

Tjitske Kleefstra

List of Publications by Year in descending order

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Version: 2024-02-01

100
papers

8,293
citations

76196

40
h-index

54797

84
g-index

108
all docs

108
docs citations

108
times ranked

13694
citing authors

#	ARTICLE	IF	CITATIONS
1	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. <i>New England Journal of Medicine</i> , 2012, 367, 1921-1929.	13.9	1,367
2	Genome sequencing identifies major causes of severe intellectual disability. <i>Nature</i> , 2014, 511, 344-347.	13.7	996
3	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. <i>Nature Neuroscience</i> , 2016, 19, 1194-1196.	7.1	407
4	Loss-of-Function Mutations in Euchromatin Histone Methyl Transferase 1 (EHMT1) Cause the 9q34 Subtelomeric Deletion Syndrome. <i>American Journal of Human Genetics</i> , 2006, 79, 370-377.	2.6	343
5	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. <i>Nature Genetics</i> , 2014, 46, 380-384.	9.4	293
6	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. <i>American Journal of Human Genetics</i> , 2016, 98, 149-164.	2.6	270
7	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	2.6	230
8	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. <i>Genetics in Medicine</i> , 2017, 19, 1055-1063.	1.1	220
9	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. <i>American Journal of Human Genetics</i> , 2012, 91, 73-82.	2.6	214
10	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 356-370.	2.6	171
11	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	2.6	136
12	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2016, 98, 541-552.	2.6	132
13	De novo gain-of-function and loss-of-function mutations of <i>SCN8A</i> in patients with intellectual disabilities and epilepsy. <i>Journal of Medical Genetics</i> , 2015, 52, 330-337.	1.5	124
14	BCL11A Haploinsufficiency Causes an Intellectual Disability Syndrome and Dysregulates Transcription. <i>American Journal of Human Genetics</i> , 2016, 99, 253-274.	2.6	118
15	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. <i>PLoS Genetics</i> , 2017, 13, e1006864.	1.5	116
16	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. <i>Biological Psychiatry</i> , 2019, 85, 287-297.	0.7	108
17	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	2.6	101
18	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. <i>American Journal of Human Genetics</i> , 2016, 98, 373-381.	2.6	95

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19	Neuronal network dysfunction in a model for Kleefstra syndrome mediated by enhanced NMDAR signaling. <i>Nature Communications</i> , 2019, 10, 4928.	5.8	92
20	Involvement of the kinesin family members <i>KIF4A</i> and <i>KIF5C</i> in intellectual disability and synaptic function. <i>Journal of Medical Genetics</i> , 2014, 51, 487-494.	1.5	90
21	Identification of rare de novo epigenetic variations in congenital disorders. <i>Nature Communications</i> , 2018, 9, 2064.	5.8	82
22	The genetics of cognitive epigenetics. <i>Neuropharmacology</i> , 2014, 80, 83-94.	2.0	78
23	Histone Methylation by the Kleefstra Syndrome Protein EHMT1 Mediates Homeostatic Synaptic Scaling. <i>Neuron</i> , 2016, 91, 341-355.	3.8	74
24	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. <i>American Journal of Human Genetics</i> , 2015, 97, 493-500.	2.6	71
25	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	5.8	70
26	Phenotypes and genotypes in individuals with <i>SMC1A</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2108-2125.	0.7	69
27	Further delineation of the KBC syndrome phenotype caused by ANKRD11 aberrations. <i>European Journal of Human Genetics</i> , 2015, 23, 1176-1185.	1.4	67
28	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. <i>Nature Genetics</i> , 2016, 48, 877-887.	9.4	67
29	Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability. <i>Human Genetics</i> , 2017, 136, 821-834.	1.8	66
30	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 716-724.	2.6	66
31	Reduced Euchromatin histone methyltransferase 1 causes developmental delay, hypotonia, and cranial abnormalities associated with increased bone gene expression in Kleefstra syndrome mice. <i>Developmental Biology</i> , 2014, 386, 395-407.	0.9	65
32	Human neuronal networks on micro-electrode arrays are a highly robust tool to study disease-specific genotype-phenotype correlations in vitro. <i>Stem Cell Reports</i> , 2021, 16, 2182-2196.	2.3	63
33	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. <i>American Journal of Human Genetics</i> , 2018, 102, 985-994.	2.6	59
34	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	2.6	59
35	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. <i>Genetics in Medicine</i> , 2016, 18, 1158-1162.	1.1	58
36	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 650-658.	2.6	56

