Guntram Borck

List of Publications by Year
in descending order

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91 papers	6,429 citations	81900 39 h-index	74163 75 g-index
92 all docs	92 docs citations	92 times ranked	14653 citing authors

#	Article	IF	Citations
1	Elucidating the relationship between migraine risk and brain structure using genetic data. Brain, 2022, 145, 3214-3224.	7.6	7
2	The murine ortholog of Kaufman oculocerebrofacial syndrome protein Ube3b regulates synapse number by ubiquitinating Ppp3cc. Molecular Psychiatry, 2021, 26, 1980-1995.	7.9	18
3	SQSTM1/p62 variants in 486 patients with familial ALS from Germany and Sweden. Neurobiology of Aging, 2020, 87, 139.e9-139.e15.	3.1	23
4	Habitual sleep disturbances and migraine: a Mendelian randomization study. Annals of Clinical and Translational Neurology, 2020, 7, 2370-2380.	3.7	18
5	A genome-wide cross-phenotype meta-analysis of the association of blood pressure with migraine. Nature Communications, 2020, 11, 3368.	12.8	49
6	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. International Journal of Epidemiology, 2020, 49, 1022-1031.	1.9	34
7	Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. Brain, 2019, 142, e67-e67.	7.6	1
8	Next-generation sequencing of 32 genes associated with hereditary aortopathies and related disorders of connective tissue in a cohort of 199 patients. Genetics in Medicine, 2019, 21, 1832-1841.	2.4	26
9	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	3.8	16
10	Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan. Molecular Genetics & Genomic Medicine, 2019, 7, e539.	1.2	9
11	Genotypes of amyotrophic lateral sclerosis in Mongolia. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1300-1302.	1.9	1
12	Functional and Phenotypic Characteristics of Human Leptin Receptor Mutations. Journal of the Endocrine Society, 2019, 3, 27-41.	0.2	47
13	Dominant Noonan syndrome-causing LZTR1 mutations specifically affect the Kelch domain substrate-recognition surface and enhance RAS-MAPK signaling. Human Molecular Genetics, 2019, 28, 1007-1022.	2.9	58
14	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708.	2.8	22
15	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. Genetics in Medicine, 2018, 20, 1175-1185.	2.4	133
16	A novel homozygous missense variant inNECTIN4 (PVRL4)causing ectodermal dysplasia cutaneous syndactyly syndrome. Annals of Human Genetics, 2018, 82, 232-238.	0.8	7
17	Early childhood BMI trajectories in monogenic obesity due to leptin, leptin receptor, and melanocortin 4 receptor deficiency. International Journal of Obesity, 2018, 42, 1602-1609.	3.4	44
18	Hot-spot KIF5A mutations cause familial ALS. Brain, 2018, 141, 688-697.	7.6	167

