Hans-Hilger Ropers

List of Publications by Year
in descending order

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49 papers	5,117 citations	159585 30 h-index	206112 48 g-index
50 all docs	50 docs citations	50 times ranked	7437 citing authors

#	Article	IF	Citations
1	Rare diseases: human genome research is coming home.. Cold Spring Harbor Molecular Case Studies, 2022, 8, .	1.0	0
2	Comprehensive <scp>genotypeâ€phenotype</scp> correlation in <scp>AP</scp>â€4 deficiency syndrome; Adding data from a large cohort of Iranian patients. Clinical Genetics, 2021, 99, 187-192.	2.0	2
3	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
4	Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1418.	1.2	1
5	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. Zeitschrift FÃœr Kinder- Und Jugendpsychiatrie Und Psychotherapie, 2020, 48, 478-489.	0.7	5
6	Identification of diseaseâ€causing variants in the EXOSC gene family underlying autosomal recessive intellectual disability in Iranian families. Clinical Genetics, 2019, 95, 718-725.	2.0	5
7	A mouse model for intellectual disability caused by mutations in the X-linked 2â€²â€²Oâ€methyltransferase Ftsj1 gene. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2083-2093.	3.8	17
8	Effect of inbreeding on intellectual disability revisited by trio sequencing. Clinical Genetics, 2019, 95, 151-159.	2.0	49
9	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	7.9	131
10	Pathogenic variants in E3 ubiquitin ligase RLIM/RNF12 lead to a syndromic X-linked intellectual disability and behavior disorder. Molecular Psychiatry, 2019, 24, 1748-1768.	7.9	26
11	KlÃ¼verâ€“Bucy syndrome associated with a recessive variant in HGSNAT in two siblings with Mucopolysaccharidosis type IIIC (Sanfilippo C). European Journal of Human Genetics, 2017, 25, 253-256.	2.8	5
12	Homozygous ARHGEF2 mutation causes intellectual disability and midbrain-hindbrain malformation. PLoS Genetics, 2017, 13, e1006746.	3.5	27
13	Homozygous YME1L1 mutation causes mitochondriopathy with optic atrophy and mitochondrial network fragmentation. ELife, 2016, 5, .	6.0	88
14	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
15	Comprehensive genotyping and clinical characterisation reveal 27 novel NKX2-1 mutations and expand the phenotypic spectrum. Journal of Medical Genetics, 2014, 51, 375-387.	3.2	77
16	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	2.9	19
17	Previously reported new type of autosomal recessive primary microcephaly is caused by compound heterozygousASPMgene mutations. Cell Cycle, 2014, 13, 1650-1651.	2.6	8
18	On the future of genetic risk assessment. Journal of Community Genetics, 2012, 3, 229-236.	1.2	69

