

# Tomasz Zemojtel

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4212732/publications.pdf>

Version: 2024-02-01

47  
papers

4,142  
citations

257450

24  
h-index

223800

46  
g-index

47  
all docs

47  
docs citations

47  
times ranked

9016  
citing authors

#	ARTICLE	IF	CITATIONS
1	CLN8 Mutations Presenting with a Phenotypic Continuum of Neuronal Ceroid Lipofuscinosis Literature Review and Case Report. <i>Genes</i> , 2021, 12, 956.	2.4	3
2	Increased risk of severe clinical course of COVID-19 in carriers of HLA-C*04:01. <i>EClinicalMedicine</i> , 2021, 40, 101099.	7.1	52
3	Longitudinal therapy monitoring of ALK-positive lung cancer by combined copy number and targeted mutation profiling of cell-free DNA. <i>EBioMedicine</i> , 2020, 62, 103103.	6.1	32
4	SEMA3A and IGSF10 Are Novel Contributors to Combined Pituitary Hormone Deficiency (CPHD). <i>Frontiers in Endocrinology</i> , 2020, 11, 368.	3.5	13
5	Pretreatment with methanolic extract of <i>Pistacia lentiscus</i> L. increases sensitivity to DNA damaging drugs in primary high-grade serous ovarian cancer cells. <i>European Journal of Integrative Medicine</i> , 2020, 37, 101163.	1.7	1
6	Stable expansion of high-grade serous ovarian cancer organoids requires a low Wnt environment. <i>EMBO Journal</i> , 2020, 39, e104013.	7.8	70
7	The phenotype-driven computational analysis yields clinical diagnosis for patients with atypical manifestations of known intellectual disability syndromes. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1263.	1.2	15
8	Detection of TP53 Mutations in Tissue or Liquid Rebiopsies at Progression Identifies ALK+ Lung Cancer Patients with Poor Survival. <i>Cancers</i> , 2019, 11, 124.	3.7	36
9	Role of Donor Clonal Hematopoiesis in Allogeneic Hematopoietic Stem-Cell Transplantation. <i>Journal of Clinical Oncology</i> , 2019, 37, 375-385.	1.6	163
10	Multi-Parameter Analysis of Biobanked Human Bone Marrow Stromal Cells Shows Little Influence for Donor Age and Mild Comorbidities on Phenotypic and Functional Properties. <i>Frontiers in Immunology</i> , 2019, 10, 2474.	4.8	64
11	Serial liquid biopsies for detection of treatment failure and profiling of resistance mechanisms in <i>KLC1</i> -rearranged lung cancer. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a004630.	1.2	13
12	Genomic landscape and clonal evolution of acute myeloid leukemia with t(8;21): an international study on 331 patients. <i>Blood</i> , 2019, 133, 1140-1151.	1.4	96
13	Phenotype of two Polish patients with Schaaf-Yang syndrome confirmed by identifying mutation in <i>MAGEL2</i> gene. <i>Clinical Dysmorphology</i> , 2018, 27, 49-52.	0.3	13
14	Hematopoietic lineage distribution and evolutionary dynamics of clonal hematopoiesis. <i>Leukemia</i> , 2018, 32, 1908-1919.	7.2	137
15	Ocular findings in Loays-Dietz syndrome. <i>British Journal of Ophthalmology</i> , 2018, 102, 1036-1040.	3.9	18
16	The Human Phenotype Ontology in 2017. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	14.5	699
17	Mutations in proteasome-related genes are associated with thyroid hemiagenesis. <i>Endocrine</i> , 2017, 56, 279-285.	2.3	18
18	The germline variants in DNA repair genes in pediatric medulloblastoma: a challenge for current therapeutic strategies. <i>BMC Cancer</i> , 2017, 17, 239.	2.6	12

