

# Peter M Krawitz

## List of Publications by Year in Descending Order

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**Version:** 2024-04-28

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

94  
papers

5,394  
citations

32  
h-index

73  
g-index

115  
ext. papers

6,739  
ext. citations

9.3  
avg. IF

4.99  
L-index

#	Paper	IF	Citations
94	Cross-tissue transcriptome-wide association studies identify susceptibility genes shared between schizophrenia and inflammatory bowel disease.. <i>Communications Biology</i> , <b>2022</b> , 5, 80	6.7	1
93	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors.. <i>Nature Genetics</i> , <b>2022</b> ,	36.3	5
92	variants of cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway.. <i>Human Genetics and Genomics Advances</i> , <b>2022</b> , 3, 100111	0.8	0
91	Reconstruction of the origin of the first major SARS-CoV-2 outbreak in Germany.. <i>Computational and Structural Biotechnology Journal</i> , <b>2022</b> , 20, 2292-2296	6.8	
90	Genetik in der pädiatrischen Rheumatologie. <i>Springer Reference Medizin</i> , <b>2022</b> , 41-53	0	
89	Breast and prostate cancer risk: The interplay of polygenic risk, rare pathogenic germline variants, and family history.. <i>Genetics in Medicine</i> , <b>2021</b> ,	8.1	4
88	Extending the allelic spectrum at noncoding risk loci of orofacial clefting. <i>Human Mutation</i> , <b>2021</b> , 42, 1066-1078	4.7	1
87	Genome sequencing in families with congenital limb malformations. <i>Human Genetics</i> , <b>2021</b> , 140, 1229-1239	2.9	0
86	CADA: phenotype-driven gene prioritization based on a case-enriched knowledge graph. <i>NAR Genomics and Bioinformatics</i> , <b>2021</b> , 3, lqab078	3.7	2
85	TBK1 and TNFRSF13B mutations and an autoinflammatory disease in a child with lethal COVID-19. <i>Npj Genomic Medicine</i> , <b>2021</b> , 6, 55	6.2	8
84	Genetik in der pädiatrischen Rheumatologie. <i>Springer Reference Medizin</i> , <b>2021</b> , 1-13	0	
83	Multimodal Machine Learning Workflows for Prediction of Psychosis in Patients With Clinical High-Risk Syndromes and Recent-Onset Depression. <i>JAMA Psychiatry</i> , <b>2021</b> , 78, 195-209	14.5	31
82	Tumor rejection in mice depends on IL-9 and Th9 cells <b>2021</b> , 9,		1
81	Knowledge transfer to enhance the performance of deep learning models for automated classification of B cell neoplasms. <i>Patterns</i> , <b>2021</b> , 2, 100351	5.1	1
80	A CRISPR-Cas9-engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2021</b> , 118,	11.5	4
79	Lessons learned from 40 novel PIGA patients and a review of the literature. <i>Epilepsia</i> , <b>2020</b> , 61, 1142-1155	5.4	15
78	Hematologist-Level Classification of Mature B-Cell Neoplasm Using Deep Learning on Multiparameter Flow Cytometry Data. <i>Cytometry Part A: the Journal of the International Society for Analytical Cytology</i> , <b>2020</b> , 97, 1073-1080	4.6	9

77	Significantly different clinical phenotypes associated with mutations in synthesis and transamidase+remodeling glycosylphosphatidylinositol (GPI)-anchor biosynthesis genes. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 40	4.2	12
76	A post glycosylphosphatidylinositol (GPI) attachment to proteins, type 2 (PGAP2) variant identified in Mabry syndrome index cases: Molecular genetics of the prototypical inherited GPI disorder. <i>European Journal of Medical Genetics</i> , <b>2020</b> , 63, 103822	2.6	5
75	Evidence of the milder phenotypic spectrum of c.1582G>A PIGT variant: Delineation based on seven novel Polish patients. <i>Clinical Genetics</i> , <b>2020</b> , 98, 468-476	4	3
74	Sex-Specific Genetic Associations for Barrett's Esophagus and Esophageal Adenocarcinoma. <i>Gastroenterology</i> , <b>2020</b> , 159, 2065-2076.e1	13.3	4
73	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , <b>2019</b> , 10, 611	4.5	7
72	PEDIA: prioritization of exome data by image analysis. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 2807-2814	8.1	20
71	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 749-757	11	15
70	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 2216-2223	8.1	18
69	Klinisch-bioinformatische Analyse bei Intelligenzminderung. <i>Medizinische Genetik</i> , <b>2019</b> , 31, 20-23	0.5	1
68	Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 395-402	11	27
67	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 384-394	11	29
66	Complement and inflammasome overactivation mediates paroxysmal nocturnal hemoglobinuria with autoinflammation. <i>Journal of Clinical Investigation</i> , <b>2019</b> , 129, 5123-5136	15.9	16
65	Identifying facial phenotypes of genetic disorders using deep learning. <i>Nature Medicine</i> , <b>2019</b> , 25, 60-64	50.5	229
64	Advances in computer-assisted syndrome recognition by the example of inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , <b>2018</b> , 41, 533-539	5.4	26
63	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. <i>Genome Medicine</i> , <b>2018</b> , 10, 3	14.4	53
62	Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. <i>Human Mutation</i> , <b>2017</b> , 38, 1394-1401	4.7	17
61	Chromatin-remodeling factor SMARCD2 regulates transcriptional networks controlling differentiation of neutrophil granulocytes. <i>Nature Genetics</i> , <b>2017</b> , 49, 742-752	36.3	58
60	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 833-843	11	37

