

Marije E C Meuwissen

List of Publications by Year in descending order

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Version: 2024-02-01

20
papers

1,020
citations

687363

13
h-index

794594

19
g-index

20
all docs

20
docs citations

20
times ranked

2442
citing authors

#	ARTICLE	IF	CITATIONS
1	Genotype-phenotype correlations in <i>SCN8A</i> -related disorders reveal prognostic and therapeutic implications. <i>Brain</i> , 2022, 145, 2991-3009.	7.6	69
2	Mutations in <i>RNU7-1</i> Weaken Secondary RNA Structure, Induce MCP-1 and CXCL10 in CSF, and Result in Aicardi-Goutières Syndrome with Severe End-Organ Involvement. <i>Journal of Clinical Immunology</i> , 2022, 42, 962-974.	3.8	8
3	Heterozygous variants in <i>CTR9</i> , which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, , .	2.4	1
4	<i>JARID2</i> haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021, 23, 374-383.	2.4	13
5	Intracerebral hemorrhage in a neonate with an intragenic <i>COL4A2</i> duplication. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 571-574.	1.2	4
6	Rare deleterious mutations of <i>HNRNP</i> genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	8.2	50
7	<i>PUM1</i> haploinsufficiency is associated with syndromic neurodevelopmental delay and epilepsy. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 591-594.	1.2	1
8	Sleep-disordered breathing and nocturnal hypoventilation in children with the <i>MECP2</i> duplication syndrome: A case series and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2437-2441.	1.2	6
9	Lessons learned from 40 novel <i>PIGA</i> patients and a review of the literature. <i>Epilepsia</i> , 2020, 61, 1142-1155.	5.1	32
10	Loss of <i>USP18</i> in microglia induces white matter pathology. <i>Acta Neuropathologica Communications</i> , 2019, 7, 106.	5.2	15
11	Aberrant Function of the C-Terminal Tail of <i>HIST1H1E</i> Accelerates Cellular Senescence and Causes Premature Aging. <i>American Journal of Human Genetics</i> , 2019, 105, 493-508.	6.2	48
12	De novo variants in <i>FBXO11</i> cause a syndromic form of intellectual disability with behavioral problems and dysmorphisms. <i>European Journal of Human Genetics</i> , 2019, 27, 738-746.	2.8	32
13	Genetic variants in the <i>KDM6B</i> gene are associated with neurodevelopmental delays and dysmorphic features. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1276-1286.	1.2	38
14	Novel <i>BRPF1</i> mutation in a boy with intellectual disability, coloboma, facial nerve palsy and hypoplasia of the corpus callosum. <i>European Journal of Medical Genetics</i> , 2019, 62, 103691.	1.3	15
15	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in <i>ADNP</i> . <i>Biological Psychiatry</i> , 2019, 85, 287-297.	1.3	108
16	The Belgian MicroArray Prenatal (BEMAPRE) database: A systematic nationwide repository of fetal genomic aberrations. <i>Prenatal Diagnosis</i> , 2018, 38, 1120-1128.	2.3	24
17	Human <i>USP18</i> deficiency underlies type 1 interferonopathy leading to severe pseudo-TORCH syndrome. <i>Journal of Experimental Medicine</i> , 2016, 213, 1163-1174.	8.5	224
18	The expanding phenotype of <i>COL4A1</i> and <i>COL4A2</i> mutations: clinical data on 13 newly identified families and a review of the literature. <i>Genetics in Medicine</i> , 2015, 17, 843-853.	2.4	204

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19	<i>ACTA2</i> mutation with childhood cardiovascular, autonomic and brain anomalies and severe outcome. American Journal of Medical Genetics, Part A, 2013, 161, 1376-1380.	1.2	36
20	Neurological findings in incontinentia pigmenti; a review. European Journal of Medical Genetics, 2012, 55, 323-331.	1.3	92