# 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 | The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215. | 6.2 | 574 |
| 2 | A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214. | 28.9 | 322 |
| 3 | COPA mutations impair ER-Golgi transport and cause hereditary autoimmune-mediated lung disease and arthritis. Nature Genetics, 2015, 47, 654-660. | 21.4 | 302 |
| 4 | Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245. | 2.9 | 261 |
| 5 | Genes that Affect Brain Structure and Function Identified by Rare Variant Analyses of Mendelian Neurologic Disease. Neuron, 2015, 88, 499-513. | 8.1 | 258 |
| 6 | Detection of clinically relevant exonic copy-number changes by array CGH. Human Mutation, 2010, 31, 1326-1342. | 2.5 | 225 |
| 7 | Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. Cell Reports, 2015, 12, 1169-1183. | 6.4 | 211 |
| 8 | Human CLP1 Mutations Alter tRNA Biogenesis, Affecting Both Peripheral and Central Nervous System Function. Cell, 2014, 157, 636-650. | 28.9 | 189 |
| 9 | Molecular diagnostic experience of whole-exome sequencing in adult patients. Genetics in Medicine, 2016, 18, 678-685. | 2.4 | 186 |
| 10 | Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26. | 8.2 | 184 |
| 11 | 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 |
| 12 | 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 |
| 13 | Global transcriptional disturbances underlie Cornelia de Lange syndrome and related phenotypes. Journal of Clinical Investigation, 2015, 125, 636-651. | 8.2 | 136 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | 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 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Homozygous and hemizygous CNV detection from exome sequencing data in a Mendelian disease cohort. Nucleic Acids Research, 2017, 45, gkw1237. | 14.5 | 98 |
| 20 | 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 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586. | 3.8 | 85 |
| 25 | Molecular etiology of arthrogryposis in multiple families of mostly Turkish origin. Journal of Clinical Investigation, 2016, 126, 762-778. | 8.2 | 82 |
| 26 | Genomic Hypomethylation in the Human Germline Associates with Selective Structural Mutability in the Human Genome. PLoS Genetics, 2012, 8, e1002692. | 3.5 | 80 |
| 27 | 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 |
| 28 | Genome-wide analyses of LINEâ€“LINE-mediated nonallelic homologous recombination. Nucleic Acids Research, 2015, 43, 2188-2198. | 14.5 | 79 |
| 29 | 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 |
| 30 | 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 |
| 31 | Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68. | 9.0 | 71 |
| 32 | An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7. | 28.9 | 66 |
| 33 | Human endogenous retroviral elements promote genome instability via non-allelic homologous recombination. BMC Biology, 2014, 12, 74. | 3.8 | 60 |
| 34 | 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 |
| 35 | 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 |
| 36 | Complex Genomic Rearrangements at the PLP1 Locus Include Triplication and Quadruplication. PLoS Genetics, 2015, 11, e1005050. | 3.5 | 57 |

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|---|---|---|---|
| 37 | A clinical survey of mosaic single nucleotide variants in disease-causing genes detected by exome sequencing. Genome Medicine, 2019, 11, 48. | 8.2 | 55 |
| 38 | Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339. | 2.8 | 52 |
| 39 | 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 |
| 40 | Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83. | 8.2 | 50 |
| 41 | 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 |
| 42 | Inverted Low-Copy Repeats and Genome Instability-A Genome-Wide Analysis. Human Mutation, 2013, 34, 210-220. | 2.5 | 48 |
| 43 | Secondary findings and carrier test frequencies in a large multiethnic sample. Genome Medicine, 2015, 7, 54. | 8.2 | 47 |
| 44 | 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 |
| 45 | 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 |
| 46 | Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797. | 2.4 | 44 |
| 47 | MIPEP recessive variants cause a syndrome of left ventricular non-compaction, hypotonia, and infantile death. Genome Medicine, 2016, 8, 106. | 8.2 | 43 |
| 48 | 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 |
| 49 | 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 |
| 50 | 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 |
| 51 | Comprehensive genomic analysis of patients with disorders of cerebral cortical development. European Journal of Human Genetics, 2018, 26, 1121-1131. | 2.8 | 35 |
| 52 | Population dynamics of miniature inverted-repeat transposable elements (MITEs) in Medicago truncatula. Gene, 2009, 448, 214-220. | 2.2 | 30 |
| 53 | New syndrome with retinitis pigmentosa is caused by nonsense mutations in retinol dehydrogenase RDH11. Human Molecular Genetics, 2014, 23, 5774-5780. | 2.9 | 30 |
| 54 | 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 |

