MariÃ«lle Alders

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5930264/publications.pdf

Version: 2024-02-01

31	2,123	257101	433756
papers	citations	24 h-index	31 g-index

31	31	31	4474
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in CCBE1 cause generalized lymph vessel dysplasia in humans. Nature Genetics, 2009, 41, 1272-1274.	9.4	269
2	Clinical Aspects of Type 3 Long-QT Syndrome. Circulation, 2016, 134, 872-882.	1.6	162
3	A Mutation in CALM1 Encoding Calmodulin in Familial Idiopathic Ventricular Fibrillation in Childhood and Adolescence. Journal of the American College of Cardiology, 2014, 63, 259-266.	1.2	160
4	Titin gene mutations are common in families with both peripartum cardiomyopathy and dilated cardiomyopathy. European Heart Journal, 2014, 35, 2165-2173.	1.0	159
5	Haplotype-Sharing Analysis Implicates Chromosome 7q36 Harboring DPP6 in Familial IdiopathicÂVentricular Fibrillation. American Journal of Human Genetics, 2009, 84, 468-476.	2.6	158
6	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	2.6	138
7	The 2373insG mutation in the MYBPC3 gene is a founder mutation, which accounts for nearly one-fourth of the HCM cases in the Netherlands. European Heart Journal, 2003, 24, 1848-1853.	1.0	127
8	Hennekam syndrome can be caused by FAT4 mutations and be allelic to Van Maldergem syndrome. Human Genetics, 2014, 133, 1161-1167.	1.8	122
9	Mutations in TBL1X Are Associated With Central Hypothyroidism. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 4564-4573.	1.8	73
10	Homozygous Premature Truncation of the HERG Protein. Circulation, 1999, 100, 1264-1267.	1.6	67
11	A homozygous missense mutation in ERAL1, encoding a mitochondrial rRNA chaperone, causes Perrault syndrome. Human Molecular Genetics, 2017, 26, 2541-2550.	1.4	61
12	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	2.6	59
13	Determination of KCNQ1OT1 and H19 methylation levels in BWS and SRS patients using methylation-sensitive high-resolution melting analysis. European Journal of Human Genetics, 2009, 17, 467-473.	1.4	47
14	Mutations in Histone Acetylase Modifier BRPF1 Cause an Autosomal-Dominant Form of Intellectual Disability with Associated Ptosis. American Journal of Human Genetics, 2017, 100, 105-116.	2.6	46
15	CREBBP mutations in individuals without Rubinsteinâ€“Taybi syndrome phenotype. American Journal of Medical Genetics, Part A, 2016, 170, 2681-2693.	0.7	43
16	A de novo mutation in KCNN3 associated with autosomal dominant idiopathic non-cirrhotic portal hypertension. Journal of Hepatology, 2016, 64, 974-977.	1.8	42
17	Clues for Polygenic Inheritance of Pituitary Stalk Interruption Syndrome From Exome Sequencing in 20 Patients. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 415-428.	1.8	41
18	An inactivating mutation in the histone deacetylase SIRT6 causes human perinatal lethality. Genes and Development, 2018, 32, 373-388.	2.7	41

#	Article	IF	Citations
19	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. American Journal of Human Genetics, 2018, 103, 752-768.	2.6	40
20	Genetic Analyses in Small-for-Gestational-Age Newborns. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 917-925.	1.8	38
21	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	2.6	37
22	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogryposis. European Journal of Human Genetics, 2018, 26, 1752-1758.	1.4	32
23	Low rate of cardiac events in first-degree relatives of diagnosis-negative young sudden unexplained death syndrome victims during follow-up. Heart Rhythm, 2014, 11, 1728-1732.	0.3	30
24	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	1.1	28
25	Methylation analysis in tongue tissue of BWS patients identifies the (EPI)genetic cause in 3 patients with normal methylation levels in blood. European Journal of Medical Genetics, 2014, 57, 293-297.	0.7	27
26	Mutations in IRS4 are associated with central hypothyroidism. Journal of Medical Genetics, 2018, 55, 693-700.	1.5	27
27	Oralâ€facialâ€digital syndrome type 1 in males: Congenital heart defects are included in its phenotypic spectrum. American Journal of Medical Genetics, Part A, 2017, 173, 1383-1389.	0.7	16
28	Van Maldergem syndrome and Hennekam syndrome: Further delineation of allelic phenotypes. American Journal of Medical Genetics, Part A, 2018, 176, 1166-1174.	0.7	14
29	Variants in KAT6A and pituitary anomalies. American Journal of Medical Genetics, Part A, 2017, 173, 2562-2565.	0.7	12
30	Two Siblings With a CDKL5 Mutation. Journal of Child Neurology, 2015, 30, 1515-1519.	0.7	6
31	Prenatal NeuN+ neurons of Down syndrome display aberrant integrative DNA methylation and gene expression profiles. Epigenomics, 2022, 14, 375-390.	1.0	1