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19	Comprehensive analysis of the mutation spectrum in 301 German ALS families. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 817-827.	1.9	80
20	Expanding the phenotype associated with biallelic WDR60 mutations: Siblings with retinal degeneration and polydactyly lacking other features of short rib thoracic dystrophies. American Journal of Medical Genetics, Part A, 2018, 176, 438-442.	1.2	10
21	Noncoding copy-number variations are associated with congenital limb malformation. Genetics in Medicine, 2018, 20, 599-607.	2.4	42
22	Association Between Germline Mutations in BRF1, a Subunit of the RNA Polymerase III Transcription Complex, and Hereditary Colorectal Cancer. Gastroenterology, 2018, 154, 181-194.e20.	1.3	32
23	Kaufman oculocerebrofacial syndrome: Novel UBE3B mutations and clinical features in four unrelated patients. American Journal of Medical Genetics, Part A, 2018, 176, 187-193.	1.2	13
24	Reduced immunoglobulin gene diversity in patients with Cornelia de Lange syndrome. Journal of Allergy and Clinical Immunology, 2018, 141, 408-411.e8.	2.9	6
25	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	12.6	1,085
26	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
27	EIF2S3Mutations Associated with Severe X-Linked Intellectual Disability Syndrome MEHMO. Human Mutation, 2017, 38, 409-425.	2.5	57
28	4.7â€‰Mb deletion encompassing TGFB2 associated with features of Loeysâ€“Dietz syndrome and osteoporosis in adulthood. American Journal of Medical Genetics, Part A, 2017, 173, 2289-2292.	1.2	4
29	DDX3X mutations in two girls with a phenotype overlapping Toriælloâ€“Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373.	1.2	41
30	A Biallelic Mutation in the Homologous Recombination Repair Gene SPIDR Is Associated With Human Gonadal Dysgenesis. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 681-688.	3.6	47
31	Mutations of PTPN23 in developmental and epileptic encephalopathy. Human Genetics, 2017, 136, 1455-1461.	3.8	15
32	Diagnostic and prognostic significance of neurofilament light chain NF-L, but not progranulin and S100B, in the course of amyotrophic lateral sclerosis: Data from the German MND-net. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 112-119.	1.7	63
33	Estimated prevalence of potentially damaging variants in the leptin gene. Molecular and Cellular Pediatrics, 2017, 4, 10.	1.8	19
34	Meta-analysis Reveals Genome-Wide Significance at 15q13 for Nonsyndromic Clefting of Both the Lip and the Palate, and Functional Analyses Implicate GREM1 As a Plausible Causative Gene. PLoS Genetics, 2016, 12, e1005914.	3.5	66
35	Femoral facial syndrome associated with a de novo complex chromosome 2q37 rearrangement. American Journal of Medical Genetics, Part A, 2016, 170, 1202-1207.	1.2	9
36	Prevalence ofBRCA1/2germline mutations in 21â€…401 families with breast and ovarian cancer. Journal of Medical Genetics, 2016, 53, 465-471.	3.2	179

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37	Identification of two novel ALS2 mutations in infantile-onset ascending hereditary spastic paraplegia. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 260-265.	1.7	12
38	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. Nature Genetics, 2016, 48, 856-866.	21.4	520
39	Early-onset familial hemiplegic migraine due to a novel SCN1A mutation. Cephalalgia, 2016, 36, 1238-1247.	3.9	36
40	Sequencing the GRHL3 Coding Region Reveals Rare Truncating Mutations and a Common Susceptibility Variant for Nonsyndromic Cleft Palate. American Journal of Human Genetics, 2016, 98, 755-762.	6.2	92
41	NEK1mutations in familial amyotrophic lateral sclerosis. Brain, 2016, 139, e28-e28.	7.6	105
42	Exome sequencing and CRISPR/Cas genome editing identify mutations of ZAK as a cause of limb defects in humans and mice. Genome Research, 2016, 26, 183-191.	5.5	52
43	Screening forCHCHD10mutations in a large cohort of sporadic ALS patients: no evidence for pathogenicity of the p.P34S variant: Table 1. Brain, 2016, 139, e8-e8.	7.6	20
44	A hypomorphic BMPR1B mutation causes du Pan acromesomelic dysplasia. Orphanet Journal of Rare Diseases, 2015, 10, 84.	2.7	18
45	A post GWAS association study of SNPs associated with cleft lip with or without cleft palate in submucous cleft palate. American Journal of Medical Genetics, Part A, 2015, 167, 670-673.	1.2	9
46	Genome wide association study identifies variants in NBEA associated with migraine in bipolar disorder. Journal of Affective Disorders, 2015, 172, 453-461.	4.1	15
47	BRF1 mutations alter RNA polymerase IIIâ€“dependent transcription and cause neurodevelopmental anomalies. Genome Research, 2015, 25, 155-166.	5.5	85
48	STIL mutation causes autosomal recessive microcephalic lobar holoprosencephaly. Human Genetics, 2015, 134, 45-51.	3.8	32
49	Exome sequencing identifies a novel heterozygous TGFB3 mutation in a disorder overlapping with Marfan and Loeys-Dietz syndrome. Molecular and Cellular Probes, 2015, 29, 330-334.	2.1	22
50	ILDR1 null mice, a model of human deafness DFNB42, show structural aberrations of tricellular tight junctions and degeneration of auditory hair cells. Human Molecular Genetics, 2015, 24, 609-624.	2.9	58
51	A recurrent synonymous KAT6B mutation causes Sayâ€Barberâ€Biesecker/Youngâ€Simpson syndrome by inducing aberrant splicing. American Journal of Medical Genetics, Part A, 2015, 167, 3006-3010.	1.2	17
52	KAT6B Is a Tumor Suppressor Histone H3 Lysine 23 Acetyltransferase Undergoing Genomic Loss in Small Cell Lung Cancer. Cancer Research, 2015, 75, 3936-3945.	0.9	65
53	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
54	Homozygous missense and nonsense mutations in BMPR1B cause acromesomelic chondrodysplasia-type Grebe. European Journal of Human Genetics, 2014, 22, 726-733.	2.8	23