59	Mutations in cause a recessive form of central hypoventilation with autonomic dysfunction. <i>Journal of Medical Genetics</i> , <b>2017</b> , 54, 754-761	5.8	14
58	A likelihood ratio-based method to predict exact pedigrees for complex families from next-generation sequencing data. <i>Bioinformatics</i> , <b>2017</b> , 33, 72-78	7.2	6
57	Challenges ahead for matchmaking. <i>IT - Information Technology</i> , <b>2016</b> , 58, 140-144	0.4	
56	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 310-21	11	65
55	Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. <i>Human Mutation</i> , <b>2016</b> , 37, 737-44	4.7	32
54	Juvenile arthritis caused by a novel FAMIN (LACC1) mutation in two children with systemic and extended oligoarticular course. <i>Pediatric Rheumatology</i> , <b>2016</b> , 14, 63	3.5	26
53	Missense variant in CCDC22 causes X-linked recessive intellectual disability with features of Ritscher-Schinzel/3C syndrome. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 633-8	5.3	21
52	Key features and clinical variability of COG6-CDG. <i>Molecular Genetics and Metabolism</i> , <b>2015</b> , 116, 163-70	3.7	39
51	Somatic neurofibromatosis type 1 (NF1) inactivation events in cutaneous neurofibromas of a single NF1 patient. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 870-3	5.3	16
50	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. <i>Nature Genetics</i> , <b>2015</b> , 47, 647-53	36.3	118
49	Disruptions of topological chromatin domains cause pathogenic rewiring of gene-enhancer interactions. <i>Cell</i> , <b>2015</b> , 161, 1012-1025	56.2	1207
48	Molecular mechanism of CHRDL1-mediated X-linked megalocornea in humans and in <i>Xenopus</i> model. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 3119-32	5.6	17
47	Strategies to improve the performance of rare variant association studies by optimizing the selection of controls. <i>Bioinformatics</i> , <b>2015</b> , 31, 3577-83	7.2	
46	The genomic birthday paradox: how much is enough?. <i>Human Mutation</i> , <b>2015</b> , 36, 989-97	4.7	11
45	Crowdsourced direct-to-consumer genomic analysis of a family quartet. <i>BMC Genomics</i> , <b>2015</b> , 16, 910	4.5	13
44	Jannovar: a java library for exome annotation. <i>Human Mutation</i> , <b>2014</b> , 35, 548-55	4.7	44
43	Mutations in PGAP3 impair GPI-anchor maturation, causing a subtype of hyperphosphatasia with mental retardation. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 278-87	11	80
42	First description of a patient with Vici syndrome due to a mutation affecting the penultimate exon of EPG5 and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , <b>2014</b> , 164A, 3170-5	2.5	28

41	Improved exome prioritization of disease genes through cross-species phenotype comparison. <i>Genome Research</i> , <b>2014</b> , 24, 340-8	9.7	219
40	Screening for single nucleotide variants, small indels and exon deletions with a next-generation sequencing based gene panel approach for Usher syndrome. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2014</b> , 2, 393-401	2.3	21
39	Sensory neuropathy with bone destruction due to a mutation in the membrane-shaping atlastin GTPase 3. <i>Brain</i> , <b>2014</b> , 137, 683-92	11.2	56
38	Delineation of PIGV mutation spectrum and associated phenotypes in hyperphosphatasia with mental retardation syndrome. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22, 762-7	5.3	30
37	Homozygous and compound-heterozygous mutations in TGDS cause Catel-Manzke syndrome. <i>American Journal of Human Genetics</i> , <b>2014</b> , 95, 763-70	11	30
36	Use of animal models for exome prioritization of rare disease genes. <i>Orphanet Journal of Rare Diseases</i> , <b>2014</b> , 9, O19	4.2	78
35	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. <i>Science Translational Medicine</i> , <b>2014</b> , 6, 252ra123	17.5	165
34	Efficient CRISPR/Cas9 genome editing with low off-target effects in zebrafish. <i>Development (Cambridge)</i> , <b>2013</b> , 140, 4982-7	6.6	334
33	Estimating exome genotyping accuracy by comparing to data from large scale sequencing projects. <i>Genome Medicine</i> , <b>2013</b> , 5, 69	14.4	20
32	A case of paroxysmal nocturnal hemoglobinuria caused by a germline mutation and a somatic mutation in PIGT. <i>Blood</i> , <b>2013</b> , 122, 1312-5	2.2	68
31	PGAP2 mutations, affecting the GPI-anchor-synthesis pathway, cause hyperphosphatasia with mental retardation syndrome. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 584-9	11	86
30	Loss-of-function mutations in the IL-21 receptor gene cause a primary immunodeficiency syndrome. <i>Journal of Experimental Medicine</i> , <b>2013</b> , 210, 433-43	16.6	156
29	Mutations in WNT1 cause different forms of bone fragility. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 565-74	11	197
28	Doubly heterozygous LMNA and TTN mutations revealed by exome sequencing in a severe form of dilated cardiomyopathy. <i>European Journal of Human Genetics</i> , <b>2013</b> , 21, 1105-11	5.3	69
27	Whole exome sequencing identified a novel zinc-finger gene ZNF141 associated with autosomal recessive postaxial polydactyly type A. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 47-53	5.8	30
26	Whole exome sequencing identifies FGF16 nonsense mutations as the cause of X-linked recessive metacarpal 4/5 fusion. <i>Journal of Medical Genetics</i> , <b>2013</b> , 50, 579-84	5.8	26
25	Distinct global shifts in genomic binding profiles of limb malformation-associated HOXD13 mutations. <i>Genome Research</i> , <b>2013</b> , 23, 2091-102	9.7	24
24	Filtering for compound heterozygous sequence variants in non-consanguineous pedigrees. <i>PLoS ONE</i> , <b>2013</b> , 8, e70151	3.7	30

