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	A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism. Nature Genetics, 2006, 38, 999-1001.	21.4	418
2	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
3	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
4	Mutations in the chromatin modifier gene KANSL1 cause the 17q21.31 microdeletion syndrome. Nature Genetics, 2012, 44, 639-641.	21.4	194
5	Genomic microarrays in mental retardation: A practical workflow for diagnostic applications. Human Mutation, 2009, 30, 283-292.	2.5	136
6	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
7	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
8	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
9	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
10	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
11	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
12	Aminoacyl-tRNA synthetase deficiencies in search of common themes. Genetics in Medicine, 2019, 21, 319-330.	2.4	70
13	PLPHP deficiency: clinical, genetic, biochemical, and mechanistic insights. Brain, 2019, 142, 542-559.	7.6	67
14	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
15	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
16	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
17	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
18	SLC13A5is the second gene associated with KohlschÃ¼tterâ€“TÃ¶nz syndrome. Journal of Medical Genetics, 2017, 54, 54-62.	3.2	45

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19	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
20	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
21	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
22	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
23	Imbalanced autophagy causes synaptic deficits in a human model for neurodevelopmental disorders. Autophagy, 2022, 18, 423-442.	9.1	42
24	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
25	Loss-of-function mutations in UDP-Glucose 6-Dehydrogenase cause recessive developmental epileptic encephalopathy. Nature Communications, 2020, 11, 595.	12.8	35
26	Variability in dentofacial phenotypes in four families with WNT10A mutations. European Journal of Human Genetics, 2014, 22, 1063-1070.	2.8	34
27	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	2.4	34
28	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
29	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
30	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
31	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
32	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
33	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
34	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
35	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
36	The epileptology of Koolenâ€de Vries syndrome: Electroâ€clinicoâ€radiologic findings in 31 patients. Epilepsia, 2017, 58, 1085-1094.	5.1	20

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37	Duplications of SLC1A3: Associated with ADHD and autism. European Journal of Medical Genetics, 2016, 59, 373-376.	1.3	19
38	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
39	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
40	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
41	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
42	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
43	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
44	Quantitative facial phenotyping for Koolen-de Vries and 22q11.2 deletion syndrome. European Journal of Human Genetics, 2021, 29, 1418-1423.	2.8	12
45	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. Genetics in Medicine, 2022, 24, 645-653.	2.4	6
46	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
47	De novo variants in MPP5 cause global developmental delay and behavioral changes. Human Molecular Genetics, 2020, 29, 3388-3401.	2.9	5
48	Quantification of Phenotype Information Aids the Identification of Novel Disease Genes. Human Mutation, 2017, 38, 594-599.	2.5	3