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55	Dopamine transporter deficiency syndrome: phenotypic spectrum from infancy to adulthood. Brain, 2014, 137, 1107-1119.	7.6	265
56	High mutation detection rates in cerebral cavernous malformation upon stringent inclusion criteria: oneâ€‘third of probands are minors. Molecular Genetics & Genomic Medicine, 2014, 2, 176-185.	1.2	53
57	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. Human Genetics, 2014, 133, 939-949.	3.8	29
58	Homozygous truncating PTPRF mutation causes athelia. Human Genetics, 2014, 133, 1041-1047.	3.8	10
59	Blepharophimosis, short humeri, developmental delay and hirschsprung disease: Expanding the phenotypic spectrum of MED12 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 1821-1825.	1.2	19
60	Biallelic SZT2 Mutations Cause Infantile Encephalopathy with Epilepsy and Dysmorphic Corpus Callosum. American Journal of Human Genetics, 2013, 93, 524-529.	6.2	81
61	De novo mutations of the gene encoding the histone acetyltransferase KAT6B in two patients with Sayâ€‘Barber/Biesecker/Youngâ€‘Simpson syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 884-888.	1.2	34
62	Mutation Spectrum inRAB3GAP1,RAB3GAP2, andRAB18and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. Human Mutation, 2013, 34, 686-696.	2.5	114
63	Mutations in GMPPA Cause a Glycosylation Disorder Characterized by Intellectual Disability and Autonomic Dysfunction. American Journal of Human Genetics, 2013, 93, 727-734.	6.2	57
64	Genome-wide meta-analysis identifies new susceptibility loci for migraine. Nature Genetics, 2013, 45, 912-917.	21.4	338
65	A regulatory role for the cohesin loader NIPBL in nonhomologous end joining during immunoglobulin class switch recombination. Journal of Experimental Medicine, 2013, 210, 2503-2513.	8.5	33
66	A Novel <i>MYO6</i> Splice Site Mutation Causes Autosomal Dominant Sensorineural Hearing Loss Type DFNA22 with a Favourable Outcome after Cochlear Implantation. Audiology and Neuro-Otology, 2013, 18, 192-199.	1.3	13
67	Exon skipping and severe childhoodâ€‘onset obesity caused by a leptin receptor mutation. American Journal of Medical Genetics, Part A, 2013, 161, 2672-2674.	1.2	10
68	Testosterone production during puberty in two 46,XY patients with disorders of sex development and novel NR5A1 (SF-1) mutations. European Journal of Endocrinology, 2012, 167, 125-130.	3.7	40
69	eIF2Î³ Mutation that Disrupts eIF2 Complex Integrity Links Intellectual Disability to Impaired Translation Initiation. Molecular Cell, 2012, 48, 641-646.	9.7	63
70	A homozygous splice site mutation in TRAPPC9 causes intellectual disability and microcephaly. European Journal of Medical Genetics, 2012, 55, 727-731.	1.3	41
71	A Mutation in PNPT1, Encoding Mitochondrial-RNA-Import Protein PNPase, Causes Hereditary Hearing Loss. American Journal of Human Genetics, 2012, 91, 919-927.	6.2	82
72	Deficiency for the Ubiquitin Ligase UBE3B in a Blepharophimosis-Ptosis-Intellectual-Disability Syndrome. American Journal of Human Genetics, 2012, 91, 998-1010.	6.2	82

