Tjitske Kleefstra

List of Publications by Year
in descending order

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100 papers	8,293 citations	76326 40 h-index	54911 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. New England Journal of Medicine, 2012, 367, 1921-1929.	27.0	1,367
2	Genome sequencing identifies major causes of severe intellectual disability. Nature, 2014, 511, 344-347.	27.8	996
3	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	14.8	407
4	Loss-of-Function Mutations in Euchromatin Histone Methyl Transferase 1 (EHMT1) Cause the 9q34 Subtelomeric Deletion Syndrome. American Journal of Human Genetics, 2006, 79, 370-377.	6.2	343
5	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. Nature Genetics, 2014, 46, 380-384.	21.4	293
6	Systematic Phenomics Analysis Deconvolutes Genes Mutated in Intellectual Disability into Biologically Coherent Modules. American Journal of Human Genetics, 2016, 98, 149-164.	6.2	270
7	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
8	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	2.4	220
9	Disruption of an EHMT1-Associated Chromatin-Modification Module Causes Intellectual Disability. American Journal of Human Genetics, 2012, 91, 73-82.	6.2	214
10	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	6.2	171
11	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
12	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
13	De novo gain-of-function and loss-of-function mutations ofSCN8Ain patients with intellectual disabilities and epilepsy. Journal of Medical Genetics, 2015, 52, 330-337.	3.2	124
14	BCL11A Haploinsufficiency Causes an Intellectual Disability Syndrome and Dysregulates Transcription. American Journal of Human Genetics, 2016, 99, 253-274.	6.2	118
15	Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder. PLoS Genetics, 2017, 13, e1006864.	3.5	116
16	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
17	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
18	De Novo Loss-of-Function Mutations in USP9X Cause a Female-Specific Recognizable Syndrome with Developmental Delay and Congenital Malformations. American Journal of Human Genetics, 2016, 98, 373-381.	6.2	95

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19	Neuronal network dysfunction in a model for Kleefstra syndrome mediated by enhanced NMDAR signaling. Nature Communications, 2019, 10, 4928.	12.8	92
20	Involvement of the kinesin family membersKIF4AandKIF5Cin intellectual disability and synaptic function. Journal of Medical Genetics, 2014, 51, 487-494.	3.2	90
21	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	12.8	82
22	The genetics of cognitive epigenetics. Neuropharmacology, 2014, 80, 83-94.	4.1	78
23	Histone Methylation by the Kleefstra Syndrome Protein EHMT1 Mediates Homeostatic Synaptic Scaling. Neuron, 2016, 91, 341-355.	8.1	74
24	De Novo Mutations in CHAMP1 Cause Intellectual Disability with Severe Speech Impairment. American Journal of Human Genetics, 2015, 97, 493-500.	6.2	71
25	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
26	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
27	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. European Journal of Human Genetics, 2015, 23, 1176-1185.	2.8	67
28	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor SIN3A causes mild intellectual disability by affecting the development of cortical integrity. Nature Genetics, 2016, 48, 877-887.	21.4	67
29	Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability. Human Genetics, 2017, 136, 821-834.	3.8	66
30	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	6.2	66
31	Reduced Euchromatin histone methyltransferase 1 causes developmental delay, hypotonia, and cranial abnormalities associated with increased bone gene expression in Kleefstra syndrome mice. Developmental Biology, 2014, 386, 395-407.	2.0	65
32	Human neuronal networks on micro-electrode arrays are a highly robust tool to study disease-specific genotype-phenotype correlations inÂvitro. Stem Cell Reports, 2021, 16, 2182-2196.	4.8	63
33	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
34	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
35	Novel mutations in LRP6 highlight the role of WNT signaling in tooth agenesis. Genetics in Medicine, 2016, 18, 1158-1162.	2.4	58
36	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	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. Journal of Medical Genetics, 2016, 53, 523-532.	3.2	51
38	The molecular and phenotypic spectrum of <scp>IQSEC</scp>2â€related epilepsy. Epilepsia, 2016, 57, 1858-1869.	5.1	46
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	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
41	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. American Journal of Human Genetics, 2017, 101, 139-148.	6.2	45
42	Distinct Pathogenic Genes Causing Intellectual Disability and Autism Exhibit a Common Neuronal Network Hyperactivity Phenotype. Cell Reports, 2020, 30, 173-186.e6.	6.4	44
43	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
44	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	12.8	43
45	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
46	Mitochondrial dysfunction and organic aciduria in five patients carrying mutations in the Ras-MAPK pathway. European Journal of Human Genetics, 2011, 19, 138-144.	2.8	42
47	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
48	Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. American Journal of Human Genetics, 2019, 105, 395-402.	6.2	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. BMC Biotechnology, 2017, 17, 90.	3.3	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. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
51	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype. American Journal of Human Genetics, 2017, 101, 824-832.	6.2	36
52	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35
53	Variability in dentofacial phenotypes in four families with WNT10A mutations. European Journal of Human Genetics, 2014, 22, 1063-1070.	2.8	34
54	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. European Journal of Human Genetics, 2016, 24, 1145-1153.	2.8	34

