

MariÃ«lle Alders

List of Publications by Year in descending order

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Version: 2024-02-01

31
papers

2,123
citations

257101

24
h-index

433756

31
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31
all docs

31
docs citations

31
times ranked

4474
citing authors

#	ARTICLE	IF	CITATIONS
1	Prenatal NeuN+ neurons of Down syndrome display aberrant integrative DNA methylation and gene expression profiles. <i>Epigenomics</i> , 2022, 14, 375-390.	1.0	1
2	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 102, 309-320.	2.6	138
3	Clues for Polygenic Inheritance of Pituitary Stalk Interruption Syndrome From Exome Sequencing in 20 Patients. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 415-428.	1.8	41
4	Van Maldergem syndrome and Hennekam syndrome: Further delineation of allelic phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1166-1174.	0.7	14
5	Genetic Analyses in Small-for-Gestational-Age Newborns. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 917-925.	1.8	38
6	An inactivating mutation in the histone deacetylase SIRT6 causes human perinatal lethality. <i>Genes and Development</i> , 2018, 32, 373-388.	2.7	41
7	NFIB Haploinsufficiency Is Associated with Intellectual Disability and Macrocephaly. <i>American Journal of Human Genetics</i> , 2018, 103, 752-768.	2.6	40
8	Mutations in IRS4 are associated with central hypothyroidism. <i>Journal of Medical Genetics</i> , 2018, 55, 693-700.	1.5	27
9	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogyriposis. <i>European Journal of Human Genetics</i> , 2018, 26, 1752-1758.	1.4	32
10	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 1195-1203.	2.6	37
11	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 281-296.	2.6	59
12	Oral-facial-digital syndrome type 1 in males: Congenital heart defects are included in its phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1383-1389.	0.7	16
13	A homozygous missense mutation in ERAL1, encoding a mitochondrial rRNA chaperone, causes Perrault syndrome. <i>Human Molecular Genetics</i> , 2017, 26, 2541-2550.	1.4	61
14	Mutations in Histone Acetylase Modifier BRPF1 Cause an Autosomal-Dominant Form of Intellectual Disability with Associated Ptosis. <i>American Journal of Human Genetics</i> , 2017, 100, 105-116.	2.6	46
15	Equivalent missense variant in the <i>FOXP2</i> and <i>FOXP1</i> transcription factors causes distinct neurodevelopmental disorders. <i>Human Mutation</i> , 2017, 38, 1542-1554.	1.1	28
16	Variants in <i>KAT6A</i> and pituitary anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2562-2565.	0.7	12
17	<i>CREBBP</i> mutations in individuals without Rubinstein-Taybi syndrome phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2681-2693.	0.7	43
18	Clinical Aspects of Type 3 Long-QT Syndrome. <i>Circulation</i> , 2016, 134, 872-882.	1.6	162

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19	Mutations in <i>TBL1X</i> Are Associated With Central Hypothyroidism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 4564-4573.	1.8	73
20	A de novo mutation in <i>KCNN3</i> associated with autosomal dominant idiopathic non-cirrhotic portal hypertension. <i>Journal of Hepatology</i> , 2016, 64, 974-977.	1.8	42
21	Two Siblings With a <i>CDKL5</i> Mutation. <i>Journal of Child Neurology</i> , 2015, 30, 1515-1519.	0.7	6
22	Titin gene mutations are common in families with both peripartum cardiomyopathy and dilated cardiomyopathy. <i>European Heart Journal</i> , 2014, 35, 2165-2173.	1.0	159
23	A Mutation in <i>CALM1</i> Encoding Calmodulin in Familial Idiopathic Ventricular Fibrillation in Childhood and Adolescence. <i>Journal of the American College of Cardiology</i> , 2014, 63, 259-266.	1.2	160
24	Methylation analysis in tongue tissue of BWS patients identifies the (EPI)genetic cause in 3 patients with normal methylation levels in blood. <i>European Journal of Medical Genetics</i> , 2014, 57, 293-297.	0.7	27
25	Hennekam syndrome can be caused by <i>FAT4</i> mutations and be allelic to Van Maldergem syndrome. <i>Human Genetics</i> , 2014, 133, 1161-1167.	1.8	122
26	Low rate of cardiac events in first-degree relatives of diagnosis-negative young sudden unexplained death syndrome victims during follow-up. <i>Heart Rhythm</i> , 2014, 11, 1728-1732.	0.3	30
27	Determination of <i>KCNQ1OT1</i> and <i>H19</i> methylation levels in BWS and SRS patients using methylation-sensitive high-resolution melting analysis. <i>European Journal of Human Genetics</i> , 2009, 17, 467-473.	1.4	47
28	Mutations in <i>CCBE1</i> cause generalized lymph vessel dysplasia in humans. <i>Nature Genetics</i> , 2009, 41, 1272-1274.	9.4	269
29	Haplotype-Sharing Analysis Implicates Chromosome 7q36 Harboring <i>DPP6</i> in Familial Idiopathic Ventricular Fibrillation. <i>American Journal of Human Genetics</i> , 2009, 84, 468-476.	2.6	158
30	The 2373insG mutation in the <i>MYBPC3</i> gene is a founder mutation, which accounts for nearly one-fourth of the HCM cases in the Netherlands. <i>European Heart Journal</i> , 2003, 24, 1848-1853.	1.0	127
31	Homozygous Premature Truncation of the <i>HERG</i> Protein. <i>Circulation</i> , 1999, 100, 1264-1267.	1.6	67