Tomasz Gambin

List of Publications by Year
in descending order

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112	6,610	61984	74163
papers	citations	43 h-index	75 g-index

119	119	119	13564
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Exacerbation of mild lung disorders to lethal pulmonary hypoplasia by a noncoding hypomorphic <scp>SNV</scp> in a lungâ€specific enhancer in trans to the frameshifting <scp>TBX4</scp> variant. American Journal of Medical Genetics, Part A, 2022, 188, 1420-1425.	1.2	7
2	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	2.4	44
3	The Thousand Polish Genomesâ€"A Database of Polish Variant Allele Frequencies. International Journal of Molecular Sciences, 2022, 23, 4532.	4.1	15
4	Variants in the pancreatic CUB and zona pellucida-like domains 1 (CUZD1) gene in early-onset chronic pancreatitis - A possible new susceptibility gene. Pancreatology, 2022, 22, 564-571.	1.1	4
5	Do paternal deletions involving the FOXF1 locus on chromosome 16q24.1 manifest with more severe non-lung anomalies?. European Journal of Medical Genetics, 2022, 65, 104519.	1.3	3
6	Transcriptome and Immunohistochemical Analyses in TBX4- and FGF10-Deficient Lungs Imply TMEM100 as a Mediator of Human Lung Development. American Journal of Respiratory Cell and Molecular Biology, 2022, 66, 694-697.	2.9	6
7	Potential interactions between the TBX4-FGF10 and SHH-FOXF1 signaling during human lung development revealed using ChIP-seq. Respiratory Research, 2021, 22, 26.	3.6	11
8	Variants in FLRT3 and SLC35E2B identified using exome sequencing in seven high myopia families from Central Europe. Advances in Medical Sciences, 2021, 66, 192-198.	2.1	5
9	Lungâ€specific distant enhancer cis regulates expression ofFOXF1and lncRNAFENDRR. Human Mutation, 2021, 42, 694-698.	2.5	10
10	Perturbation of semaphorin and VEGF signaling in ACDMPV lungs due to FOXF1 deficiency. Respiratory Research, 2021, 22, 212.	3.6	11
11	Loss of function TRPV6 variants are associated with chronic pancreatitis in nonalcoholic early-onset Polish and German patients. Pancreatology, 2021, 21, 1434-1442.	1.1	10
12	Detection of low-level parental somatic mosaicism for clinically relevant SNVs and indels identified in a large exome sequencing dataset. Human Genomics, 2021, 15, 72.	2.9	11
13	Low-level parental somatic mosaic SNVs in exomes from a large cohort of trios with diverse suspected Mendelian conditions. Genetics in Medicine, 2020, 22, 1768-1776.	2.4	30
14	Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498.	1.9	11
15	A de novo 2.2â€‰Mb recurrent 17q23.1q23.2 deletion unmasks novel putative regulatory non-coding SNVs associated with lethal lung hypoplasia and pulmonary hypertension: a case report. BMC Medical Genomics, 2020, 13, 34.	1.5	12
16	<scp>Wolffâ€"Parkinsonâ€"White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	1.2	14
17	Accumulation of sequence variants in genes of Wnt signaling and focal adhesion pathways in human corneas further explains their involvement in keratoconus. PeerJ, 2020, 8, e8982.	2.0	12
18	SeQuiLa-cov: A fast and scalable library for depth of coverage calculations. GigaScience, 2019, 8, .	6.4	8

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19	A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48.	8.2	55
20	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	6.2	24
21	Comparison of kNN and k-means optimization methods of reference set selection for improved CNV callers performance. BMC Bioinformatics, 2019, 20, 266.	2.6	13
22	Clinical, Histopathological, and Molecular Diagnostics in Lethal Lung Developmental Disorders. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 1093-1101.	5.6	47
23	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 2296-2299.	2.9	87
24	Familial ataxia, tremor, and dementia in a polish family with a novel mutation in the CCDC88C gene. Movement Disorders, 2019, 34, 142-144.	3.9	10
25	SeQuiLa: an elastic, fast and scalable SQL-oriented solution for processing and querying genomic intervals. Bioinformatics, 2019, 35, 2156-2158.	4.1	9
26	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
27	A Patient with Berardinelli-Seip Syndrome, Novel <i>AGPAT2</i> Splicesite Mutation and Concomitant Development of Non-diabetic Polyneuropathy. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 319-326.	0.9	6
28	DNA methylation signature in blood does not predict calendar age in patients with chronic lymphocytic leukemia but may alert to the presence of disease. Forensic Science International: Genetics, 2018, 34, e15-e17.	3.1	13
29	The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075.	2.9	16
30	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	2.8	52
31	Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131.	2.8	35
32	Phenotype expansion and development in Kosaki overgrowth syndrome. Clinical Genetics, 2018, 93, 919-924.	2.0	17
33	TADeus-a tool for clinical interpretation of structural variants modifying chromatin organization. , 2018, , .		3
34	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	6.2	160
35	iGAP: Interactive Genomic Analysis Platform. , 2018, , .		0
36	Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237.	14.5	98