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37	A detailed clinical analysis of 13 patients with AUTS2 syndrome further delineates the phenotypic spectrum and underscores the behavioural phenotype. <i>Journal of Medical Genetics</i> , 2016, 53, 523-532.	1.5	51
38	The molecular and phenotypic spectrum of <i>IQSEC2</i> -related epilepsy. <i>Epilepsia</i> , 2016, 57, 1858-1869.	2.6	46
39	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. <i>Human Genetics</i> , 2018, 137, 375-388.	1.8	46
40	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. <i>Genetics in Medicine</i> , 2015, 17, 460-466.	1.1	45
41	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. <i>American Journal of Human Genetics</i> , 2017, 101, 139-148.	2.6	45
42	Distinct Pathogenic Genes Causing Intellectual Disability and Autism Exhibit a Common Neuronal Network Hyperactivity Phenotype. <i>Cell Reports</i> , 2020, 30, 173-186.e6.	2.9	44
43	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. <i>Nature Communications</i> , 2019, 10, 4679.	5.8	43
44	Germline AGO2 mutations impair RNA interference and human neurological development. <i>Nature Communications</i> , 2020, 11, 5797.	5.8	43
45	Characterization of SETD1A haploinsufficiency in humans and <i>Drosophila</i> defines a novel neurodevelopmental syndrome. <i>Molecular Psychiatry</i> , 2021, 26, 2013-2024.	4.1	43
46	Mitochondrial dysfunction and organic aciduria in five patients carrying mutations in the Ras-MAPK pathway. <i>European Journal of Human Genetics</i> , 2011, 19, 138-144.	1.4	42
47	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor β Signaling. <i>Biological Psychiatry</i> , 2020, 87, 100-112.	0.7	42
48	Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. <i>American Journal of Human Genetics</i> , 2019, 105, 395-402.	2.6	39
49	Computer face-matching technology using two-dimensional photographs accurately matches the facial gestalt of unrelated individuals with the same syndromic form of intellectual disability. <i>BMC Biotechnology</i> , 2017, 17, 90.	1.7	37
50	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. <i>American Journal of Human Genetics</i> , 2020, 107, 164-172.	2.6	37
51	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. <i>American Journal of Human Genetics</i> , 2017, 101, 824-832.	2.6	36
52	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 403-412.	2.6	35
53	Variability in dentofacial phenotypes in four families with WNT10A mutations. <i>European Journal of Human Genetics</i> , 2014, 22, 1063-1070.	1.4	34
54	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in <i>Drosophila</i> . <i>European Journal of Human Genetics</i> , 2016, 24, 1145-1153.	1.4	34

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55	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2019, 104, 758-766.	2.6	34
56	Increased H3K9 methylation and impaired expression of Protocadherins are associated with the cognitive dysfunctions of the Kleefstra syndrome. <i>Nucleic Acids Research</i> , 2018, 46, 4950-4965.	6.5	32
57	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. <i>European Journal of Human Genetics</i> , 2018, 26, 54-63.	1.4	32
58	Adaptive and maladaptive functioning in Kleefstra syndrome compared to other rare genetic disorders with intellectual disabilities. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1821-1830.	0.7	31
59	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. <i>American Journal of Human Genetics</i> , 2021, 108, 1053-1068.	2.6	31
60	A 3-base pair deletion, c.9711_9713del, in DMD results in intellectual disability without muscular dystrophy. <i>European Journal of Human Genetics</i> , 2014, 22, 480-485.	1.4	30
61	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	2.6	30
62	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGF β 2 signaling and cause autosomal dominant spondylocarpotarsal synostosis. <i>Scientific Reports</i> , 2017, 7, 41803.	1.6	29
63	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. <i>American Journal of Human Genetics</i> , 2015, 96, 386-396.	2.6	27
64	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. <i>Gene</i> , 2017, 605, 92-98.	1.0	26
65	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. <i>Human Mutation</i> , 2021, 42, 445-459.	1.1	26
66	Loss-of-function variants in the schizophrenia risk gene SETD1A alter neuronal network activity in human neurons through the cAMP/PKA pathway. <i>Cell Reports</i> , 2022, 39, 110790.	2.9	26
67	Haploinsufficiency of EHMT1 improves pattern separation and increases hippocampal cell proliferation. <i>Scientific Reports</i> , 2017, 7, 40284.	1.6	25
68	The intellectual disability-associated CAMK2G p.Arg292Pro mutation acts as a pathogenic gain-of-function. <i>Human Mutation</i> , 2018, 39, 2008-2024.	1.1	25
69	Mutations in TBR1 gene leads to cortical malformations and intellectual disability. <i>European Journal of Medical Genetics</i> , 2018, 61, 759-764.	0.7	24
70	Damaging de novo missense variants in <i>EEF1A2</i> lead to a developmental and degenerative epileptic dyskinetic encephalopathy. <i>Human Mutation</i> , 2020, 41, 1263-1279.	1.1	24
71	The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. <i>Science Advances</i> , 2021, 7, .	4.7	24
72	Phenotypic spectrum of 20 novel patients with molecularly defined supernumerary marker chromosomes 15 and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2221-2229.	0.7	23

