

Guntram Borck

List of Publications by Year in descending order

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Version: 2024-02-01

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	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , 2018, 360, .	12.6	1,085
2	Meta-analysis of 375,000 individuals identifies 38 susceptibility loci for migraine. <i>Nature Genetics</i> , 2016, 48, 856-866.	21.4	520
3	Genome-wide meta-analysis identifies new susceptibility loci for migraine. <i>Nature Genetics</i> , 2013, 45, 912-917.	21.4	338
4	Genome-wide association study of migraine implicates a common susceptibility variant on 8q22.1. <i>Nature Genetics</i> , 2010, 42, 869-873.	21.4	332
5	Dopamine transporter deficiency syndrome: phenotypic spectrum from infancy to adulthood. <i>Brain</i> , 2014, 137, 1107-1119.	7.6	265
6	Adaptor Protein Complex 4 Deficiency Causes Severe Autosomal-Recessive Intellectual Disability, Progressive Spastic Paraplegia, Shy Character, and Short Stature. <i>American Journal of Human Genetics</i> , 2011, 88, 788-795.	6.2	206
7	Prevalence of <i>BRCA1/2</i> germline mutations in 21â€¦401 families with breast and ovarian cancer. <i>Journal of Medical Genetics</i> , 2016, 53, 465-471.	3.2	179
8	Hot-spot KIF5A mutations cause familial ALS. <i>Brain</i> , 2018, 141, 688-697.	7.6	167
9	Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants. <i>Genetics in Medicine</i> , 2018, 20, 1175-1185.	2.4	133
10	Mutation Spectrum in <i>RAB3</i> , <i>GAP1</i> , <i>RAB3</i> , <i>GAP2</i> , and <i>RAB18</i> and Genotype-Phenotype Correlations in Warburg Micro Syndrome and Martsolf Syndrome. <i>Human Mutation</i> , 2013, 34, 686-696.	2.5	114
11	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. <i>American Journal of Human Genetics</i> , 2011, 88, 127-137.	6.2	108
12	A mutation screen in patients with Kabuki syndrome. <i>Human Genetics</i> , 2011, 130, 715-724.	3.8	106
13	<i>NEK1</i> mutations in familial amyotrophic lateral sclerosis. <i>Brain</i> , 2016, 139, e28-e28.	7.6	105
14	Sequencing the GRHL3 Coding Region Reveals Rare Truncating Mutations and a Common Susceptibility Variant for Nonsyndromic Cleft Palate. <i>American Journal of Human Genetics</i> , 2016, 98, 755-762.	6.2	92
15	<i>BRF1</i> mutations alter RNA polymerase IIIâ€œdependent transcription and cause neurodevelopmental anomalies. <i>Genome Research</i> , 2015, 25, 155-166.	5.5	85
16	A Mutation in PNPT1, Encoding Mitochondrial-RNA-Import Protein PNPase, Causes Hereditary Hearing Loss. <i>American Journal of Human Genetics</i> , 2012, 91, 919-927.	6.2	82
17	Deficiency for the Ubiquitin Ligase UBE3B in a Blepharophimosis-Ptosis-Intellectual-Disability Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 998-1010.	6.2	82
18	Biallelic SZT2 Mutations Cause Infantile Encephalopathy with Epilepsy and Dysmorphic Corpus Callosum. <i>American Journal of Human Genetics</i> , 2013, 93, 524-529.	6.2	81

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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	A homozygous RAB3GAP2 mutation causes Warburg Micro syndrome. Human Genetics, 2011, 129, 45-50.	3.8	79
21	Incidence and clinical features of X-linked Cornelia de Lange syndrome due toSMC1L1 mutations. Human Mutation, 2007, 28, 205-206.	2.5	71
22	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
23	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
24	Angiotensin-Converting Enzyme and Heart Chymase gene Polymorphisms in Hypertrophic Cardiomyopathy**This study was supported by a grant-in-aid from the Bundesministerium für Bildung		