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37	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
38	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	28.9	66
39	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	3.8	36
40	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
41	Iterative Sequencing and Variant Screening (ISVS) as a novel pathogenic mutations search strategy - application for TMPRSS3 mutations screen. Scientific Reports, 2017, 7, 2543.	3.3	10
42	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. Genetics in Medicine, 2017, 19, 412-420.	2.4	73
43	Variants in SKP1, PROB1, and IL17B genes at keratoconus 5q31.1â€“q35.3 susceptibility locus identified by whole-exome sequencing. European Journal of Human Genetics, 2017, 25, 73-78.	2.8	19
44	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
45	Benchmarking distributed data warehouse solutions for storing genomic variant information. Database: the Journal of Biological Databases and Curation, 2017, 2017, .	3.0	8
46	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
47	Scalable Framework for the Analysis of Population Structure Using the Next Generation Sequencing Data. Lecture Notes in Computer Science, 2017, , 471-480.	1.3	2
48	Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778.	8.2	82
49	Evidence against <scp>ZNF</scp>469 being causative for keratoconus in Polish patients. Acta Ophthalmologica, 2016, 94, 289-294.	1.1	20
50	A potential founder variant inCARMIL2/RLTPRin three Norwegian families with warts, molluscum contagiosum, and T-cell dysfunction. Molecular Genetics & Genomic Medicine, 2016, 4, 604-616.	1.2	59
51	MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106.	8.2	43
52	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
53	CAV3 mutation in a patient with transient hyperCKemia and myalgia. Neurologia I Neurochirurgia Polska, 2016, 50, 468-473.	1.2	8
54	Hutteriteâ€type cataract maps to chromosome 6p21.32â€p21.31, cosegregates with a homozygous mutation in <scp>LEMD</scp>2, and is associated with sudden cardiac death. Molecular Genetics & Genomic Medicine, 2016, 4, 77-94.	1.2	28

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55	Exome sequencing in mostly consanguineous Arab families with neurologic disease provides a high potential molecular diagnosis rate. BMC Medical Genomics, 2016, 9, 42.	1.5	80
56	PEHO Syndrome May Represent Phenotypic Expansion at the Severe End of the Early-Onset Encephalopathies. Pediatric Neurology, 2016, 60, 83-87.	2.1	25
57	Two male sibs with severe micrognathia and a missense variant in MED12. European Journal of Medical Genetics, 2016, 59, 367-372.	1.3	11
58	Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685.	2.4	186
59	Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. Human Mutation, 2016, 37, 231-234.	2.5	18
60	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
61	Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68.	9.0	71
62	Secondary findings and carrier test frequencies in a large multiethnic sample. Genome Medicine, 2015, 7, 54.	8.2	47
63	Rare variants in the notch signaling pathway describe a novel type of autosomal recessive Klippelâ€“Feil syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2795-2799.	1.2	47
64	Comparative Genomic Analyses of the Human NPHP1 Locus Reveal Complex Genomic Architecture and Its Regional Evolution in Primates. PLoS Genetics, 2015, 11, e1005686.	3.5	21
65	Whole Exome Sequencing Identifies an Adult-Onset Case of Methylmalonic Aciduria and Homocystinuria Type C (cblC) with Non-Syndromic Bullâ€™s Eye Maculopathy. Ophthalmic Genetics, 2015, 36, 270-275.	1.2	17
66	Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183.	6.4	211
67	Genome-wide analyses of LINEâ€“LINE-mediated nonallelic homologous recombination. Nucleic Acids Research, 2015, 43, 2188-2198.	14.5	79
68	Whole-Exome Sequencing Identifies Homozygous GPR161 Mutation in a Family with Pituitary Stalk Interruption Syndrome. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E140-E147.	3.6	60
69	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	6.2	574
70	Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. PLoS Genetics, 2015, 11, e1005050.	3.5	57
71	COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660.	21.4	302
72	Exome sequencing identifies a homozygous C5orf42 variant in a Turkish kindred with oralâ€facialâ€digital syndrome type VI. American Journal of Medical Genetics, Part A, 2015, 167, 2132-2137.	1.2	12

