Peter M Krawitz

List of Publications by Citations

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	Disruptions of topological chromatin domains cause pathogenic rewiring of gene-enhancer interactions. *Cell*, **2015**, 161, 1012-1025	56.2	1207
93	Efficient CRISPR/Cas9 genome editing with low off-target effects in zebrafish. *Development (Cambridge)*, **2013**, 140, 4982-7	6.6	334
92	Clinical diagnostics in human genetics with semantic similarity searches in ontologies. *American Journal of Human Genetics*, **2009**, 85, 457-64	11	328
91	Identity-by-descent filtering of exome sequence data identifies PIGV mutations in hyperphosphatasia mental retardation syndrome. *Nature Genetics*, **2010**, 42, 827-9	36.3	250
90	Identifying facial phenotypes of genetic disorders using deep learning. *Nature Medicine*, **2019**, 25, 60-64	50.5	229
89	Improved exome prioritization of disease genes through cross-species phenotype comparison. *Genome Research*, **2014**, 24, 340-8	9.7	219
88	Mutations in WNT1 cause different forms of bone fragility. *American Journal of Human Genetics*, **2013**, 92, 565-74	11	197
87	Effective diagnosis of genetic disease by computational phenotype analysis of the disease-associated genome. *Science Translational Medicine*, **2014**, 6, 252ra123	17.5	165
86	Loss-of-function mutations in the IL-21 receptor gene cause a primary immunodeficiency syndrome. *Journal of Experimental Medicine*, **2013**, 210, 433-43	16.6	156
85	PDE3A mutations cause autosomal dominant hypertension with brachydactyly. *Nature Genetics*, **2015**, 47, 647-53	36.3	118
84	Mutations in PIGO, a member of the GPI-anchor-synthesis pathway, cause hyperphosphatasia with mental retardation. *American Journal of Human Genetics*, **2012**, 91, 146-51	11	113
83	Homeotic arm-to-leg transformation associated with genomic rearrangements at the PITX1 locus. *American Journal of Human Genetics*, **2012**, 91, 629-35	11	95
82	PGAP2 mutations, affecting the GPI-anchor-synthesis pathway, cause hyperphosphatasia with mental retardation syndrome. *American Journal of Human Genetics*, **2013**, 92, 584-9	11	86
81	Microindel detection in short-read sequence data. *Bioinformatics*, **2010**, 26, 722-9	7.2	82
80	Mutations in PGAP3 impair GPI-anchor maturation, causing a subtype of hyperphosphatasia with mental retardation. *American Journal of Human Genetics*, **2014**, 94, 278-87	11	80
79	Use of animal models for exome prioritization of rare disease genes. *Orphanet Journal of Rare Diseases*, **2014**, 9, O19	4.2	78
78	Mechanism for release of alkaline phosphatase caused by glycosylphosphatidylinositol deficiency in patients with hyperphosphatasia mental retardation syndrome. *Journal of Biological Chemistry*, **2012**, 287, 6318-25	5.4	73

77	Differential localization and identification of a critical aspartate suggest non-redundant proteolytic functions of the presenilin homologues SPPL2b and SPPL3. *Journal of Biological Chemistry*, **2005**, 280, 39515-23	5.4	72
76	Basin entropy in Boolean network ensembles. *Physical Review Letters*, **2007**, 98, 158701	7.4	70
75	Doubly heterozygous LMNA and TTN mutations revealed by exome sequencing in a severe form of dilated cardiomyopathy. *European Journal of Human Genetics*, **2013**, 21, 1105-11	5.3	69
74	A case of paroxysmal nocturnal hemoglobinuria caused by a germline mutation and a somatic mutation in PIGT. *Blood*, **2013**, 122, 1312-5	2.2	68
73	CCDC115 Deficiency Causes a Disorder of Golgi Homeostasis with Abnormal Protein Glycosylation. *American Journal of Human Genetics*, **2016**, 98, 310-21	11	65
72	Chromatin-remodeling factor SMARCD2 regulates transcriptional networks controlling differentiation of neutrophil granulocytes. *Nature Genetics*, **2017**, 49, 742-752	36.3	58
71	Sensory neuropathy with bone destruction due to a mutation in the membrane-shaping atlastin GTPase 3. *Brain*, **2014**, 137, 683-92	11.2	56
70	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. *Genome Medicine*, **2018**, 10, 3	14.4	53
69	Jannovar: a java library for exome annotation. *Human Mutation*, **2014**, 35, 548-55	4.7	44
68	GeneTalk: an expert exchange platform for assessing rare sequence variants in personal genomes. *Bioinformatics*, **2012**, 28, 2515-6	7.2	42
67	Hyperphosphatasia-mental retardation syndrome due to PIGV mutations: expanded clinical spectrum. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1917-22	2.5	41
66	Dependence of age-specific incidence of acute myeloid leukemia on karyotype. *Blood*, **2001**, 98, 3500	2.2	41
65	Key features and clinical variability of COG6-CDG. *Molecular Genetics and Metabolism*, **2015**, 116, 163-70	3.7	39
64	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. *American Journal of Human Genetics*, **2017**, 101, 833-843	11	37
63	The allele distribution in next-generation sequencing data sets is accurately described as the result of a stochastic branching process. *Nucleic Acids Research*, **2012**, 40, 2426-31	20.1	33
62	Phenotypic variability in hyperphosphatasia with seizures and neurologic deficit (Mabry syndrome). *American Journal of Medical Genetics, Part A*, **2012**, 158A, 553-8	2.5	32
61	Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. *Human Mutation*, **2016**, 37, 737-44	4.7	32
60	Multimodal Machine Learning Workflows for Prediction of Psychosis in Patients With Clinical High-Risk Syndromes and Recent-Onset Depression. *JAMA Psychiatry*, **2021**, 78, 195-209	14.5	31