#	ARTICLE	IF	CITATIONS
19	Clonal Hematopoiesis: Cell of Origin, Lineage Repartition and Dynamic Evolution during Chemotherapy. <i>Blood</i> , 2017, 130, 632-632.	1.4	7
20	Identification of a molecular defect in a stillborn fetus with perinatal lethal hypophosphatasia using a disease-associated genome sequencing approach. <i>Polish Journal of Pathology</i> , 2016, 1, 78-83.	0.3	3
21	<i>BRAT1</i> mutations are associated with infantile epileptic encephalopathy, mitochondrial dysfunction, and survival into childhood. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2274-2281.	1.2	25
22	Alternate-locus aware variant calling in whole genome sequencing. <i>Genome Medicine</i> , 2016, 8, 130.	8.2	16
23	Exome sequencing reveals two novel compound heterozygous <i>XYLT1</i> mutations in a Polish patient with Desbuquois dysplasia type 2 and growth hormone deficiency. <i>Journal of Human Genetics</i> , 2016, 61, 577-583.	2.3	18
24	Clinical characterization and mutation spectrum of German patients with familial hypercholesterolemia. <i>Atherosclerosis</i> , 2016, 253, 88-93.	0.8	35
25	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. <i>American Journal of Human Genetics</i> , 2016, 99, 595-606.	6.2	223
26	Next-generation diagnostics and disease-gene discovery with the Exomiser. <i>Nature Protocols</i> , 2015, 10, 2004-2015.	12.0	296
27	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. <i>American Journal of Human Genetics</i> , 2015, 97, 111-124.	6.2	203
28	<i>FGFR2</i> mutation in a patient without typical features of Pfeiffer syndrome – The emerging role of combined NGS and phenotype based strategies. <i>European Journal of Medical Genetics</i> , 2015, 58, 376-380.	1.3	9
29	Somatic neurofibromatosis type 1 (NF1) inactivation events in cutaneous neurofibromas of a single NF1 patient. <i>European Journal of Human Genetics</i> , 2015, 23, 870-873.	2.8	20
30	Recessive Inheritance of Population-Specific Intronic LINE-1 Insertion Causes a Rotor Syndrome Phenotype. <i>Human Mutation</i> , 2015, 36, 327-332.	2.5	28
31	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. <i>American Journal of Human Genetics</i> , 2015, 97, 483-492.	6.2	70
32	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. <i>Bioinformatics</i> , 2014, 30, 3215-3222.	4.1	91
33	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. <i>Science Translational Medicine</i> , 2014, 6, 252ra123.	12.4	223
34	Whole exome sequencing identifies <i>FGF16</i> nonsense mutations as the cause of X-linked recessive metacarpal 4/5 fusion. <i>Journal of Medical Genetics</i> , 2013, 50, 579-584.	3.2	31
35	P53 Binding Sites in Transposons. <i>Frontiers in Genetics</i> , 2012, 3, 40.	2.3	3
36	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. <i>Nature</i> , 2011, 478, 57-63.	27.8	805

#	ARTICLE	IF	CITATIONS
37	Systems-Level Evidence of Transcriptional Co-Regulation of Yeast Protein Complexes. <i>Journal of Computational Biology</i> , 2009, 16, 331-339.	1.6	7
38	Methylation and deamination of CpGs generate p53-binding sites on a genomic scale. <i>Trends in Genetics</i> , 2009, 25, 63-66.	6.7	61
39	The Spt-Ada-Gcn5-acetyltransferase complex interaction motif of E2a is essential for a subset of transcriptional and oncogenic properties of E2a-Pbx1. <i>Leukemia and Lymphoma</i> , 2009, 50, 816-828.	1.3	9
40	Corrigendum to "Mammalian mitochondrial nitric oxide synthase: Characterization of a novel candidate" [FEBS Lett. 580 (2006) 455-462]. <i>FEBS Letters</i> , 2007, 581, 2072-2073.	2.8	0
41	Rap1A-Deficient T and B Cells Show Impaired Integrin-Mediated Cell Adhesion. <i>Molecular and Cellular Biology</i> , 2006, 26, 643-653.	2.3	103
42	Plant nitric oxide synthase: a never-ending story?. <i>Trends in Plant Science</i> , 2006, 11, 524-525.	8.8	297
43	A novel conserved family of nitric oxide synthase?. <i>Trends in Biochemical Sciences</i> , 2004, 29, 224-226.	7.5	41
44	ECEPE proteins: a novel family of eukaryotic cysteine proteinases. <i>Trends in Biochemical Sciences</i> , 2004, 29, 524-526.	7.5	3
45	NO-Bound Myoglobin: Structural Diversity and Dynamics of the NO Ligand. <i>Journal of the American Chemical Society</i> , 2004, 126, 1930-1931.	13.7	26
46	In search of the prototype of nitric oxide synthase. <i>FEBS Letters</i> , 2003, 554, 1-5.	2.8	24
47	Kinetics of CO and NO Ligation with the Cys331 → Ala Mutant of Neuronal Nitric-oxide Synthase. <i>Journal of Biological Chemistry</i> , 2001, 276, 4733-4736.	3.4	10