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37	A Biallelic Mutation in the Homologous Recombination Repair Gene SPIDR Is Associated With Human Gonadal Dysgenesis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017, 102, 681-688.	3.6	47
38	Functional and Phenotypic Characteristics of Human Leptin Receptor Mutations. <i>Journal of the Endocrine Society</i> , 2019, 3, 27-41.	0.2	47
39	Goitrous Congenital Hypothyroidism and Hearing Impairment Associated with Mutations in the TPO and SLC26A4/PDS Genes. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2006, 91, 2678-2681.	3.6	46
40	Early childhood BMI trajectories in monogenic obesity due to leptin, leptin receptor, and melanocortin 4 receptor deficiency. <i>International Journal of Obesity</i> , 2018, 42, 1602-1609.	3.4	44
41	Noncoding copy-number variations are associated with congenital limb malformation. <i>Genetics in Medicine</i> , 2018, 20, 599-607.	2.4	42
42	Clinical, cellular, and neuropathological consequences of <i>AP1S2</i> mutations: further delineation of a recognizable X-linked mental retardation syndrome. <i>Human Mutation</i> , 2008, 29, 966-974.	2.5	41
43	A homozygous splice site mutation in TRAPPC9 causes intellectual disability and microcephaly. <i>European Journal of Medical Genetics</i> , 2012, 55, 727-731.	1.3	41
44	<i>DDX3X</i> mutations in two girls with a phenotype overlapping Toriello's "Carey syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1369-1373.	1.2	41
45	Testosterone production during puberty in two 46,XY patients with disorders of sex development and novel NR5A1 (SF-1) mutations. <i>European Journal of Endocrinology</i> , 2012, 167, 125-130.	3.7	40
46	Early-onset familial hemiplegic migraine due to a novel <i>SCN1A</i> mutation. <i>Cephalalgia</i> , 2016, 36, 1238-1247.	3.9	36
47	Mutations in the PDS Gene in German Families with Pendred's Syndrome: V138F Is a Founder Mutation. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2003, 88, 2916-2921.	3.6	35
48	De novo mutations of the gene encoding the histone acetyltransferase KAT6B in two patients with Say-Barber/Biesecker/Young-Simpson syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 884-888.	1.2	34
49	Cross-trait analyses with migraine reveal widespread pleiotropy and suggest a vascular component to migraine headache. <i>International Journal of Epidemiology</i> , 2020, 49, 1022-1031.	1.9	34
50	Four New Cases of Congenital Secondary Hypothyroidism due to a Splice Site Mutation in the Thyrotropin- β Gene: Phenotypic Variability and Founder Effect. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2004, 89, 4136-4141.	3.6	33
51	A regulatory role for the cohesin loader NIPBL in nonhomologous end joining during immunoglobulin class switch recombination. <i>Journal of Experimental Medicine</i> , 2013, 210, 2503-2513.	8.5	33
52	STIL mutation causes autosomal recessive microcephalic lobar holoprosencephaly. <i>Human Genetics</i> , 2015, 134, 45-51.	3.8	32
53	Association Between Germline Mutations in BRF1, a Subunit of the RNA Polymerase III Transcription Complex, and Hereditary Colorectal Cancer. <i>Gastroenterology</i> , 2018, 154, 181-194.e20.	1.3	32
54	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. <i>Human Mutation</i> , 2018, 39, 1246-1261.	2.5	31

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55	Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. <i>Human Genetics</i> , 2014, 133, 939-949.	3.8	29
56	Intrafamilial Variability of the Deafness and Goiter Phenotype in Pendred Syndrome Caused by a T416P Mutation in the SLC26A4 Gene. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2004, 89, 5347-5351.	3.6	28
57	Next-generation sequencing of 32 genes associated with hereditary aortopathies and related disorders of connective tissue in a cohort of 199 patients. <i>Genetics in Medicine</i> , 2019, 21, 1832-1841.	2.4	26
58	Compound heterozygous ASPM mutations associated with microcephaly and simplified cortical gyration in a consanguineous Algerian family. <i>European Journal of Medical Genetics</i> , 2009, 52, 180-184.	1.3	25
59	Molecular karyotyping in human constitutional cytogenetics. <i>European Journal of Medical Genetics</i> , 2005, 48, 214-231.	1.3	24
60	Homozygous missense and nonsense mutations in BMPR1B cause acromesomelic chondrodysplasia-type Grebe. <i>European Journal of Human Genetics</i> , 2014, 22, 726-733.	2.8	23
61	SQSTM1/p62 variants in 486 patients with familial ALS from Germany and Sweden. <i>Neurobiology of Aging</i> , 2020, 87, 139.e9-139.e15.	3.1	23
62	Exome sequencing identifies a novel heterozygous TGFB3 mutation in a disorder overlapping with Marfan and Loeys-Dietz syndrome. <i>Molecular and Cellular Probes</i> , 2015, 29, 330-334.	2.1	22
63	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. <i>European Journal of Human Genetics</i> , 2018, 26, 695-708.	2.8	22
64	An Alu repeat-mediated genomic GCNT2 deletion underlies congenital cataracts and adult i blood group. <i>Human Genetics</i> , 2012, 131, 209-216.	3.8	20
65	Screening for <i>CHCHD10</i> mutations in a large cohort of sporadic ALS patients: no evidence for pathogenicity of the p.P34S variant: Table 1. <i>Brain</i> , 2016, 139, e8-e8.	7.6	20
66	Blepharophimosis, short humeri, developmental delay and hirschsprung disease: Expanding the phenotypic spectrum of <i>MED12</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1821-1825.	1.2	19
67	Estimated prevalence of potentially damaging variants in the leptin gene. <i>Molecular and Cellular Pediatrics</i> , 2017, 4, 10.	1.8	19
68	A hypomorphic BMPR1B mutation causes du Pan acromesomelic dysplasia. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 84.	2.7	18
69	Habitual sleep disturbances and migraine: a Mendelian randomization study. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 2370-2380.	3.7	18
70	The murine ortholog of Kaufman oculocerebrofacial syndrome protein Ube3b regulates synapse number by ubiquitinating Ppp3cc. <i>Molecular Psychiatry</i> , 2021, 26, 1980-1995.	7.9	18
71	A recurrent synonymous <i>KAT6B</i> mutation causes Say-Barber-Biesecker/Young-Simpson syndrome by inducing aberrant splicing. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 3006-3010.	1.2	17
72	New case of interstitial deletion 12(q15-q21.2) in a girl with facial dysmorphism and mental retardation. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 93-96.	1.2	16

