Dominik Seelow

List of Publications by Year
in descending order

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48 papers	10,328 citations	270111 25 h-index	252626 46 g-index

53 all docs	53 docs citations	53 times ranked	24310 citing authors

#	Article	IF	Citations
1	Biallelic truncating variants in ATP9A cause a novel neurodevelopmental disorder involving postnatal microcephaly and failure to thrive. Journal of Medical Genetics, 2022, 59, 662-668.	1.5	9
2	AutozygosityMapper: Identification of disease-mutations in consanguineous families. Nucleic Acids Research, 2022, 50, W83-W89.	6.5	2
3	FABIAN-variant: predicting the effects of DNA variants on transcription factor binding. Nucleic Acids Research, 2022, 50, W322-W329.	6.5	12
4	Deep phenotyping: symptom annotation made simple with SAMS. Nucleic Acids Research, 2022, 50, W677-W681.	6.5	5
5	MutationTaster2021. Nucleic Acids Research, 2021, 49, W446-W451.	6.5	122
6	Aviator: a web service for monitoring the availability of web services. Nucleic Acids Research, 2021, 49, W46-W51.	6.5	3
7	SIGLEC1 (CD169): a marker of active neuroinflammation in the brain but not in the blood of multiple sclerosis patients. Scientific Reports, 2021, 11, 10299.	1.6	14
8	Public data sources for regulatory genomic features. Medizinische Genetik, 2021, 33, 167-177.	0.1	1
9	Novel sequencing technologies and bioinformatic tools for deciphering the non-coding genome. Medizinische Genetik, 2021, 33, 133-145.	0.1	1
10	An intronic splice site alteration in combination with a large deletion affecting VPS13B (COH1) causes Cohen syndrome. European Journal of Medical Genetics, 2020, 63, 103973.	0.7	5
11	VarFish: comprehensive DNA variant analysis for diagnostics and research. Nucleic Acids Research, 2020, 48, W162-W169.	6.5	39
12	Pervasive and CpG-dependent promoter-like characteristics of transcribed enhancers. Nucleic Acids Research, 2020, 48, 5306-5317.	6.5	24
13	RegulationSpotter: annotation and interpretation of extratranscriptic DNA variants. Nucleic Acids Research, 2019, 47, W106-W113.	6.5	17
14	MutationDistiller: user-driven identification of pathogenic DNA variants. Nucleic Acids Research, 2019, 47, W114-W120.	6.5	37
15	Phenotero: Annotate as you write. Clinical Genetics, 2019, 95, 287-292.	1.0	3
16	De novo mutation inELOVL1causes ichthyosis,acanthosis nigricans, hypomyelination, spastic paraplegia, high frequency deafness and optic atrophy. Journal of Medical Genetics, 2019, 56, 164-175.	1.5	54
17	Harmonising phenomics information for a better interoperability in the rare disease field. European Journal of Medical Genetics, 2018, 61, 706-714.	0.7	29
18	A systematic, large-scale comparison of transcription factor binding site models. BMC Genomics, 2016, 17, 388.	1.2	15