59	Delineation of PIGV mutation spectrum and associated phenotypes in hyperphosphatasia with mental retardation syndrome. *European Journal of Human Genetics*, **2014**, 22, 762-7	5.3	30
58	Homozygous and compound-heterozygous mutations in TGDS cause Catel-Manzke syndrome. *American Journal of Human Genetics*, **2014**, 95, 763-70	11	30
57	Whole exome sequencing identified a novel zinc-finger gene ZNF141 associated with autosomal recessive postaxial polydactyly type A. *Journal of Medical Genetics*, **2013**, 50, 47-53	5.8	30
56	Filtering for compound heterozygous sequence variants in non-consanguineous pedigrees. *PLoS ONE*, **2013**, 8, e70151	3.7	30
55	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. *American Journal of Human Genetics*, **2019**, 105, 384-394	11	29
54	First description of a patient with Vici syndrome due to a mutation affecting the penultimate exon of EPG5 and review of the literature. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 3170-5	2.5	28
53	Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. *American Journal of Human Genetics*, **2019**, 105, 395-402	11	27
52	Identity-by-descent filtering of exome sequence data for disease-gene identification in autosomal recessive disorders. *Bioinformatics*, **2011**, 27, 829-36	7.2	27
51	Advances in computer-assisted syndrome recognition by the example of inborn errors of metabolism. *Journal of Inherited Metabolic Disease*, **2018**, 41, 533-539	5.4	26
50	Whole exome sequencing identifies FGF16 nonsense mutations as the cause of X-linked recessive metacarpal 4/5 fusion. *Journal of Medical Genetics*, **2013**, 50, 579-84	5.8	26
49	Entropy of complex relevant components of Boolean networks. *Physical Review E*, **2007**, 76, 036115	2.4	26
48	Juvenile arthritis caused by a novel FAMIN (LACC1) mutation in two children with systemic and extended oligoarticular course. *Pediatric Rheumatology*, **2016**, 14, 63	3.5	26
47	Distinct global shifts in genomic binding profiles of limb malformation-associated HOXD13 mutations. *Genome Research*, **2013**, 23, 2091-102	9.7	24
46	Missense variant in CCDC22 causes X-linked recessive intellectual disability with features of Ritscher-Schinzel/3C syndrome. *European Journal of Human Genetics*, **2015**, 23, 633-8	5.3	21
45	Screening for single nucleotide variants, small indels and exon deletions with a next-generation sequencing based gene panel approach for Usher syndrome. *Molecular Genetics & Genomic Medicine*, **2014**, 2, 393-401	2.3	21
44	PEDIA: prioritization of exome data by image analysis. *Genetics in Medicine*, **2019**, 21, 2807-2814	8.1	20
43	Estimating exome genotyping accuracy by comparing to data from large scale sequencing projects. *Genome Medicine*, **2013**, 5, 69	14.4	20
42	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. *Genetics in Medicine*, **2019**, 21, 2216-2223	8.1	18