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73	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. <i>Human Genetics</i> , 2019, 138, 593-600.	3.8	16
74	Genome wide association study identifies variants in NBEA associated with migraine in bipolar disorder. <i>Journal of Affective Disorders</i> , 2015, 172, 453-461.	4.1	15
75	Mutations of PTPN23 in developmental and epileptic encephalopathy. <i>Human Genetics</i> , 2017, 136, 1455-1461.	3.8	15
76	A Novel MYO6 Splice Site Mutation Causes Autosomal Dominant Sensorineural Hearing Loss Type DFNA22 with a Favourable Outcome after Cochlear Implantation. <i>Audiology and Neuro-Otology</i> , 2013, 18, 192-199.	1.3	13
77	Kaufman oculocerebrofacial syndrome: Novel UBE3B mutations and clinical features in four unrelated patients. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 187-193.	1.2	13
78	Identification of two novel ALS2 mutations in infantile-onset ascending hereditary spastic paraplegia. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2016, 17, 260-265.	1.7	12
79	Exon skipping and severe childhood-onset obesity caused by a leptin receptor mutation. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2672-2674.	1.2	10
80	Homozygous truncating PTPRF mutation causes athelia. <i>Human Genetics</i> , 2014, 133, 1041-1047.	3.8	10
81	Expanding the phenotype associated with biallelic WDR60 mutations: Siblings with retinal degeneration and polydactyly lacking other features of short rib thoracic dystrophies. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 438-442.	1.2	10
82	A post GWAS association study of SNPs associated with cleft lip with or without cleft palate in submucous cleft palate. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 670-673.	1.2	9
83	Femoral facial syndrome associated with a de novo complex chromosome 2q37 rearrangement. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1202-1207.	1.2	9
84	Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e539.	1.2	9
85	A novel homozygous missense variant in NECTIN4 (PVRL4) causing ectodermal dysplasia cutaneous syndactyly syndrome. <i>Annals of Human Genetics</i> , 2018, 82, 232-238.	0.8	7
86	Elucidating the relationship between migraine risk and brain structure using genetic data. <i>Brain</i> , 2022, 145, 3214-3224.	7.6	7
87	Genetic Causes of Goiter and Deafness: Pendred Syndrome in a Girl and Cooccurrence of Pendred Syndrome and Resistance to Thyroid Hormone in Her Sister. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 2106-2109.	3.6	6
88	Reduced immunoglobulin gene diversity in patients with Cornelia de Lange syndrome. <i>Journal of Allergy and Clinical Immunology</i> , 2018, 141, 408-411.e8.	2.9	6
89	4.7 Mb deletion encompassing TGFβ2 associated with features of Loey's-Dietz syndrome and osteoporosis in adulthood. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2289-2292.	1.2	4
90	Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. <i>Brain</i> , 2019, 142, e67-e67.	7.6	1

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91	Genotypes of amyotrophic lateral sclerosis in Mongolia. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1300-1302.	1.9	1