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55	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	6.2	34
56	Increased H3K9 methylation and impaired expression of Protocadherins are associated with the cognitive dysfunctions of the Kleefstra syndrome. Nucleic Acids Research, 2018, 46, 4950-4965.	14.5	32
57	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
58	Adaptive and maladaptive functioning in Kleefstra syndrome compared to other rare genetic disorders with intellectual disabilities. American Journal of Medical Genetics, Part A, 2017, 173, 1821-1830.	1.2	31
59	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
60	A 3-base pair deletion, c.9711_9713del, in DMD results in intellectual disability without muscular dystrophy. European Journal of Human Genetics, 2014, 22, 480-485.	2.8	30
61	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
62	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGFÎ² signaling and cause autosomal dominant spondylocarpotarsal synostosis. Scientific Reports, 2017, 7, 41803.	3.3	29
63	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	6.2	27
64	Mutations in two large pedigrees highlight the role of ZNF711 in X-linked intellectual disability. Gene, 2017, 605, 92-98.	2.2	26
65	TAOK1 is associated with neurodevelopmental disorder and essential for neuronal maturation and cortical development. Human Mutation, 2021, 42, 445-459.	2.5	26
66	Loss-of-function variants in the schizophrenia risk gene SETD1A alter neuronal network activity in human neurons through the cAMP/PKA pathway. Cell Reports, 2022, 39, 110790.	6.4	26
67	Haploinsufficiency of EHMT1 improves pattern separation and increases hippocampal cell proliferation. Scientific Reports, 2017, 7, 40284.	3.3	25
68	The intellectual disability-associated CAMK2G p.Arg292Pro mutation acts as a pathogenic gain-of-function. Human Mutation, 2018, 39, 2008-2024.	2.5	25
69	Mutations in TBR1 gene leads to cortical malformations and intellectual disability. European Journal of Medical Genetics, 2018, 61, 759-764.	1.3	24
70	Damaging de novo missense variants inEEF1A2lead to a developmental and degenerative epilepticâ€dyskinetic encephalopathy. Human Mutation, 2020, 41, 1263-1279.	2.5	24
71	The CHD8/CHD7/Kismet family links blood-brain barrier glia and serotonin to ASD-associated sleep defects. Science Advances, 2021, 7, .	10.3	24
72	Phenotypic spectrum of 20 novel patients with molecularly defined supernumerary marker chromosomes 15 and a review of the literature. American Journal of Medical Genetics, Part A, 2010, 152A, 2221-2229.	1.2	23

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73	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	6.2	23
74	Behavioral phenotype in the 9q subtelomeric deletion syndrome: A report about two adult patients. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 536-541.	1.7	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. American Journal of Medical Genetics, Part A, 2006, 140A, 618-623.	1.2	21
76	The histone methyltransferase G9a regulates tolerance to oxidative stressâ€“induced energy consumption. PLoS Biology, 2019, 17, e2006146.	5.6	21
77	The phenotypic spectrum of germline YARS2 variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2. Haematologica, 2018, 103, 2008-2015.	3.5	19
78	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	3.8	17
79	Heterozygous variants that disturb the transcriptional repressor activity of FOXP4 cause a developmental disorder with speech/language delays and multiple congenital abnormalities. Genetics in Medicine, 2021, 23, 534-542.	2.4	17
80	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
81	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
82	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	2.3	14
83	SETD1A Mediated H3K4 Methylation and Its Role in Neurodevelopmental and Neuropsychiatric Disorders. Frontiers in Molecular Neuroscience, 2021, 14, 772000.	2.9	14
84	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 880-893.	2.4	14
85	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotypeâ€“phenotype associations in the muscular dystrophy-dystroglycanopathies. Genome Medicine, 2017, 9, 118.	8.2	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. Neuropsychiatric Disease and Treatment, 2016, 12, 1135.	2.2	12
87	Exploring the behavioral and cognitive phenotype of KBG syndrome. Genes, Brain and Behavior, 2019, 18, e12553.	2.2	12
88	Clinical delineation of SETBP1 haploinsufficiency disorder. European Journal of Human Genetics, 2021, 29, 1198-1205.	2.8	12
89	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. Gene, 2014, 538, 30-35.	2.2	11
90	Haploinsufficiency of the HIRA gene located in the 22q11 deletion syndrome region is associated with abnormal neurodevelopment and impaired dendritic outgrowth. Human Genetics, 2021, 140, 885-896.	3.8	10

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91	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
92	A novel MBD5 mutation in an intellectually disabled adult female patient with epilepsy: Suggestive of early onset dementia?. Molecular Genetics & Genomic Medicine, 2019, 7, e849.	1.2	8
93	Human KCNQ5 de novo mutations underlie epilepsy and intellectual disability. Journal of Neurophysiology, 2022, 128, 40-61.	1.8	8
94	A MT-TL1 variant identified by whole exome sequencing in an individual with intellectual disability, epilepsy, and spastic tetraparesis. European Journal of Human Genetics, 2021, 29, 1359-1368.	2.8	7
95	Phenotype based prediction of exome sequencing outcome using machine learning for neurodevelopmental disorders. Genetics in Medicine, 2022, 24, 645-653.	2.4	6
96	Speechâ€language profiles in the context of cognitive and adaptive functioning in SATB2 â€associated syndrome. Genes, Brain and Behavior, 2021, 20, e12761.	2.2	4
97	Following Excitation/Inhibition Ratio Homeostasis from Synapse to EEG in Monogenetic Neurodevelopmental Disorders. Genes, 2022, 13, 390.	2.4	4
98	Genome-wide variant calling in reanalysis of exome sequencing data uncovered a pathogenic TUBB3 variant. European Journal of Medical Genetics, 2022, 65, 104402.	1.3	2
99	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	2.4	1
100	A de novo microdeletion in NRXN1 in a Dutch patient with mild intellectual disability, microcephaly and gonadal dysgenesis. Genetical Research, 2015, 97, e19.	0.9	0