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73	An Alu repeat-mediated genomic GCNT2 deletion underlies congenital cataracts and adult i blood group. Human Genetics, 2012, 131, 209-216.	3.8	20
74	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	6.2	108
75	Adaptor Protein Complex 4 Deficiency Causes Severe Autosomal-Recessive Intellectual Disability, Progressive Spastic Paraplegia, Shy Character, and Short Stature. American Journal of Human Genetics, 2011, 88, 788-795.	6.2	206
76	A homozygous RAB3GAP2 mutation causes Warburg Micro syndrome. Human Genetics, 2011, 129, 45-50.	3.8	79
77	A mutation screen in patients with Kabuki syndrome. Human Genetics, 2011, 130, 715-724.	3.8	106
78	Arterial rupture in classic Ehlersâ€“Danlos syndrome with COL5A1 mutation. American Journal of Medical Genetics, Part A, 2010, 152A, 2090-2093.	1.2	52
79	Genome-wide association study of migraine implicates a common susceptibility variant on 8q22.1. Nature Genetics, 2010, 42, 869-873.	21.4	332
80	Genetic Causes of Goiter and Deafness: Pendred Syndrome in a Girl and Cooccurrence of Pendred Syndrome and Resistance to Thyroid Hormone in Her Sister. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 2106-2109.	3.6	6
81	Compound heterozygous ASPM mutations associated with microcephaly and simplified cortical gyration in a consanguineous Algerian family. European Journal of Medical Genetics, 2009, 52, 180-184.	1.3	25
82	Clinical, cellular, and neuropathological consequences ofAP1S2mutations: further delineation of a recognizable X-linked mental retardation syndrome. Human Mutation, 2008, 29, 966-974.	2.5	41
83	New case of interstitial deletion 12(q15â€q21.2) in a girl with facial dysmorphism and mental retardation. American Journal of Medical Genetics, Part A, 2008, 146A, 93-96.	1.2	16
84	Incidence and clinical features of X-linked Cornelia de Lange syndrome due toSMC1L1 mutations. Human Mutation, 2007, 28, 205-206.	2.5	71
85	Goitrous Congenital Hypothyroidism and Hearing Impairment Associated with Mutations in the TPO and SLC26A4/PDS Genes. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 2678-2681.	3.6	46
86	Father-to-daughter transmission of Cornelia de Lange syndrome caused by a mutation in the 5â€² untranslated region of theNIPBL Gene. Human Mutation, 2006, 27, 731-735.	2.5	58
87	Molecular karyotyping in human constitutional cytogenetics. European Journal of Medical Genetics, 2005, 48, 214-231.	1.3	24
88	Intrafamilial Variability of the Deafness and Goiter Phenotype in Pendred Syndrome Caused by a T416P Mutation in the SLC26A4 Gene. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 5347-5351.	3.6	28
89	Four New Cases of Congenital Secondary Hypothyroidism due to a Splice Site Mutation in the Thyrotropin-Î² Gene: Phenotypic Variability and Founder Effect. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 4136-4141.	3.6	33
90	Mutations in the PDS Gene in German Families with Pendredâ€™s Syndrome: V138F Is a Founder Mutation. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 2916-2921.	3.6	35

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91	Angiotensin-Converting Enzyme and Heart Chymase gene Polymorphisms in Hypertrophic Cardiomyopathy**This study was supported by a grant-in-aid from the Bundesministerium fÃ¼r Bildung		