Tommaso Pippucci

List of Publications by Year
in descending order

85	3,645	136950 32	155660 55
papers	citations	h-index	g-index

91	91	91	8252
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Functional analysis of TLK2 variants and their proximal interactomes implicates impaired kinase activity and chromatin maintenance defects in their pathogenesis. Journal of Medical Genetics, 2022, 59, 170-179.	3.2	9
2	Defective lipid signalling caused by mutations inPIK3C2Bunderlies focal epilepsy. Brain, 2022, 145, 2313-2331.	7.6	10
3	A New Homozygous CACNB2 Mutation has Functional Relevance and Supports a Role for Calcium Channels in Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2021, 51, 377-381.	2.7	5
4	unCOVERApp: an interactive graphical application for clinical assessment of sequence coverage at the base-pair level. Bioinformatics, 2021, 37, 723-725.	4.1	0
5	Is Focal Cortical Dysplasia/Epilepsy Caused by Somatic MTOR Mutations Always a Unilateral Disorder?. Neurology: Genetics, 2021, 7, e540.	1.9	26
6	Expanding the clinical phenotype of the ultraâ€rare <scp>Skrabanâ€Deardorff</scp> syndrome: Two novel individuals with <scp>WDR26</scp> lossâ€ofâ€function variants and a literature review. American Journal of Medical Genetics, Part A, 2021, 185, 1712-1720.	1.2	6
7	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 1330-1341.	6.2	18
8	Sleep-related hypermotor epilepsy (SHE): Contribution of known genes in 103 patients. Seizure: the Journal of the British Epilepsy Association, 2020, 74, 60-64.	2.0	25
9	AUDACITY: A comprehensive approach for the detection and classification of Runs of Homozygosity in medical and population genomics. Computational and Structural Biotechnology Journal, 2020, 18, 1956-1967.	4.1	3
10	SLC12A2 variants cause a neurodevelopmental disorder or cochleovestibular defect. Brain, 2020, 143, 2380-2387.	7.6	34
11	ACE2 gene variants may underlie interindividual variability and susceptibility to COVID-19 in the Italian population. European Journal of Human Genetics, 2020, 28, 1602-1614.	2.8	208
12	Accurate Detection of Hot-Spot MTOR Somatic Mutations in Archival Surgical Specimens of Focal Cortical Dysplasia by Molecular Inversion Probes. Molecular Diagnosis and Therapy, 2020, 24, 571-577.	3.8	5
13	Wholeâ€exome sequencing in adult patients with developmental and epileptic encephalopathy: It is never too late. Clinical Genetics, 2020, 98, 477-485.	2.0	25
14	Autozygosity-driven genetic diagnosis in consanguineous families from Italy and the Greater Middle East. Human Genetics, 2020, 139, 1429-1441.	3.8	8
15	Advances in genetic testing and optimization of clinical management in children and adults with epilepsy. Expert Review of Neurotherapeutics, 2020, 20, 251-269.	2.8	45
16	Loss of function mutations in CCDC32 cause a congenital syndrome characterized by craniofacial, cardiac and neurodevelopmental anomalies. Human Molecular Genetics, 2020, 29, 1489-1497.	2.9	6
17	In silico analysis of a novel causative mutation in Cadherin23 gene identified in an Omani family with hearing loss. Journal of Genetic Engineering and Biotechnology, 2020, 18, 8.	3.3	4
18	The landscape of epilepsy-related GATOR1 variants. Genetics in Medicine, 2019, 21, 398-408.	2.4	137

#	Article	IF	Citations
19	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
20	A novel mutation in SPART gene causes a severe neurodevelopmental delay due to mitochondrial dysfunction with complex I impairments and altered pyruvate metabolism. FASEB Journal, 2019, 33, 11284-11302.	0.5	15
21	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
22	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
23	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
24	The Epilepsy Genetics Initiative: Systematic reanalysis of diagnostic exomes increases yield. Epilepsia, 2019, 60, 797-806.	5.1	52
25	Contribution of ultrarare variants in mTOR pathway genes to sporadic focal epilepsies. Annals of Clinical and Translational Neurology, 2019, 6, 475-485.	3.7	15
26	Exploring by whole exome sequencing patients with initial diagnosis of Rubinsteinâ€“Taybi syndrome: the interconnections of epigenetic machinery disorders. Human Genetics, 2019, 138, 257-269.	3.8	25
27	Loss-of-function mutations in PTPRJ cause a new form of inherited thrombocytopenia. Blood, 2019, 133, 1346-1357.	1.4	40
28	Spontaneous remission in a Diamondâ€Blackfan anaemia patient due to a revertant uniparental disomy ablating a de novo RPS19 mutation. British Journal of Haematology, 2019, 185, 994-998.	2.5	24
29	De Novo SOX4 Variants Cause a Neurodevelopmental Disease Associated with Mild Dysmorphism. American Journal of Human Genetics, 2019, 104, 246-259.	6.2	40
30	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
31	SCN1A mutations in focal epilepsy with auditory features: widening the spectrum of GEFS plus. Epileptic Disorders, 2019, 21, 185-191.	1.3	5
32	Epilepsy with auditory features: Longâ€term outcome and predictors of terminal remission. Epilepsia, 2018, 59, 834-843.	5.1	8
33	Somatic APC mosaicism and oligogenic inheritance in genetically unsolved colorectal adenomatous polyposis patients. European Journal of Human Genetics, 2018, 26, 387-395.	2.8	26
34	Phenotype variability of GLUT1 deficiency syndrome: Description of a case series with novel SLC2A1 gene mutations. Epilepsy and Behavior, 2018, 79, 169-173.	1.7	8
35	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132.	6.2	46
36	Profile of neuropsychological impairment in Sleep-related Hypermotor Epilepsy. Sleep Medicine, 2018, 48, 8-15.	1.6	13