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73	New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133.	2.5	28
74	Homozygous Loss-of-function Mutations inSOHLH1in Patients With Nonsyndromic Hypergonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E808-E814.	3.6	29
75	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	6.2	110
76	FBN1 contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836.	1.2	24
77	Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513.	8.1	258
78	Social cognition, psychopathological symptoms, and family functioning in a sample of inpatient adolescents using variableâ€centered and personâ€centered approaches. Journal of Adolescence, 2015, 45, 31-43.	2.4	14
79	Alpha-Thalassemia Mutations in Adana Province, Southern Turkey: Genotype-Phenotype Correlation. Indian Journal of Hematology and Blood Transfusion, 2015, 31, 223-228.	0.6	14
80	Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651.	8.2	136
81	Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. BMC Biology, 2014, 12, 74.	3.8	60
82	Molecular and clinical analyses of 16q24.1 duplications involving FOXF1 identify an evolutionarily unstable large minisatellite. BMC Medical Genetics, 2014, 15, 128.	2.1	11
83	Whole-exome sequencing links TMCO1 defect syndrome with cerebro-facio-thoracic dysplasia. European Journal of Human Genetics, 2014, 22, 1145-1148.	2.8	19
84	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258.	3.5	122
85	Rule-Based Algorithm Transforming OWL Ontology Into Relational Database. Communications in Computer and Information Science, 2014, , 148-159.	0.5	2
86	Compound Heterozygous CORO1A Mutations in Siblings with a Mucocutaneous-Immunodeficiency Syndrome of Epidermodysplasia Verruciformis-HPV, Molluscum Contagiosum and Granulomatous Tuberculoid Leprosy. Journal of Clinical Immunology, 2014, 34, 871-890.	3.8	78
87	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	2.8	112
88	Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650.	28.9	189
89	Application of array comparative genomic hybridization in 256 patients with developmental delay or intellectual disability. Journal of Applied Genetics, 2014, 55, 125-144.	1.9	37
90	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	28.9	322

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91	Whole exome sequencing identifies three novel mutations in ANTXR1 in families with GAPO syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2328-2334.	1.2	20
92	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978.	2.8	51
93	New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780.	2.9	30
94	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	6.2	148
95	Multiple samples aCGH analysis for rare CNVs detection. Journal of Clinical Bioinformatics, 2013, 3, 12.	1.2	2
96	Functional performance of aCGH design for clinical cytogenetics. Computers in Biology and Medicine, 2013, 43, 775-785.	7.0	4
97	Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. Human Mutation, 2013, 34, 210-220.	2.5	48
98	Application of custom-designed oligonucleotide array CGH in 145 patients with autistic spectrum disorders. European Journal of Human Genetics, 2013, 21, 620-625.	2.8	37
99	TIRfinder: A Web Tool for Mining Class II Transposons Carrying Terminal Inverted Repeats. Evolutionary Bioinformatics, 2013, 9, EBO.S10619.	1.2	7
100	Confounding by Repetitive Elements and CpG Islands Does Not Explain the Association between Hypomethylation and Genomic Instability. PLoS Genetics, 2013, 9, e1003333.	3.5	3
101	NAHR-mediated copy-number variants in a clinical population: Mechanistic insights into both genomic disorders and Mendelizing traits. Genome Research, 2013, 23, 1395-1409.	5.5	120
102	Genomic Hypomethylation in the Human Germline Associates with Selective Structural Mutability in the Human Genome. PLoS Genetics, 2012, 8, e1002692.	3.5	80
103	Detection of â‰¥1â€‰Mb microdeletions and microduplications in a single cell using custom oligonucleotide arrays. Prenatal Diagnosis, 2012, 32, 10-20.	2.3	29
104	Application of array comparative genomic hybridization in 102 patients with epilepsy and additional neurodevelopmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 760-771.	1.7	48
105	Assessment of the role of copy-number variants in 150 patients with congenital heart defects. , 2012, 16, 175-82.		12
106	Efficient Multiple Samples aCGH Analysis for Rare CNVs Detection. , 2011, , .		0
107	Use of array CGH to detect exonic copy number variants throughout the genome in autism families detects a novel deletion in TMLHE. Human Molecular Genetics, 2011, 20, 4360-4370.	2.9	101
108	Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342.	2.5	225

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109	A new classification method using array Comparative Genome Hybridization data, based on the concept of Limited Jumping Emerging Patterns. BMC Bioinformatics, 2009, 10, S64.	2.6	4
110	Population dynamics of miniature inverted-repeat transposable elements (MITEs) in Medicago truncatula. Gene, 2009, 448, 214-220.	2.2	30
111	Classification based on the highest impact jumping emerging patterns. , 2009, , .		3
112	Diversity and structure of PIF/Harbinger-like elements in the genome of Medicago truncatula. BMC Genomics, 2007, 8, 409.	2.8	25