

# Tjitske Kleefstra

## List of Publications by Year in descending order

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Version: 2024-02-01

100  
papers

8,293  
citations

87401

40  
h-index

62345

84  
g-index

108  
all docs

108  
docs citations

108  
times ranked

14766  
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	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
3	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, 24, 880-893.	1.1	14
4	Following Excitation/Inhibition Ratio Homeostasis from Synapse to EEG in Monogenetic Neurodevelopmental Disorders. <i>Genes</i> , 2022, 13, 390.	1.0	4
5	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	1.1	9
6	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
7	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
8	Human <i>KCNQ5</i> de novo mutations underlie epilepsy and intellectual disability. <i>Journal of Neurophysiology</i> , 2022, 128, 40-61.	0.9	8
9	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
10	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
11	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
12	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
13	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
14	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
15	Clinical delineation of SETBP1 haploinsufficiency disorder. <i>European Journal of Human Genetics</i> , 2021, 29, 1198-1205.	1.4	12
16	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
17	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
18	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

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19	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
20	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
21	Speech and 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
22	SETD1A Mediated H3K4 Methylation and Its Role in Neurodevelopmental and Neuropsychiatric Disorders. <i>Frontiers in Molecular Neuroscience</i> , 2021, 14, 772000.	1.4	14
23	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor $\beta$ Signaling. <i>Biological Psychiatry</i> , 2020, 87, 100-112.	0.7	42
24	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
25	Germline AGO2 mutations impair RNA interference and human neurological development. <i>Nature Communications</i> , 2020, 11, 5797.	5.8	43
26	Missense variant contribution to USP9X-female syndrome. <i>Npj Genomic Medicine</i> , 2020, 5, 53.	1.7	17
27	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
28	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
29	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
30	A novel MBD5 mutation in an intellectually disabled adult female patient with epilepsy: Suggestive of early onset dementia?. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e849.	0.6	8
31	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
32	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
33	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. <i>Nature Communications</i> , 2019, 10, 4679.	5.8	43
34	Neuronal network dysfunction in a model for Kleefstra syndrome mediated by enhanced NMDAR signaling. <i>Nature Communications</i> , 2019, 10, 4928.	5.8	92
35	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. <i>Frontiers in Genetics</i> , 2019, 10, 611.	1.1	14
36	The histone methyltransferase G9a regulates tolerance to oxidative stress-induced energy consumption. <i>PLoS Biology</i> , 2019, 17, e2006146.	2.6	21

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37	Pathogenic Variants in GPC4 Cause Keipert Syndrome. American Journal of Human Genetics, 2019, 104, 914-924.	2.6	23
38	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.	2.6	34
39	Exploring the behavioral and cognitive phenotype of KBG syndrome. Genes, Brain and Behavior, 2019, 18, e12553.	1.1	12
40	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.	2.6	59
41	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
42	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.	2.6	59
43	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.	6.5	32
44	A genotype-first approach identifies an intellectual disability-overweight syndrome caused by PHIP haploinsufficiency. European Journal of Human Genetics, 2018, 26, 54-63.	1.4	32
45	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	5.8	70
46	Mutations in TBR1 gene leads to cortical malformations and intellectual disability. European Journal of Medical Genetics, 2018, 61, 759-764.	0.7	24
47	The intellectual disability-associated CAMK2G p.Arg292Pro mutation acts as a pathogenic gain-of-function. Human Mutation, 2018, 39, 2008-2024.	1.1	25
48	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	5.8	82
49	The phenotypic spectrum of germline <i>YARS2</i> variants: from isolated sideroblastic anemia to mitochondrial myopathy, lactic acidosis and sideroblastic anemia 2. Haematologica, 2018, 103, 2008-2015.	1.7	19
50	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	1.8	46
51	Haploinsufficiency of EHMT1 improves pattern separation and increases hippocampal cell proliferation. Scientific Reports, 2017, 7, 40284.	1.6	25
52	A postnatal role for embryonic myosin revealed by MYH3 mutations that alter TGF $\beta$ 2 signaling and cause autosomal dominant spondylarcarpotarsal synostosis. Scientific Reports, 2017, 7, 41803.	1.6	29
53	Heterozygous HNRNPU variants cause early onset epilepsy and severe intellectual disability. Human Genetics, 2017, 136, 821-834.	1.8	66
54	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.	0.7	31