#	Article	IF	Citations
37	A new form of inherited thrombocytopenia due to monoallelic loss of function mutation in the thrombopoietin gene. British Journal of Haematology, 2018, 181, 698-701.	2.5	21
38	Whole Exome Sequencing allows the identification of two novel groups of Xeroderma pigmentosum in Tunisia, XP-D and XP-E: Impact on molecular diagnosis. Journal of Dermatological Science, 2018, 89, 172-180.	1.9	20
39	Distal renal tubular acidosis in a Libyan patient: Evidence for digenic inheritance. European Journal of Medical Genetics, 2018, 61, 1-7.	1.3	7
40	<scp>ACTN</scp>1 mutations lead to a benign form of platelet macrocytosis not always associated with thrombocytopenia. British Journal of Haematology, 2018, 183, 276-288.	2.5	16
41	Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. Journal of Medical Genetics, 2018, 55, 753-764.	3.2	39
42	Guideline recommendations for diagnosis and clinical management of Ring14 syndromeâ€"first report of an ad hoc task force. Orphanet Journal of Rare Diseases, 2017, 12, 69.	2.7	18
43	Somatic APC Mosaicism Causes a High Proportion of Unexplained Colorectal Polyposis. Gastroenterology, 2017, 152, S555.	1.3	0
44	A stereo EEG study in a patient with sleep-related hypermotor epilepsy due to DEPDC5 mutation. Seizure: the Journal of the British Epilepsy Association, 2017, 53, 51-54.	2.0	11
45	5â€™UTR point substitutions and N-terminal truncating mutations of ANKRD26 in acute myeloid leukemia. Journal of Hematology and Oncology, 2017, 10, 18.	17.0	33
46	A novel founder MYO15A frameshift duplication is the major cause of genetic hearing loss in Oman. Journal of Human Genetics, 2017, 62, 259-264.	2.3	21
47	XCAVATOR: accurate detection and genotyping of copy number variants from second and third generation whole-genome sequencing experiments. BMC Genomics, 2017, 18, 747.	2.8	29
48	BRCA1 p.His1673del is a pathogenic mutation associated with a predominant ovarian cancer phenotype. Oncotarget, 2017, 8, 22640-22648.	1.8	10
49	SLFN14-related thrombocytopenia: identification within a large series of patients with inherited thrombocytopenia. Thrombosis and Haemostasis, 2016, 115, 1076-1079.	3.4	28
50	From Whole Gene Deletion to Point Mutations ofEP300-Positive Rubinstein-Taybi Patients: New Insights into the Mutational Spectrum and Peculiar Clinical Hallmarks. Human Mutation, 2016, 37, 175-183.	2.5	36
51	Identity by descent fine mapping of familial adult myoclonus epilepsy (FAME) to 2p11.2â€“2q11.2. Human Genetics, 2016, 135, 1117-1125.	3.8	29
52	<scp>DEPDC</scp>5 mutations in epilepsy with auditory features. Epilepsia, 2016, 57, 335-335.	5.1	6
53	GATOR1 complex: the common genetic actor in focal epilepsies. Journal of Medical Genetics, 2016, 53, 503-510.	3.2	58
54	Clinical and pathogenic features of ETV6 -related thrombocytopenia with predisposition to acute lymphoblastic leukemia. Haematologica, 2016, 101, 1333-1342.	3.5	92