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73	Pathogenic Variants in GPC4 Cause Keipert Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 914-924.	2.6	23
74	Behavioral phenotype in the 9q subtelomeric deletion syndrome: A report about two adult patients. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010, 153B, 536-541.	1.1	22
75	Interstitial 2.2 Mb deletion at 9q34 in a patient with mental retardation but without classical features of the 9q subtelomeric deletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 618-623.	0.7	21
76	The histone methyltransferase G9a regulates tolerance to oxidative stress-induced energy consumption. <i>PLoS Biology</i> , 2019, 17, e2006146.	2.6	21
77	The phenotypic spectrum of germline <i>YARS2</i> variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2. <i>Haematologica</i> , 2018, 103, 2008-2015.	1.7	19
78	Missense variant contribution to USP9X-female syndrome. <i>Npj Genomic Medicine</i> , 2020, 5, 53.	1.7	17
79	Heterozygous variants that disturb the transcriptional repressor activity of FOXP4 cause a developmental disorder with speech/language delays and multiple congenital abnormalities. <i>Genetics in Medicine</i> , 2021, 23, 534-542.	1.1	17
80	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. <i>European Journal of Human Genetics</i> , 2021, 29, 625-636.	1.4	17
81	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021, 108, 1138-1150.	2.6	17
82	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019, 10, 611.	1.1	14
83	SETD1A Mediated H3K4 Methylation and Its Role in Neurodevelopmental and Neuropsychiatric Disorders. <i>Frontiers in Molecular Neuroscience</i> , 2021, 14, 772000.	1.4	14
84	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 880-893.	1.1	14
85	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies. <i>Genome Medicine</i> , 2017, 9, 118.	3.6	13
86	Absence epilepsy and the CHD2 gene: an adolescent male with moderate intellectual disability, short-lasting psychoses, and an interstitial deletion in 15q26.1–q26.2. <i>Neuropsychiatric Disease and Treatment</i> , 2016, 12, 1135.	1.0	12
87	Exploring the behavioral and cognitive phenotype of KBG syndrome. <i>Genes, Brain and Behavior</i> , 2019, 18, e12553.	1.1	12
88	Clinical delineation of SETBP1 haploinsufficiency disorder. <i>European Journal of Human Genetics</i> , 2021, 29, 1198-1205.	1.4	12
89	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. <i>Gene</i> , 2014, 538, 30-35.	1.0	11
90	Haploinsufficiency of the HIRA gene located in the 22q11 deletion syndrome region is associated with abnormal neurodevelopment and impaired dendritic outgrowth. <i>Human Genetics</i> , 2021, 140, 885-896.	1.8	10

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91	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	1.1	9
92	A novel MBD5 mutation in an intellectually disabled adult female patient with epilepsy: Suggestive of early onset dementia?. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e849.	0.6	8
93	Human <i>KCNQ5</i> de novo mutations underlie epilepsy and intellectual disability. <i>Journal of Neurophysiology</i> , 2022, 128, 40-61.	0.9	8
94	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. <i>European Journal of Human Genetics</i> , 2021, 29, 1359-1368.	1.4	7
95	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. <i>Genetics in Medicine</i> , 2022, 24, 645-653.	1.1	6
96	Speech&language profiles in the context of cognitive and adaptive functioning in SATB2 associated syndrome. <i>Genes, Brain and Behavior</i> , 2021, 20, e12761.	1.1	4
97	Following Excitation/Inhibition Ratio Homeostasis from Synapse to EEG in Monogenetic Neurodevelopmental Disorders. <i>Genes</i> , 2022, 13, 390.	1.0	4
98	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. <i>European Journal of Medical Genetics</i> , 2022, 65, 104402.	0.7	2
99	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, , .	1.1	1
100	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. <i>Genetical Research</i> , 2015, 97, e19.	0.3	0