Ute Moog

List of Publications by Year
in descending order

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

Version: 2024-02-01

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	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
2	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
3	Germ cell mosaicism for AUTS2 exon 6 deletion. American Journal of Medical Genetics, Part A, 2021, 185, 1261-1265.	0.7	2
4	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
5	Frequency of KCNQ1 variants causing loss of methylation of Imprinting Centre 2 in Beckwith-Wiedemann syndrome. Clinical Epigenetics, 2020, 12, 63.	1.8	11
6	POLR3A variants with striatal involvement and extrapyramidal movement disorder. Neurogenetics, 2020, 21, 121-133.	0.7	24
7	Recurrent sebaceous carcinoma on the shoulder. JDDG - Journal of the German Society of Dermatology, 2020, 18, 247-250.	0.4	0
8	Disorders Caused by Genetic Mosaicism. Deutsches Ärzteblatt International, 2020, 116, 119-125.	0.6	12
9	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
10	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
11	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
12	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
13	An update on oculocerebrocutaneous (Delleman-Oorthuys) syndrome. , 2018, 178, 414-422.		11
14	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
15	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
16	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
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	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

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19	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	3.7	102
20	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	0.5	50
21	Double germline mutations in APC and BRCA2 in an individual with a pancreatic tumor. Familial Cancer, 2017, 16, 303-309.	0.9	5
22	Trichothiodystrophy causative TFIIEÎ² mutation affects transcription in highly differentiated tissue. Human Molecular Genetics, 2017, 26, 4689-4698.	1.4	38
23	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
24	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	2.6	88
25	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
26	Phenotypic and molecular insights into CASK-related disorders in males. Orphanet Journal of Rare Diseases, 2015, 10, 44.	1.2	68
27	Transcriptional regulator PRDM12 is essential for human pain perception. Nature Genetics, 2015, 47, 803-808.	9.4	137
28	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
29	Next-generation sequencing in X-linked intellectual disability. European Journal of Human Genetics, 2015, 23, 1513-1518.	1.4	112
30	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
31	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
32	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
33	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
34	Genome-wide UPD screening in patients with intellectual disability. European Journal of Human Genetics, 2014, 22, 1233-1235.	1.4	3
35	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
36	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

#	Article	IF	Citations
37	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
38	Phenotypic spectrum associated with CASK loss-of-function mutations. Journal of Medical Genetics, 2011, 48, 741-751.	1.5	114
39	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
40	Invited comment. Acta Psychiatrica Scandinavica, 2010, 122, 166-166.	2.2	4
41	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
42	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
43	Brain anomalies in encephalocraniocutaneous lipomatosis. American Journal of Medical Genetics, Part A, 2007, 143A, 2963-2972.	0.7	56
44	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
45	Acampomelic campomelic syndrome. American Journal of Medical Genetics Part A, 2001, 104, 239-245.	2.4	32
46	Acampomelic campomelic syndrome. American Journal of Medical Genetics Part A, 2001, 104, 239-245.	2.4	1