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19	Recessive REEP1 mutation is associated with congenital axonal neuropathy and diaphragmatic palsy. Neurology: Genetics, 2015, 1, e32.	0.9	21
20	Clinical application of whole exome sequencing reveals a novel compound heterozygous TK2-mutation in two brothers with rapidly progressive combined muscle-brain atrophy, axonal neuropathy, and status epilepticus. Mitochondrion, 2015, 20, 1-6.	1.6	18
21	Improved exome prioritization of disease genes through cross-species phenotype comparison. Genome Research, 2014, 24, 340-348.	2.4	300
22	MutationTaster2: mutation prediction for the deep-sequencing age. Nature Methods, 2014, 11, 361-362.	9.0	3,203
23	GrabBlur - a framework to facilitate the secure exchange of whole-exome and -genome SNV data using VCF files. BMC Genomics, 2014, 15, S8.	1.2	6
24	ZC4H2 Mutations Are Associated with Arthrogryposis Multiplex Congenita and Intellectual Disability through Impairment of Central and Peripheral Synaptic Plasticity. American Journal of Human Genetics, 2013, 92, 681-695.	2.6	68
25	Identification of a Ninein (NIN) mutation in a family with spondyloepimetaphyseal dysplasia with joint laxity (leptodactylic type)-like phenotype. Matrix Biology, 2013, 32, 387-392.	1.5	15
26	CNVinspector:a web-based tool for the interactive evaluation of copy number variations in single patients and in cohorts. Journal of Medical Genetics, 2013, 50, 529-533.	1.5	3
27	HomozygosityMapper2012–bridging the gap between homozygosity mapping and deep sequencing. Nucleic Acids Research, 2012, 40, W516-W520.	6.5	69
28	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	2.6	108
29	Systematic Comparison of Three Methods for Fragmentation of Long-Range PCR Products for Next Generation Sequencing. PLoS ONE, 2011, 6, e28240.	1.1	106
30	MutationTaster evaluates disease-causing potential of sequence alterations. Nature Methods, 2010, 7, 575-576.	9.0	2,538
31	Fatal Cardiac Arrhythmia and Long-QT Syndrome in a New Form of Congenital Generalized Lipodystrophy with Muscle Rippling (CGL4) Due to PTRF-CAVIN Mutations. PLoS Genetics, 2010, 6, e1000874.	1.5	198
32	Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the ADAM9 gene. Molecular and Cellular Probes, 2010, 24, 357-363.	0.9	20
33	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	3.9	102
34	A Systematic Approach to Mapping Recessive Disease Genes in Individuals from Outbred Populations. PLoS Genetics, 2009, 5, e1000353.	1.5	144
35	HomozygosityMapper--an interactive approach to homozygosity mapping. Nucleic Acids Research, 2009, 37, W593-W599.	6.5	331
36	FragIdent â€“ Automatic identification and characterisation of cDNA-fragments. BMC Genomics, 2009, 10, 95.	1.2	0

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37	Acetylcholine Receptor Pathway Mutations Explain Various Fetal Akinesia Deformation Sequence Disorders. American Journal of Human Genetics, 2008, 82, 464-476.	2.6	124
38	The Human Phenotype Ontology: A Tool for Annotating and Analyzing Human Hereditary Disease. American Journal of Human Genetics, 2008, 83, 610-615.	2.6	797
39	GeneDistillerâ€"Distilling Candidate Genes from Linkage Intervals. PLoS ONE, 2008, 3, e3874.	1.1	98
40	AssociationDB: web-based exploration of genomic association. Bioinformatics, 2007, 23, 2643-2644.	1.8	0
41	RAB23 Mutations in Carpenter Syndrome Imply an Unexpected Role for Hedgehog Signaling in Cranial-Suture Development and Obesity. American Journal of Human Genetics, 2007, 80, 1162-1170.	2.6	229
42	Loss of GLIS2 causes nephronophthisis in humans and mice by increased apoptosis and fibrosis. Nature Genetics, 2007, 39, 1018-1024.	9.4	221
43	Mutations in the Tight-Junction Gene Claudin 19 (CLDN19) Are Associated with Renal Magnesium Wasting, Renal Failure, and Severe Ocular Involvement. American Journal of Human Genetics, 2006, 79, 949-957.	2.6	446
44	Mutations in the Gene Encoding the Wnt-Signaling Component R-Spondin 4 (RSPO4) Cause Autosomal Recessive Anonychia. American Journal of Human Genetics, 2006, 79, 1105-1109.	2.6	94
45	Positional cloning uncovers mutations in PLCE1 responsible for a nephrotic syndrome variant that may be reversible. Nature Genetics, 2006, 38, 1397-1405.	9.4	510
46	d-matrix - database exploration, visualization and analysis. BMC Bioinformatics, 2004, 5, 168.	1.2	3
47	Genome-Wide Array Analysis of Normal and Malformed Human Hearts. Circulation, 2003, 107, 2467-2474.	1.6	109
48	Periodic catatonia: confirmation of linkage to chromosome 15 and further evidence for genetic heterogeneity. Human Genetics, 2002, 111, 323-330.	1.8	53