23	Phenotypic variability in hyperphosphatasia with seizures and neurologic deficit (Mabry syndrome). <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 553-8	2.5	32
22	Mutations in PIGO, a member of the GPI-anchor-synthesis pathway, cause hyperphosphatasia with mental retardation. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 146-51	11	113
21	Homeotic arm-to-leg transformation associated with genomic rearrangements at the PITX1 locus. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 629-35	11	95
20	GeneTalk: an expert exchange platform for assessing rare sequence variants in personal genomes. <i>Bioinformatics</i> , <b>2012</b> , 28, 2515-6	7.2	42
19	The allele distribution in next-generation sequencing data sets is accurately described as the result of a stochastic branching process. <i>Nucleic Acids Research</i> , <b>2012</b> , 40, 2426-31	20.1	33
18	Mechanism for release of alkaline phosphatase caused by glycosylphosphatidylinositol deficiency in patients with hyperphosphatasia mental retardation syndrome. <i>Journal of Biological Chemistry</i> , <b>2012</b> , 287, 6318-25	5.4	73
17	Hyperphosphatasia-mental retardation syndrome due to PIGV mutations: expanded clinical spectrum. <i>American Journal of Medical Genetics, Part A</i> , <b>2011</b> , 155A, 1917-22	2.5	41
16	Misregulation of mitotic chromosome segregation in a new type of autosomal recessive primary microcephaly. <i>Cell Cycle</i> , <b>2011</b> , 10, 2967-77	4.7	11
15	Identity-by-descent filtering of exome sequence data for disease-gene identification in autosomal recessive disorders. <i>Bioinformatics</i> , <b>2011</b> , 27, 829-36	7.2	27
14	Identity-by-descent filtering of exome sequence data identifies PIGV mutations in hyperphosphatasia mental retardation syndrome. <i>Nature Genetics</i> , <b>2010</b> , 42, 827-9	36.3	250
13	Microindel detection in short-read sequence data. <i>Bioinformatics</i> , <b>2010</b> , 26, 722-9	7.2	82
12	VZV meningitis following varicella vaccine. <i>Journal of Clinical Virology</i> , <b>2010</b> , 48, 275-7	14.5	10
11	Clinical diagnostics in human genetics with semantic similarity searches in ontologies. <i>American Journal of Human Genetics</i> , <b>2009</b> , 85, 457-64	11	328
10	Basin entropy in Boolean network ensembles. <i>Physical Review Letters</i> , <b>2007</b> , 98, 158701	7.4	70
9	Entropy of complex relevant components of Boolean networks. <i>Physical Review E</i> , <b>2007</b> , 76, 036115	2.4	26
8	Differential localization and identification of a critical aspartate suggest non-redundant proteolytic functions of the presenilin homologues SPPL2b and SPPL3. <i>Journal of Biological Chemistry</i> , <b>2005</b> , 280, 39515-23	5.4	72
7	Dependence of age-specific incidence of acute myeloid leukemia on karyotype. <i>Blood</i> , <b>2001</b> , 98, 3500	2.2	41
6	De novo variants in the PSMC3 proteasome AAA-ATPase subunit gene cause neurodevelopmental disorders associated with type I interferonopathies		1

5	A CRISPR-Cas9 engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions	1
4	Advances in computer-assisted syndrome recognition and differentiation in a set of metabolic disorders	2
3	PEDIA: Prioritization of Exome Data by Image Analysis	1
2	Complement- and inflammasome-mediated autoinflammation-paroxysmal nocturnal hemoglobinuria	1
1	CADA: Phenotype-driven gene prioritization based on a case-enriched knowledge graph	1