders</i> , 2021, 51, 377-381.	2.7	5
78	<i>SCN1A</i> mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. <i>Epileptic Disorders</i> , 2019, 21, 185-191.	1.3	5
79	In silico analysis of a novel causative mutation in <i>Cadherin23</i> gene identified in an Omani family with hearing loss. <i>Journal of Genetic Engineering and Biotechnology</i> , 2020, 18, 8.	3.3	4
80	AUDACITY: A comprehensive approach for the detection and classification of Runs of Homozygosity in medical and population genomics. <i>Computational and Structural Biotechnology Journal</i> , 2020, 18, 1956-1967.	4.1	3
81	Reply to ten Kate et al. <i>European Journal of Human Genetics</i> , 2014, 22, 157-158.	2.8	1
82	EX-HOM: Exome sequencing in small consanguineous pedigrees. <i>Current Opinion in Biotechnology</i> , 2011, 22, S25.	6.6	0
83	The past, present and future of consanguinity studies. <i>Current Opinion in Biotechnology</i> , 2011, 22, S26.	6.6	0
84	Somatic APC Mosaicism Causes a High Proportion of Unexplained Colorectal Polyposis. <i>Gastroenterology</i> , 2017, 152, S555.	1.3	0
85	unCOVERApp: an interactive graphical application for clinical assessment of sequence coverage at the base-pair level. <i>Bioinformatics</i> , 2021, 37, 723-725.	4.1	0