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|---|---|---|---|
| 55 | 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 |
| 56 | Homozygous Loss-of-function Mutations inSOHLH1in Patients With Nonsyndromic Hypergonadotropic Hypogonadism. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E808-E814. | 3.6 | 29 |
| 57 | New Mutations in theRAB28Gene in 2 Spanish Families With Cone-Rod Dystrophy. JAMA Ophthalmology, 2015, 133, 133. | 2.5 | 28 |
| 58 | 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 |
| 59 | Diversity and structure of PIF/Harbinger-like elements in the genome of Medicago truncatula. BMC Genomics, 2007, 8, 409. | 2.8 | 25 |
| 60 | PEHO Syndrome May Represent Phenotypic Expansion at the Severe End of the Early-Onset Encephalopathies. Pediatric Neurology, 2016, 60, 83-87. | 2.1 | 25 |
| 61 | FBN1 contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836. | 1.2 | 24 |
| 62 | Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015. | 6.2 | 24 |
| 63 | 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 |
| 64 | 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 |
| 65 | Evidence against <scp>ZNF</scp>469 being causative for keratoconus in Polish patients. Acta Ophthalmologica, 2016, 94, 289-294. | 1.1 | 20 |
| 66 | Whole-exome sequencing links TMCO1 defect syndrome with cerebro-facio-thoracic dysplasia. European Journal of Human Genetics, 2014, 22, 1145-1148. | 2.8 | 19 |
| 67 | 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 |
| 68 | Multiallelic Positions in the Human Genome: Challenges for Genetic Analyses. Human Mutation, 2016, 37, 231-234. | 2.5 | 18 |
| 69 | 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 |
| 70 | Phenotype expansion and development in Kosaki overgrowth syndrome. Clinical Genetics, 2018, 93, 919-924. | 2.0 | 17 |
| 71 | The role of FREM2 and FRAS1 in the development of congenital diaphragmatic hernia. Human Molecular Genetics, 2018, 27, 2064-2075. | 2.9 | 16 |
| 72 | The Thousand Polish Genomesâ€”A Database of Polish Variant Allele Frequencies. International Journal of Molecular Sciences, 2022, 23, 4532. | 4.1 | 15 |

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|---|---|---|---|
| 73 | 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 |
| 74 | 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 |
| 75 | <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 |
| 76 | 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 |
| 77 | 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 |
| 78 | 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 |
| 79 | 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 |
| 80 | 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 |
| 81 | Assessment of the role of copy-number variants in 150 patients with congenital heart defects. , 2012, 16, 175-82. | | 12 |
| 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 | Two male sibs with severe micrognathia and a missense variant in MED12. European Journal of Medical Genetics, 2016, 59, 367-372. | 1.3 | 11 |
| 84 | Integrated sequencing and array comparative genomic hybridization in familial Parkinson disease. Neurology: Genetics, 2020, 6, e498. | 1.9 | 11 |
| 85 | 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 |
| 86 | Perturbation of semaphorin and VEGF signaling in ACDMPV lungs due to FOXF1 deficiency. Respiratory Research, 2021, 22, 212. | 3.6 | 11 |
| 87 | 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 |
| 88 | 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 |
| 89 | 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 |
| 90 | Lungâ€specific distant enhancer cis regulates expression ofFOXF1and lncRNAFENDRR. Human Mutation, 2021, 42, 694-698. | 2.5 | 10 |

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|---|---|---|---|
| 91 | 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 |
| 92 | SeQuiLa: an elastic, fast and scalable SQL-oriented solution for processing and querying genomic intervals. Bioinformatics, 2019, 35, 2156-2158. | 4.1 | 9 |
| 93 | CAV3 mutation in a patient with transient hyperCKemia and myalgia. Neurologia I Neurochirurgia Polska, 2016, 50, 468-473. | 1.2 | 8 |
| 94 | Benchmarking distributed data warehouse solutions for storing genomic variant information. Database: the Journal of Biological Databases and Curation, 2017, 2017, . | 3.0 | 8 |
| 95 | SeQuiLa-cov: A fast and scalable library for depth of coverage calculations. GigaScience, 2019, 8, . | 6.4 | 8 |
| 96 | TIRfinder: A Web Tool for Mining Class II Transposons Carrying Terminal Inverted Repeats. Evolutionary Bioinformatics, 2013, 9, EBO.S10619. | 1.2 | 7 |
| 97 | 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 |
| 98 | 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 |
| 99 | 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 |
| 100 | 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 |
| 101 | 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 |
| 102 | Functional performance of aCGH design for clinical cytogenetics. Computers in Biology and Medicine, 2013, 43, 775-785. | 7.0 | 4 |
| 103 | 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 |
| 104 | Classification based on the highest impact jumping emerging patterns. , 2009, , . | | 3 |
| 105 | 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 |
| 106 | TADeus-a tool for clinical interpretation of structural variants modifying chromatin organization. , 2018, , . | | 3 |
| 107 | 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 |
| 108 | Multiple samples aCGH analysis for rare CNVs detection. Journal of Clinical Bioinformatics, 2013, 3, 12. | 1.2 | 2 |

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|---|---|---|---|
| 109 | Rule-Based Algorithm Transforming OWL Ontology Into Relational Database. Communications in Computer and Information Science, 2014, , 148-159. | 0.5 | 2 |
| 110 | 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 |
| 111 | Efficient Multiple Samples aCGH Analysis for Rare CNVs Detection. , 2011, , . | | 0 |
| 112 | iGAP: Interactive Genomic Analysis Platform. , 2018, , . | | 0 |