Tomasz Zemojtel

List of Publications by Year in descending order

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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	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	27.8	805
2	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	14.5	699
3	Plant nitric oxide synthase: a never-ending story?. Trends in Plant Science, 2006, 11, 524-525.	8.8	297
4	Next-generation diagnostics and disease-gene discovery with the Exomiser. Nature Protocols, 2015, 10, 2004-2015.	12.0	296
5	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. Science Translational Medicine, 2014, 6, 252ra123.	12.4	223
6	A Whole-Genome Analysis Framework for Effective Identification of Pathogenic Regulatory Variants in Mendelian Disease. American Journal of Human Genetics, 2016, 99, 595-606.	6.2	223
7	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. American Journal of Human Genetics, 2015, 97, 111-124.	6.2	203
8	Role of Donor Clonal Hematopoiesis in Allogeneic Hematopoietic Stem-Cell Transplantation. Journal of Clinical Oncology, 2019, 37, 375-385.	1.6	163
9	Hematopoietic lineage distribution and evolutionary dynamics of clonal hematopoiesis. Leukemia, 2018, 32, 1908-1919.	7.2	137
10	Rap1A-Deficient T and B Cells Show Impaired Integrin-Mediated Cell Adhesion. Molecular and Cellular Biology, 2006, 26, 643-653.	2.3	103
11	Genomic landscape and clonal evolution of acute myeloid leukemia with t(8;21): an international study on 331 patients. Blood, 2019, 133, 1140-1151.	1.4	96
12	Walking the interactome for candidate prioritization in exome sequencing studies of Mendelian diseases. Bioinformatics, 2014, 30, 3215-3222.	4.1	91
13	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. American Journal of Human Genetics, 2015, 97, 483-492.	6.2	70
14	Stable expansion of highâ€grade serous ovarian cancer organoids requires a lowâ€Wnt environment. EMBO Journal, 2020, 39, e104013.	7.8	70
15	Multi-Parameter Analysis of Biobanked Human Bone Marrow Stromal Cells Shows Little Influence for Donor Age and Mild Comorbidities on Phenotypic and Functional Properties. Frontiers in Immunology, 2019, 10, 2474.	4.8	64
16	Methylation and deamination of CpGs generate p53-binding sites on a genomic scale. Trends in Genetics, 2009, 25, 63-66.	6.7	61
17	Increased risk of severe clinical course of COVID-19 in carriers of HLA-C*04:01. EClinicalMedicine, 2021, 40, 101099.	7.1	52
18	A novel conserved family of nitric oxide synthase?. Trends in Biochemical Sciences, 2004, 29, 224-226.	7.5	41