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55	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
56	De Novo Truncating Mutations in the Last and Penultimate Exons of <i>PPM1D</i> Cause an Intellectual Disability Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 650-658.	2.6	56
57	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
58	Mutations in two large pedigrees highlight the role of <i>ZNF711</i> in X-linked intellectual disability. <i>Gene</i> , 2017, 605, 92-98.	1.0	26
59	Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of <i>RAB11B</i> Cause Intellectual Disability and a Distinctive Brain Phenotype. <i>American Journal of Human Genetics</i> , 2017, 101, 824-832.	2.6	36
60	De Novo Missense Mutations in <i>DHX30</i> Impair Global Translation and Cause a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 716-724.	2.6	66
61	De Novo Mutations in Protein Kinase Genes <i>CAMK2A</i> and <i>CAMK2B</i> Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	2.6	136
62	<i>WDR26</i> 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
63	Functional convergence of histone methyltransferases <i>EHMT1</i> and <i>KMT2C</i> involved in intellectual disability and autism spectrum disorder. <i>PLoS Genetics</i> , 2017, 13, e1006864.	1.5	116
64	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
65	<i>B3GALNT2</i> 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
66	Absence epilepsy and the <i>CHD2</i> gene: an adolescent male with moderate intellectual disability, short-lasting psychoses, and an interstitial deletion in 15q26.1&ndash;q26.2. <i>Neuropsychiatric Disease and Treatment</i> , 2016, 12, 1135.	1.0	12
67	Haploinsufficiency of MeCP2-interacting transcriptional co-repressor <i>SIN3A</i> causes mild intellectual disability by affecting the development of cortical integrity. <i>Nature Genetics</i> , 2016, 48, 877-887.	9.4	67
68	Histone Methylation by the Kleefstra Syndrome Protein <i>EHMT1</i> Mediates Homeostatic Synaptic Scaling. <i>Neuron</i> , 2016, 91, 341-355.	3.8	74
69	A detailed clinical analysis of 13 patients with <i>AUTS2</i> syndrome further delineates the phenotypic spectrum and underscores the behavioural phenotype. <i>Journal of Medical Genetics</i> , 2016, 53, 523-532.	1.5	51
70	The molecular and phenotypic spectrum of <i>IQSEC2</i> -related epilepsy. <i>Epilepsia</i> , 2016, 57, 1858-1869.	2.6	46
71	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
72	<i>BCL11A</i> Haploinsufficiency Causes an Intellectual Disability Syndrome and Dysregulates Transcription. <i>American Journal of Human Genetics</i> , 2016, 99, 253-274.	2.6	118

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73	De novo loss-of-function mutations in WAC cause a recognizable intellectual disability syndrome and learning deficits in Drosophila. <i>European Journal of Human Genetics</i> , 2016, 24, 1145-1153.	1.4	34
74	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
75	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
76	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
77	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
78	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
79	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
80	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
81	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
82	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
83	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
84	Further delineation of the KBG syndrome phenotype caused by ANKRD11 aberrations. <i>European Journal of Human Genetics</i> , 2015, 23, 1176-1185.	1.4	67
85	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
86	Variability in dentofacial phenotypes in four families with WNT10A mutations. <i>European Journal of Human Genetics</i> , 2014, 22, 1063-1070.	1.4	34
87	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
88	A SWI/SNF-related autism syndrome caused by de novo mutations in ADNP. <i>Nature Genetics</i> , 2014, 46, 380-384.	9.4	293
89	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
90	The genetics of cognitive epigenetics. <i>Neuropharmacology</i> , 2014, 80, 83-94.	2.0	78

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91	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
92	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
93	Genome sequencing identifies major causes of severe intellectual disability. <i>Nature</i> , 2014, 511, 344-347.	13.7	996
94	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. <i>New England Journal of Medicine</i> , 2012, 367, 1921-1929.	13.9	1,367
95	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
96	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
97	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
98	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
99	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
100	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