David A Koolen

List of Publications by Year
in descending order

48 papers	3,012 citations	218677 26 h-index	197818 49 g-index
51 all docs	51 docs citations	51 times ranked	6868 citing authors

#	Article	IF	Citations
1	Imbalanced autophagy causes synaptic deficits in a human model for neurodevelopmental disorders. Autophagy, 2022, 18, 423-442.	9.1	42
2	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. Genetics in Medicine, 2022, 24, 645-653.	2.4	6
3	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
4	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
5	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
6	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
7	Human disease genes website series: An international, open and dynamic library for upâ€toâ€date clinical information. American Journal of Medical Genetics, Part A, 2021, 185, 1039-1046.	1.2	19
8	Quantitative facial phenotyping for Koolen-de Vries and 22q11.2 deletion syndrome. European Journal of Human Genetics, 2021, 29, 1418-1423.	2.8	12
9	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
10	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
11	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	6.2	23
12	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
13	De novo CLTC variants are associated with a variable phenotype from mild to severe intellectual disability, microcephaly, hypoplasia of the corpus callosum, and epilepsy. Genetics in Medicine, 2020, 22, 797-802.	2.4	15
14	De novo variants in MPP5 cause global developmental delay and behavioral changes. Human Molecular Genetics, 2020, 29, 3388-3401.	2.9	5
15	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
16	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
17	Aminoacyl-tRNA synthetase deficiencies in search of common themes. Genetics in Medicine, 2019, 21, 319-330.	2.4	70
18	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	7.6	67

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19	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	2.4	34
20	High Yield of Pathogenic Germline Mutations Causative or Likely Causative of the Cancer Phenotype in Selected Children with Cancer. Clinical Cancer Research, 2018, 24, 1594-1603.	7.0	52
21	Early speech development in Koolen de Vries syndrome limited by oral praxis and hypotonia. European Journal of Human Genetics, 2018, 26, 75-84.	2.8	30
22	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	2.8	32
23	Variants in PUS7 Cause Intellectual Disability with Speech Delay, Microcephaly, Short Stature, and Aggressive Behavior. American Journal of Human Genetics, 2018, 103, 1045-1052.	6.2	89
24	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
25	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. Orphanet Journal of Rare Diseases, 2018, 13, 86.	2.7	29
26	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	1.8	75
27	Neonatal nonepileptic myoclonus is a prominent clinical feature of <scp>KCNQ</scp>2 gainâ€ofâ€function variants R201C and R201H. Epilepsia, 2017, 58, 436-445.	5.1	80
28	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. Human Mutation, 2017, 38, 594-599.	2.5	3
29	SLC13A5is the second gene associated with KohlschÃ¼tterâ€“TÃ¶nz syndrome. Journal of Medical Genetics, 2017, 54, 54-62.	3.2	45
30	The epileptology of Koolenâ€de Vries syndrome: Electroâ€clinicoâ€radiologic findings in 31 patients. Epilepsia, 2017, 58, 1085-1094.	5.1	20
31	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
32	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
33	Computer face-matching technology using two-dimensional photographs accurately matches the facial gestalt of unrelated individuals with the same syndromic form of intellectual disability. BMC Biotechnology, 2017, 17, 90.	3.3	37
34	Mutations in Complex I Assembly Factor TMEM126B Result in Muscle Weakness and Isolated Complex I Deficiency. American Journal of Human Genetics, 2016, 99, 208-216.	6.2	51
35	Biallelic Mutations in TBCD , Encoding the Tubulin Folding Cofactor D, Perturb Microtubule Dynamics and Cause Early-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 962-973.	6.2	66
36	Duplications of SLC1A3: Associated with ADHD and autism. European Journal of Medical Genetics, 2016, 59, 373-376.	1.3	19

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37	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
38	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
39	Variability in dentofacial phenotypes in four families with WNT10A mutations. European Journal of Human Genetics, 2014, 22, 1063-1070.	2.8	34
40	Beta-propeller protein-associated neurodegeneration (BPAN), a rare form of NBIA: Novel mutations and neuropsychiatric phenotype in three adult patients. Parkinsonism and Related Disorders, 2014, 20, 332-336.	2.2	42
41	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	2.5	303
42	Two families with sibling recurrence of the 17q21.31 microdeletion syndrome due to low-grade mosaicism. European Journal of Human Genetics, 2012, 20, 729-733.	2.8	17
43	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	21.4	194
44	Genomic microarrays in mental retardation: A practical workflow for diagnostic applications. Human Mutation, 2009, 30, 283-292.	2.5	136
45	Identification of non-recurrent submicroscopic genome imbalances: the advantage of genome-wide microarrays over targeted approaches. European Journal of Human Genetics, 2008, 16, 395-400.	2.8	14
46	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	21.4	418
47	Holoprosencephaly and preaxial polydactyly associated with a 1.24ÂMb duplication encompassing FBXW11 at 5q35.1. Journal of Human Genetics, 2006, 51, 721-726.	2.3	18
48	Molecular characterisation of patients with subtelomeric 22q abnormalities using chromosome specific array-based comparative genomic hybridisation. European Journal of Human Genetics, 2005, 13, 1019-1024.	2.8	42