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19	Detection of TP53 Mutations in Tissue or Liquid Rebiopsies at Progression Identifies ALK+ Lung Cancer Patients with Poor Survival. Cancers, 2019, 11, 124.	3.7	36
20	Clinical characterization and mutation spectrum of German patients with familial hypercholesterolemia. Atherosclerosis, 2016, 253, 88-93.	0.8	35
21	Longitudinal therapy monitoring of ALK-positive lung cancer by combined copy number and targeted mutation profiling of cell-free DNA. EBioMedicine, 2020, 62, 103103.	6.1	32
22	Whole exome sequencing identifiesFGF16nonsense mutations as the cause of X-linked recessive metacarpal 4/5 fusion. Journal of Medical Genetics, 2013, 50, 579-584.	3.2	31
23	Recessive Inheritance of Population-Specific Intronic LINE-1 Insertion Causes a Rotor Syndrome Phenotype. Human Mutation, 2015, 36, 327-332.	2.5	28
24	NO-Bound Myoglobin:â€‰ Structural Diversity and Dynamics of the NO Ligand. Journal of the American Chemical Society, 2004, 126, 1930-1931.	13.7	26
25	BRAT1 mutations are associated with infantile epileptic encephalopathy, mitochondrial dysfunction, and survival into childhood. American Journal of Medical Genetics, Part A, 2016, 170, 2274-2281.	1.2	25
26	In search of the prototype of nitric oxide synthase. FEBS Letters, 2003, 554, 1-5.	2.8	24
27	Somatic neurofibromatosis type 1 (NF1) inactivation events in cutaneous neurofibromas of a single NF1 patient. European Journal of Human Genetics, 2015, 23, 870-873.	2.8	20
28	Exome sequencing reveals two novel compound heterozygous XYLT1 mutations in a Polish patient with Desbuquois dysplasia type 2 and growth hormone deficiency. Journal of Human Genetics, 2016, 61, 577-583.	2.3	18
29	Mutations in proteasome-related genes are associated with thyroid hemiagenesis. Endocrine, 2017, 56, 279-285.	2.3	18
30	Ocular findings in Loeys-Dietz syndrome. British Journal of Ophthalmology, 2018, 102, 1036-1040.	3.9	18
31	Alternate-locus aware variant calling in whole genome sequencing. Genome Medicine, 2016, 8, 130.	8.2	16
32	The phenotypeâ€driven computational analysis yields clinical diagnosis for patients with atypical manifestations of known intellectual disability syndromes. Molecular Genetics & Genomic Medicine, 2020, 8, e1263.	1.2	15
33	Phenotype of two Polish patients with Schaafâ€“Yang syndrome confirmed by identifying mutation in MAGEL2 gene. Clinical Dysmorphology, 2018, 27, 49-52.	0.3	13
34	Serial liquid biopsies for detection of treatment failure and profiling of resistance mechanisms in KLC1â€“ALK-rearranged lung cancer. Journal of Physical Education and Sports Management, 2019, 5, a004630.	1.2	13
35	SEMA3A and IGSF10 Are Novel Contributors to Combined Pituitary Hormone Deficiency (CPHD). Frontiers in Endocrinology, 2020, 11, 368.	3.5	13
36	The germline variants in DNA repair genes in pediatric medulloblastoma: a challenge for current therapeutic strategies. BMC Cancer, 2017, 17, 239.	2.6	12

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37	Kinetics of CO and NO Ligation with the Cys331 â†' Ala Mutant of Neuronal Nitric-oxide Synthase. Journal of Biological Chemistry, 2001, 276, 4733-4736.	3.4	10
38	The Spt-Ada-Gcn5-acetyltransferase complex interaction motif of E2a is essential for a subset of transcriptional and oncogenic properties of E2a-Pbx1. Leukemia and Lymphoma, 2009, 50, 816-828.	1.3	9
39	FGFR2 mutation in a patient without typical features of Pfeiffer syndrome â€“ The emerging role of combined NGS and phenotype based strategies. European Journal of Medical Genetics, 2015, 58, 376-380.	1.3	9
40	Systems-Level Evidence of Transcriptional Co-Regulation of Yeast Protein Complexes. Journal of Computational Biology, 2009, 16, 331-339.	1.6	7
41	Clonal Hematopoiesis: Cell of Origin, Lineage Repartition and Dynamic Evolution during Chemotherapy. Blood, 2017, 130, 632-632.	1.4	7
42	ECEPE proteins: a novel family of eukaryotic cysteine proteinases. Trends in Biochemical Sciences, 2004, 29, 524-526.	7.5	3
43	P53 Binding Sites in Transposons. Frontiers in Genetics, 2012, 3, 40.	2.3	3
44	Identification of a molecular defect in a stillborn fetus with perinatal lethal hypophosphatasia using a disease-associated genome sequencing approach. Polish Journal of Pathology, 2016, 1, 78-83.	0.3	3
45	CLN8 Mutations Presenting with a Phenotypic Continuum of Neuronal Ceroid Lipofuscinosisâ€"Literature Review and Case Report. Genes, 2021, 12, 956.	2.4	3
46	Pretreatment with methanolic extract of Pistacia lentiscus L. increases sensitivity to DNA damaging drugs in primary high-grade serous ovarian cancer cells. European Journal of Integrative Medicine, 2020, 37, 101163.	1.7	1
47	Corrigendum to â€œMammalian mitochondrial nitric oxide synthase: Characterization of a novel candidateâ€•[FEBS Lett. 580 (2006) 455-462]. FEBS Letters, 2007, 581, 2072-2073.	2.8	0