Claudia A L Ruivenkamp

List of Publications by Year
in descending order

53	1,832	279798	315739
papers	citations	23 h-index	38 g-index

58	58	58	4253
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.7	5
2	De novo variants in ATP2B1 lead to neurodevelopmental delay. American Journal of Human Genetics, 2022, 109, 944-952.	6.2	11
3	Consolidation of the clinical and genetic definition of aSOX4-related neurodevelopmental syndrome. Journal of Medical Genetics, 2022, 59, 1058-1068.	3.2	10
4	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	7.6	8
5	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	1.2	3
6	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	5.3	2
7	Diagnostic Value of a Protocolized In-Depth Evaluation of Pediatric Bone Marrow Failure: A Multi-Center Prospective Cohort Study. Frontiers in Immunology, 2022, 13, 883826.	4.8	4
8	The Phenotypic Continuum of ATP1A3 -Related Disorders. Neurology, 2022, 99, .	1.1	16
9	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
10	Heterozygous variants in SPTBN1 cause intellectual disability and autism. American Journal of Medical Genetics, Part A, 2021, 185, 2037-2045.	1.2	9
11	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
12	KCND2 variants associated with global developmental delay differentially impair Kv4.2 channel gating. Human Molecular Genetics, 2021, 30, 2300-2314.	2.9	12
13	ZTTK syndrome: Clinical and molecular findings ofÂ15 cases and a review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 3740-3753.	1.2	11
14	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
15	A Case Series of Familial ARID1B Variants Illustrating Variable Expression and Suggestions to Update the ACMG Criteria. Genes, 2021, 12, 1275.	2.4	5
16	Two SMARCAD1 Variants Causing Basan Syndrome in a Canadian and a Dutch Family. JID Innovations, 2021, 1, 100022.	2.4	2
17	The end of the laboratory developed test as we know it? Recommendations from a national multidisciplinary taskforce of laboratory specialists on the interpretation of the IVDR and its complications. Clinical Chemistry and Laboratory Medicine, 2021, 59, 491-497.	2.3	27
18	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	3.8	11

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19	Repurposing of Diagnostic Whole Exome Sequencing Data of 1,583 Individuals for Clinical Pharmacogenetics. Clinical Pharmacology and Therapeutics, 2020, 107, 617-627.	4.7	24
20	Frameshift mutations at the C-terminus of HIST1H1E result in a specific DNA hypomethylation signature. Clinical Epigenetics, 2020, 12, 7.	4.1	40
21	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
22	A new gene associated with a Î²-thalassemia phenotype: the observation of variants in SUPT5H. Blood, 2020, 136, 1789-1793.	1.4	13
23	Identification of known and unknown genes associated with mitral valve prolapse using an exome slice methodology. Journal of Medical Genetics, 2020, 57, 843-850.	3.2	22
24	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	6.2	17
25	Dutch genome diagnostic laboratories accelerated and improved variant interpretation and increased accuracy by sharing data. Human Mutation, 2019, 40, 2230-2238.	2.5	32
26	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
27	Adultâ€onset betaâ€thalassaemia intermedia caused by a 5â€Mb somatic clonal segmental deletion in haemopoietic stem cells involving the Î²â€globin locus. British Journal of Haematology, 2019, 186, e165-e170.	2.5	3
28	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
29	Efficacy of Baricitinib in the Treatment of Chilblains Associated With AicardiâGoutiÃ¨res Syndrome, a Type I Interferonopathy. Arthritis and Rheumatology, 2019, 71, 829-831.	5.6	41
30	Response to Gorokhova et al.. Genetics in Medicine, 2019, 21, 2656-2657.	2.4	0
31	A YWHAZ Variant Associated With Cardiofaciocutaneous Syndrome Activates the RAF-ERK Pathway. Frontiers in Physiology, 2019, 10, 388.	2.8	23
32	From diagnostic yield to clinical impact: a pilot study on the implementation of prenatal exome sequencing in routine care. Genetics in Medicine, 2019, 21, 2303-2310.	2.4	41
33	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
34	Putting genome-wide sequencing in neonates into perspective. Genetics in Medicine, 2019, 21, 1074-1082.	2.4	15
35	Further delineation of an entity caused by CREBBP and EP300 mutations but not resembling Rubinsteinâ€“Taybi syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 862-876.	1.2	52
36	Two novel cases expanding the phenotype of SETD2â€related overgrowth syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1212-1215.	1.2	20

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37	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	6.2	59
38	Monosomy 18p is a risk factor for facioscapulohumeral dystrophy. Journal of Medical Genetics, 2018, 55, 469-478.	3.2	11
39	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	3.8	46
40	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
41	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	6.2	72
42	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	2.5	28
43	Genotype-phenotype correlation in ATAD3A deletions: not just of scientific relevance. Brain, 2017, 140, e66-e66.	7.6	24
44	The Prognostic Value of AJCC Staging in Uveal Melanoma Is Enhanced by Adding Chromosome 3 and 8q Status. , 2017, 58, 833.		77
45	DNA Methylation Profiling of Uniparental Disomy Subjects Provides a Map of Parental Epigenetic Bias in the Human Genome. American Journal of Human Genetics, 2016, 99, 555-566.	6.2	66
46	Clinical and molecular characterization of an infant with a tandem duplication and deletion of 19p13. American Journal of Medical Genetics, Part A, 2015, 167, 1884-1889.	1.2	4
47	Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome?. Human Mutation, 2015, 36, 648-655.	2.5	124
48	Hemizygosity forSMCHD1in Facioscapulohumeral Muscular Dystrophy Type 2: Consequences for 18p Deletion Syndrome. Human Mutation, 2015, 36, 679-683.	2.5	32
49	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73
50	Digital PCR Validates 8q Dosage as Prognostic Tool in Uveal Melanoma. PLoS ONE, 2015, 10, e0116371.	2.5	94
51	Copy number variants in patients with short stature. European Journal of Human Genetics, 2014, 22, 602-609.	2.8	60
52	Subtelomeric deletion of chromosome 10p15.3: Clinical findings and molecular cytogenetic characterization. American Journal of Medical Genetics, Part A, 2012, 158A, 2152-2161.	1.2	45
53	The clinical spectrum of complete FBN1 allele deletions. European Journal of Human Genetics, 2011, 19, 247-252.	2.8	65