Ute Moog

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4588482/publications.pdf

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46	2,656	304602	254106
papers	citations	22 h-index	43 g-index

48	48	48	6579
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Range of genetic mutations associated with severe non-syndromic sporadic intellectual disability: an exome sequencing study. Lancet, The, 2012, 380, 1674-1682.	6.3	940
2	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	9.4	137
3	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
4	Phenotypic spectrum associated with CASK loss-of-function mutations. Journal of Medical Genetics, 2011, 48, 741-751.	1.5	114
5	Next-generation sequencing in X-linked intellectual disability. European Journal of Human Genetics, 2015, 23, 1513-1518.	1.4	112
6	A Mild PUM1 Mutation Is Associated with Adult-Onset Ataxia, whereas Haploinsufficiency Causes Developmental Delay and Seizures. Cell, 2018, 172, 924-936.e11.	13.5	103
7	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	3.7	102
8	A targeted next-generation sequencing assay for the molecular diagnosis of genetic disorders with orodental involvement. Journal of Medical Genetics, 2016, 53, 98-110.	1.5	100
9	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	2.6	88
10	Phenotypic and molecular insights into CASK-related disorders in males. Orphanet Journal of Rare Diseases, 2015, 10, 44.	1.2	68
11	FOXP2variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	1.5	67
12	Is Sanfilippo type B in your mind when you see adults with mental retardation and behavioral problems?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2007, 145C, 293-301.	0.7	58
13	Brain anomalies in encephalocraniocutaneous lipomatosis. American Journal of Medical Genetics, Part A, 2007, 143A, 2963-2972.	0.7	56
14	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	0.5	50
15	Homozygous loss of CHRNA7 on chromosome 15q13.3 causes severe encephalopathy with seizures and hypotonia. American Journal of Medical Genetics, Part A, 2010, 152A, 2908-2911.	0.7	48
16	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
17	DDX3X mutations in two girls with a phenotype overlapping Torielloâ€“Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373.	0.7	41
18	Diagnosis of CoPAN by whole exome sequencing: Waking up a sleeping tiger's eye. American Journal of Medical Genetics, Part A, 2017, 173, 1878-1886.	0.7	40

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19	Encephalocraniocutaneous lipomatosis accompanied by the formation of bone cysts: Harboring clues to pathogenesis?. American Journal of Medical Genetics, Part A, 2007, 143A, 2973-2980.	0.7	38
20	Trichothiodystrophy causative TFIIEÎ² mutation affects transcription in highly differentiated tissue. Human Molecular Genetics, 2017, 26, 4689-4698.	1.4	38
21	Acampomelic campomelic syndrome. American Journal of Medical Genetics Part A, 2001, 104, 239-245.	2.4	32
22	Clinical spectrum of females with HCCS mutation: from no clinical signs to a neonatal lethal form of the microphthalmia with linear skin defects (MLS) syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 53.	1.2	26
23	POLR3A variants with striatal involvement and extrapyramidal movement disorder. Neurogenetics, 2020, 21, 121-133.	0.7	24
24	Sequencing of a Patient with Balanced Chromosome Abnormalities and Neurodevelopmental Disease Identifies Disruption of Multiple High Risk Loci by Structural Variation. PLoS ONE, 2014, 9, e90894.	1.1	20
25	Mosaic deletion of EXOC6B: Further evidence for an important role of the exocyst complex in the pathogenesis of intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 3088-3094.	0.7	17
26	SIPA1L3 identified by linkage analysis and whole-exome sequencing as a novel gene for autosomal recessive congenital cataract. European Journal of Human Genetics, 2015, 23, 1627-1633.	1.4	15
27	Exome sequencing reveals a novel CWF19L1 mutation associated with intellectual disability and cerebellar atrophy. American Journal of Medical Genetics, Part A, 2016, 170, 1502-1509.	0.7	13
28	Oculodentodigital Dysplasia: A Hypomyelinating Leukodystrophy with a Characteristic MRI Pattern of Brain Stem Involvement. American Journal of Neuroradiology, 2019, 40, 903-907.	1.2	13
29	The Frog Xenopus as a Model to Study Joubert Syndrome: The Case of a Human Patient With Compound Heterozygous Variants in PIBF1. Frontiers in Physiology, 2019, 10, 134.	1.3	13
30	Oculoectodermal syndrome: Report of a new case with a broad clinical spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 2947-2951.	0.7	12
31	Disorders Caused by Genetic Mosaicism. Deutsches Ärzteblatt International, 2020, 116, 119-125.	0.6	12
32	An update on oculocerebrocutaneous (Delleman-Oorthuys) syndrome. , 2018, 178, 414-422.		11
33	Frequency of KCNQ1 variants causing loss of methylation of Imprinting Centre 2 in Beckwith-Wiedemann syndrome. Clinical Epigenetics, 2020, 12, 63.	1.8	11
34	Characterization of the first intragenic SATB2 duplication in a girl with intellectual disability, nearly absent speech and suspected hypodontia. European Journal of Human Genetics, 2015, 23, 704-707.	1.4	10
35	Duplication Xp11.22â€p14 in females: Does Xâ€inactivation help in assessing their significance?. American Journal of Medical Genetics, Part A, 2015, 167, 553-562.	0.7	8
36	Uniparental isodisomy as a cause of recessive Mendelian disease: a diagnostic pitfall with a quick and easy solution in medium/large NGS analyses. European Journal of Human Genetics, 2018, 26, 1392-1395.	1.4	8

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37	What do parents expect from a genetic diagnosis of their child with intellectual disability?. Journal of Applied Research in Intellectual Disabilities, 2019, 32, 1129-1137.	1.3	8
38	Double germline mutations in APC and BRCA2 in an individual with a pancreatic tumor. Familial Cancer, 2017, 16, 303-309.	0.9	5
39	To Test or Not to Test? Metabolic Testing in Adolescents and Adults With Intellectual Disability. Journal of Policy and Practice in Intellectual Disabilities, 2008, 5, 167-173.	1.7	4
40	Invited comment. Acta Psychiatrica Scandinavica, 2010, 122, 166-166.	2.2	4
41	Genome-wide UPD screening in patients with intellectual disability. European Journal of Human Genetics, 2014, 22, 1233-1235.	1.4	3
42	Germ cell mosaicism for AUTS2 exon 6 deletion. American Journal of Medical Genetics, Part A, 2021, 185, 1261-1265.	0.7	2
43	The need for recognition of core professional groups in genetics healthcare services in Europe. European Journal of Human Genetics, 2022, 30, 639-640.	1.4	2
44	Acampomelic campomelic syndrome. American Journal of Medical Genetics Part A, 2001, 104, 239-245.	2.4	1
45	A boy with <scp>Silver</scp>â€“<scp>Russell</scp> syndrome and Sotos syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 549-554.	0.7	0
46	Recurrent sebaceous carcinoma on the shoulder. JDDG - Journal of the German Society of Dermatology, 2020, 18, 247-250.	0.4	0