41	Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. *Human Mutation*, **2017**, 38, 1394-1401	4.7	17
40	Molecular mechanism of CHRDL1-mediated X-linked megalocornea in humans and in Xenopus model. *Human Molecular Genetics*, **2015**, 24, 3119-32	5.6	17
39	Somatic neurofibromatosis type 1 (NF1) inactivation events in cutaneous neurofibromas of a single NF1 patient. *European Journal of Human Genetics*, **2015**, 23, 870-3	5.3	16
38	Complement and inflammasome overactivation mediates paroxysmal nocturnal hemoglobinuria with autoinflammation. *Journal of Clinical Investigation*, **2019**, 129, 5123-5136	15.9	16
37	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. *American Journal of Human Genetics*, **2019**, 104, 749-757	11	15
36	Lessons learned from 40 novel PIGA patients and a review of the literature. *Epilepsia*, **2020**, 61, 1142-1156	5.4	15
35	Mutations in cause a recessive form of central hypoventilation with autonomic dysfunction. *Journal of Medical Genetics*, **2017**, 54, 754-761	5.8	14
34	Crowdsourced direct-to-consumer genomic analysis of a family quartet. *BMC Genomics*, **2015**, 16, 910	4.5	13
33	Significantly different clinical phenotypes associated with mutations in synthesis and transamidase+remodeling glycosylphosphatidylinositol (GPI)-anchor biosynthesis genes. *Orphanet Journal of Rare Diseases*, **2020**, 15, 40	4.2	12
32	The genomic birthday paradox: how much is enough?. *Human Mutation*, **2015**, 36, 989-97	4.7	11
31	Misregulation of mitotic chromosome segregation in a new type of autosomal recessive primary microcephaly. *Cell Cycle*, **2011**, 10, 2967-77	4.7	11
30	VZV meningitis following varicella vaccine. *Journal of Clinical Virology*, **2010**, 48, 275-7	14.5	10
29	Hematologist-Level Classification of Mature B-Cell Neoplasm Using Deep Learning on Multiparameter Flow Cytometry Data. *Cytometry Part A: the Journal of the International Society for Analytical Cytology*, **2020**, 97, 1073-1080	4.6	9
28	TBK1 and TNFRSF13B mutations and an autoinflammatory disease in a child with lethal COVID-19. *Npj Genomic Medicine*, **2021**, 6, 55	6.2	8
27	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. *Frontiers in Genetics*, **2019**, 10, 611	4.5	7
26	A likelihood ratio-based method to predict exact pedigrees for complex families from next-generation sequencing data. *Bioinformatics*, **2017**, 33, 72-78	7.2	6
25	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors.. *Nature Genetics*, **2022**,	36.3	5
24	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. *European Journal of Medical Genetics*, **2020**, 63, 103822	2.6	5

23	Breast and prostate cancer risk: The interplay of polygenic risk, rare pathogenic germline variants, and family history.. *Genetics in Medicine*, **2021**,	8.1	4
22	Sex-Specific Genetic Associations for Barrett's Esophagus and Esophageal Adenocarcinoma. *Gastroenterology*, **2020**, 159, 2065-2076.e1	13.3	4
21	A CRISPR-Cas9-engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions. *Proceedings of the National Academy of Sciences of the United States of America*, **2021**, 118,	11.5	4
20	Evidence of the milder phenotypic spectrum of c.1582G>A PIGT variant: Delineation based on seven novel Polish patients. *Clinical Genetics*, **2020**, 98, 468-476	4	3
19	Advances in computer-assisted syndrome recognition and differentiation in a set of metabolic disorders		2
18	CADA: phenotype-driven gene prioritization based on a case-enriched knowledge graph. *NAR Genomics and Bioinformatics*, **2021**, 3, lqab078	3.7	2
17	Klinisch-bioinformatische Analyse bei Intelligenzminderung. *Medizinische Genetik*, **2019**, 31, 20-23	0.5	1
16	Cross-tissue transcriptome-wide association studies identify susceptibility genes shared between schizophrenia and inflammatory bowel disease.. *Communications Biology*, **2022**, 5, 80	6.7	1
15	De novo variants in the PSMC3 proteasome AAA-ATPase subunit gene cause neurodevelopmental disorders associated with type I interferonopathies		1
14	A CRISPR-Cas9-engineered mouse model for GPI-anchor deficiency mirrors human phenotypes and exhibits hippocampal synaptic dysfunctions		1
13	PEDIA: Prioritization of Exome Data by Image Analysis		1
12	Complement- and inflammasome-mediated autoinflammation-paroxysmal nocturnal hemoglobinuria		1
11	CADA: Phenotype-driven gene prioritization based on a case-enriched knowledge graph		1
10	Extending the allelic spectrum at noncoding risk loci of orofacial clefting. *Human Mutation*, **2021**, 42, 1066-1078	4.7	1
9	Tumor rejection in mice depends on IL-9 and Th9 cells **2021**, 9,		1
8	Knowledge transfer to enhance the performance of deep learning models for automated classification of B cell neoplasms. *Patterns*, **2021**, 2, 100351	5.1	1
7	Genome sequencing in families with congenital limb malformations. *Human Genetics*, **2021**, 140, 1229-1239	3.9	0
6	variants of cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway.. *Human Genetics and Genomics Advances*, **2022**, 3, 100111	0.8	0

List of Publications

5	Strategies to improve the performance of rare variant association studies by optimizing the selection of controls. *Bioinformatics*, **2015**, 31, 3577-83	7.2
4	Challenges ahead for matchmaking. *IT - Information Technology*, **2016**, 58, 140-144	0.4
3	Genetik in der pädiatrischen Rheumatologie. *Springer Reference Medizin*, **2021**, 1-13	0
2	Reconstruction of the origin of the first major SARS-CoV-2 outbreak in Germany.. *Computational and Structural Biotechnology Journal*, **2022**, 20, 2292-2296	6.8
1	Genetik in der pädiatrischen Rheumatologie. *Springer Reference Medizin*, **2022**, 41-53	0