Bernt Popp

List of Publications by Year in descending order

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50	2,015	361413	265206
papers	citations	20 h-index	42 g-index

61	61	61	4634
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes. Nature Genetics, 2010, 42, 1021-1026.	21.4	431
2	Haploinsufficiency of ARID1B, a Member of the SWI/SNF-A Chromatin-Remodeling Complex, Is a Frequent Cause of Intellectual Disability. American Journal of Human Genetics, 2012, 90, 565-572.	6.2	225
3	Clinical relevance of systematic phenotyping and exome sequencing in patients with short stature. Genetics in Medicine, 2018, 20, 630-638.	2.4	101
4	Exome Pool-Seq in neurodevelopmental disorders. European Journal of Human Genetics, 2017, 25, 1364-1376.	2.8	77
5	De novo missense mutations in the NAA10 gene cause severe non-syndromic developmental delay in males and females. European Journal of Human Genetics, 2015, 23, 602-609.	2.8	72
6	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
7	Need for high-resolution Genetic Analysis in iPSC: Results and Lessons from the ForIPS Consortium. Scientific Reports, 2018, 8, 17201.	3.3	70
8	Mutations in MBOAT7 , Encoding Lysophosphatidylinositol Acyltransferase I, Lead to Intellectual Disability Accompanied by Epilepsy and Autistic Features. American Journal of Human Genetics, 2016, 99, 912-916.	6.2	69
9	Mutations in the BAF-Complex Subunit DPF2 Are Associated with Coffin-Siris Syndrome. American Journal of Human Genetics, 2018, 102, 468-479.	6.2	63
10	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
11	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
12	The mutational and phenotypic spectrum of TUBA1A-associated tubulinopathy. Orphanet Journal of Rare Diseases, 2019, 14, 38.	2.7	48
13	Behavioral phenotype in five individuals with de novo mutations within the GRIN2B gene. Behavioral and Brain Functions, 2013, 9, 20.	3.3	47
14	TRIM28 haploinsufficiency predisposes to Wilms tumor. International Journal of Cancer, 2019, 145, 941-951.	5.1	45
15	CUSHAW3: Sensitive and Accurate Base-Space and Color-Space Short-Read Alignment with Hybrid Seeding. PLoS ONE, 2014, 9, e86869.	2.5	45
16	Single molecule real time sequencing in ADTKD-MUC1 allows complete assembly of the VNTR and exact positioning of causative mutations. Scientific Reports, 2018, 8, 4170.	3.3	40
17	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	2.4	32
18	The genetic landscape of intellectual disability and epilepsy in adults and the elderly: a systematic genetic work-up of 150 individuals. Genetics in Medicine, 2021, 23, 1492-1497.	2.4	31

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19	Matching clinical and genetic diagnoses in autosomal dominant polycystic kidney disease reveals novel phenocopies and potential candidate genes. Genetics in Medicine, 2020, 22, 1374-1383.	2.4	30
20	DYNC2LI1 mutations broaden the clinical spectrum of dynein-2 defects. Scientific Reports, 2015, 5, 11649.	3.3	28
21	Biallelic Expression of Mucin-1 in Autosomal Dominant Tubulointerstitial Kidney Disease: Implications for Nongenetic Disease Recognition. Journal of the American Society of Nephrology: JASN, 2018, 29, 2298-2309.	6.1	25
22	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24
23	Exome first approach to reduce diagnostic costs and time â€“ retrospective analysis of 111 individuals with rare neurodevelopmental disorders. European Journal of Human Genetics, 2022, 30, 117-125.	2.8	22
24	Is MED13L-related intellectual disability a recognizable syndrome?. European Journal of Medical Genetics, 2019, 62, 129-136.	1.3	21
25	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
26	Targeted sequencing of FH-deficient uterine leiomyomas reveals biallelic inactivating somatic fumarase variants and allows characterization of missense variants. Modern Pathology, 2020, 33, 2341-2353.	5.5	19
27	SWI/SNF protein expression status in fumarate hydrataseâ€“deficient renal cell carcinoma: immunohistochemical analysis of 32 tumors from 28 patients. Human Pathology, 2018, 77, 139-146.	2.0	18
28	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18
29	Prenatal diagnosis of HNF1Bâ€associated renal cysts: Is there a need to differentiate intragenic variants from 17q12 microdeletion syndrome?. Prenatal Diagnosis, 2019, 39, 1136-1147.	2.3	16
30	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
31	Genotypeâ€“phenotype correlation at codon 1740 of <scp>SETD2</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2037-2048.	1.2	14
32	Do the exome: A case of Williams-Beuren syndrome with severe epilepsy due to a truncating de novo variant in GABRA1. European Journal of Medical Genetics, 2016, 59, 549-553.	1.3	11
33	A biallelic truncating AEBP1 variant causes connective tissue disorder in two siblings. American Journal of Medical Genetics, Part A, 2019, 179, 50-56.	1.2	11
34	Evolutionary conserved networks of human height identify multiple Mendelian causes of short stature. European Journal of Human Genetics, 2019, 27, 1061-1071.	2.8	11
35	High-throughput imaging of ATG9A distribution as a diagnostic functional assay for adaptor protein complex 4-associated hereditary spastic paraplegia. Brain Communications, 2021, 3, fcab221.	3.3	11
36	De novo variants in ATP2B1 lead to neurodevelopmental delay. American Journal of Human Genetics, 2022, 109, 944-952.	6.2	11

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37	Whole exome sequencing reveals a novel de novo FOXC1 mutation in a patient with unrecognized Axenfeldâ€“Rieger syndrome and glaucoma. Gene, 2015, 568, 76-80.	2.2	10
38	Differential Coassembly of Î±1-GABA_ARs Associated with Epileptic Encephalopathy. Journal of Neuroscience, 2020, 40, 5518-5530.	3.6	10
39	Diverse molecular causes of unsolved autosomal dominant tubulointerstitial kidney diseases. Kidney International, 2022, 102, 405-420.	5.2	10
40	BDV Syndrome: an Emerging Syndrome With Profound Obesity and Neurodevelopmental Delay Resembling Prader-Willi Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 3413-3427.	3.6	9
41	Complementing the phenotypical spectrum of TUBA1A tubulinopathy and its role in early-onset epilepsies. European Journal of Human Genetics, 2022, 30, 298-306.	2.8	9
42	Breast MRI texture analysis for prediction of BRCA-associated genetic risk. BMC Medical Imaging, 2020, 20, 86.	2.7	8
43	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. Kidney International, 2022, 101, 1039-1053.	5.2	8
44	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
45	<scp>QRICH1</scp> variants in <scp>Ververiâ€Brady</scp> syndromeâ€”delineation of the genotypic and phenotypic spectrum. Clinical Genetics, 2021, 99, 199-207.	2.0	5
46	<scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	2.0	5
47	BiallelicANKS6mutations cause late-onset ciliopathy with chronic kidney disease through YAP dysregulation. Human Molecular Genetics, 2022, 31, 1357-1369.	2.9	5
48	Prenatal phenotype of PNKP-related primary microcephaly associated with variants affecting both the FHA and phosphatase domain. European Journal of Human Genetics, 2022, 30, 101-110.	2.8	3
49	The Dilemma of Regularly Missed Diagnoses: ADTKD. Archives of Clinical and Medical Case Reports, 2019, 03, .	0.1	2
50	Dissecting TSC2-mutated renal and hepatic angiomyolipomas in an individual with ARID1B-associated intellectual disability. BMC Cancer, 2019, 19, 435.	2.6	1