#	Article	IF	Citations
55	Prenatal diagnosis of Simpsonâ€“Golabiâ€“Behmel syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3258-3264.	1.2	14
56	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
57	Enhanced copy number variants detection from whole-exome sequencing data using EXCAVATOR2. Nucleic Acids Research, 2016, 44, gkw695.	14.5	75
58	ALDH18A1gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism. Brain, 2016, 139, e3-e3.	7.6	42
59	Mutations in the mammalian target of rapamycin pathway regulators NPRL2 and NPRL3 cause focal epilepsy. Annals of Neurology, 2016, 79, 120-131.	5.3	190
60	PRIMA1 mutation: a new cause of nocturnal frontal lobe epilepsy. Annals of Clinical and Translational Neurology, 2015, 2, 821-830.	3.7	21
61	ACTN1-related thrombocytopenia: identification of novel families for phenotypic characterization. Blood, 2015, 125, 869-872.	1.4	57
62	Homozygous <scp>NOTCH</scp> 3 null mutation and impaired <scp>NOTCH</scp> 3 signaling in recessive earlyâ€onset arteriopathy and cavitating leukoencephalopathy. EMBO Molecular Medicine, 2015, 7, 848-858.	6.9	48
63	Epilepsy with auditory features. Neurology: Genetics, 2015, 1, e5.	1.9	55
64	Characterization and identification of hidden rare variants in the human genome. BMC Genomics, 2015, 16, 340.	2.8	24
65	Syndromic intellectual disability: A new phenotype caused by an aromatic amino acid decarboxylase gene (DDC) variant. Gene, 2015, 559, 144-148.	2.2	27
66	HCFC1 loss-of-function mutations disrupt neuronal and neural progenitor cells of the developing brain. Human Molecular Genetics, 2015, 24, 3335-3347.	2.9	47
67	Mutations in RAD21 Disrupt Regulation of APOB in Patients With Chronic Intestinal Pseudo-Obstruction. Gastroenterology, 2015, 148, 771-782.e11.	1.3	71
68	Detection of Runs of Homozygosity from Whole Exome Sequencing Data: State of the Art and Perspectives for Clinical, Population and Epidemiological Studies. Human Heredity, 2014, 77, 63-72.	0.8	21
69	Autosomal dominant partial epilepsy with auditory features: A new locus on chromosome 19q13.11â€“q13.31. Epilepsia, 2014, 55, 841-848.	5.1	10
70	A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype. Human Molecular Genetics, 2014, 23, 3607-3617.	2.9	33
71	Reply to ten Kate et al. European Journal of Human Genetics, 2014, 22, 157-158.	2.8	1
72	H Â3 ÂM Â2 : detection of runs of homozygosity from whole-exome sequencing data. Bioinformatics, 2014, 30, 2852-2859.	4.1	88

#	Article	IF	Citations
73	Analysis of 339 pregnancies in 181 women with 13 different forms of inherited thrombocytopenia. Haematologica, 2014, 99, 1387-1394.	3.5	63
74	The homozygosity index (HI) approach reveals high allele frequency for Wilson disease in the Sardinian population. European Journal of Human Genetics, 2013, 21, 1308-1311.	2.8	43
75	A novel pedigree with familial cortical myoclonic tremor and epilepsy (<scp>FCMTE</scp>): Clinical characterization, refinement of the <scp>FCMTE</scp>2 locus, and confirmation of a founder haplotype. Epilepsia, 2013, 54, 1298-1306.	5.1	23
76	A novel missense mutation in ANO5/TMEM16E is causative for gnathodiaphyseal dyplasia in a large Italian pedigree. European Journal of Human Genetics, 2013, 21, 613-619.	2.8	53
77	EXCAVATOR: detecting copy number variants from whole-exome sequencing data. Genome Biology, 2013, 14, R120.	9.6	213
78	A Novel Null Homozygous Mutation Confirms CACNA2D2 as a Gene Mutated in Epileptic Encephalopathy. PLoS ONE, 2013, 8, e82154.	2.5	67
79	Estimating the Allele Frequency of Autosomal Recessive Disorders through Mutational Records and Consanguinity: The Homozygosity Index (HI). Annals of Human Genetics, 2012, 76, 159-167.	0.8	18
80	Read count approach for DNA copy number variants detection. Bioinformatics, 2012, 28, 470-478.	4.1	67
81	EX-HOM (EXome HOMozygosity): A Proof of Principle. Human Heredity, 2011, 72, 45-53.	0.8	27
82	FA2H-related disorders: a novel c.270+3A>T splice-site mutation leads to a complex neurodegenerative phenotype. Developmental Medicine and Child Neurology, 2011, 53, 958-961.	2.1	39
83	Mutations in the 5â€² UTR of ANKRD26, the Ankirin Repeat Domain 26 Gene, Cause an Autosomal-Dominant Form of Inherited Thrombocytopenia, THC2. American Journal of Human Genetics, 2011, 88, 115-120.	6.2	200
84	EX-HOM: Exome sequencing in small consanguineous pedigrees. Current Opinion in Biotechnology, 2011, 22, S25.	6.6	0
85	The past, present and future of consanguinity studies. Current Opinion in Biotechnology, 2011, 22, S26.	6.6	0