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19	A Noncoding, Regulatory Mutation Implicates HCFC1 in Nonsyndromic Intellectual Disability. American Journal of Human Genetics, 2012, 91, 694-702.	6.2	89
20	Breakpoint analysis of balanced chromosome rearrangements by next-generation paired-end sequencing. European Journal of Human Genetics, 2010, 18, 539-543.	2.8	61
21	Single gene disorders come into focus--again. Dialogues in Clinical Neuroscience, 2010, 12, 95-102.	3.7	9
22	Single gene disorders come into focus - again. Dialogues in Clinical Neuroscience, 2010, 12, 95-102.	3.7	10
23	A systematic, large-scale resequencing screen of X-chromosome coding exons in mental retardation. Nature Genetics, 2009, 41, 535-543.	21.4	528
24	A New Chromosome X Exon-Specific Microarray Platform for Screening of Patients with X-Linked Disorders. Journal of Molecular Diagnostics, 2009, 11, 562-568.	2.8	8
25	Submicroscopic Duplications of the Hydroxysteroid Dehydrogenase HSD17B10 and the E3 Ubiquitin Ligase HUWE1 Are Associated with Mental Retardation. American Journal of Human Genetics, 2008, 82, 432-443.	6.2	187
26	Screening of 20 patients with X-linked mental retardation using chromosome X-specific array-MAPH. European Journal of Medical Genetics, 2007, 50, 399-410.	1.3	8
27	Complex Inheritance Pattern Resembling Autosomal Recessive Inheritance Involving a Microdeletion in Thrombocytopeniaâ€“Absent Radius Syndrome. American Journal of Human Genetics, 2007, 80, 232-240.	6.2	290
28	New Perspectives for the Elucidation of Genetic Disorders. American Journal of Human Genetics, 2007, 81, 199-207.	6.2	119
29	Mutation frequencies of X-linked mental retardation genes in families from the EuroMRX consortium. Human Mutation, 2007, 28, 207-208.	2.5	103
30	X-linked mental retardation: a comprehensive molecular screen of 47 candidate genes from a 7.4 Mb interval in Xp11. European Journal of Human Genetics, 2007, 15, 68-75.	2.8	19
31	Mutations in autism susceptibility candidate 2 (AUTS2) in patients with mental retardation. Human Genetics, 2007, 121, 501-509.	3.8	116
32	Loss of SLC38A5 and FTSJ1 at Xp11.23 in three brothers with non-syndromic mental retardation due to a microdeletion in an unstable genomic region. Human Genetics, 2007, 121, 539-547.	3.8	46
33	ZNF674: A New KrÃ¼ppel-Associated Boxâ€“Containing Zinc-Finger Gene Involved in Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2006, 78, 265-278.	6.2	75
34	Breakpoint Cloning and Haplotype Analysis Indicate a Single Origin of the Common Inv(10)(p11.2q21.2) Mutation among Northern Europeans. American Journal of Human Genetics, 2006, 78, 878-883.	6.2	23
35	X-linked mental retardation: many genes for a complex disorder. Current Opinion in Genetics and Development, 2006, 16, 260-269.	3.3	147
36	Disruptions of the novel KIAA1202 gene are associated with X-linked mental retardation. Human Genetics, 2006, 118, 578-590.	3.8	55

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37	A novel X-linked recessive mental retardation syndrome comprising macrocephaly and ciliary dysfunction is allelic to oralâ€“facialâ€“digital type I syndrome. Human Genetics, 2006, 120, 171-178.	3.8	166
38	NovelJARID1C/SMCX mutations in patients with X-linked mental retardation. Human Mutation, 2006, 27, 389-389.	2.5	120
39	Mutations in the JARID1C Gene, Which Is Involved in Transcriptional Regulation and Chromatin Remodeling, Cause X-Linked Mental Retardation. American Journal of Human Genetics, 2005, 76, 227-236.	6.2	349
40	X-Linked Mental Retardation and Autism Are Associated with a Mutation in the NLGN4 Gene, a Member of the Neuroligin Family. American Journal of Human Genetics, 2004, 74, 552-557.	6.2	686
41	High Prevalence of SLC6A8 Deficiency in X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 97-105.	6.2	193
42	Mutations in the FTSJ1 Gene Coding for a Novel S-Adenosylmethionineâ€“Binding Protein Cause Nonsyndromic X-Linked Mental Retardation. American Journal of Human Genetics, 2004, 75, 305-309.	6.2	117
43	Reply to Mandel. American Journal of Human Genetics, 2004, 75, 731-732.	6.2	1
44	Nonsyndromic X-linked mental retardation: where are the missing mutations?. Trends in Genetics, 2003, 19, 316-320.	6.7	65
45	Mutations in the polyglutamine binding protein 1 gene cause X-linked mental retardation. Nature Genetics, 2003, 35, 313-315.	21.4	139
46	Mutations in the ZNF41 Gene Are Associated with Cognitive Deficits: Identification of a New Candidate for X-Linked Mental Retardation. American Journal of Human Genetics, 2003, 73, 1341-1354.	6.2	83
47	FACL4, encoding fatty acid-CoA ligase 4, is mutated in nonspecific X-linked mental retardation. Nature Genetics, 2002, 30, 436-440.	21.4	135
48	MECP2 is highly mutated in X-linked mental retardation. Human Molecular Genetics, 2001, 10, 941-946.	2.9	238
49	Mutations in ARHGEF6, encoding a guanine nucleotide exchange factor for Rho GTPases, in patients with X-linked mental retardation. Nature Genetics, 2000, 26, 247-250.	21.4	329