

# Erik-jan Kamsteeg

## List of Publications by Year in descending order

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84  
papers

3,908  
citations

186265

28  
h-index

144013

57  
g-index

85  
all docs

85  
docs citations

85  
times ranked

8130  
citing authors

#	ARTICLE	IF	CITATIONS
1	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. <i>Nature Neuroscience</i> , 2016, 19, 1194-1196.	14.8	407
2	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	6.2	337
3	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. <i>Genetics in Medicine</i> , 2017, 19, 1055-1063.	2.4	220
4	Short-chain ubiquitination mediates the regulated endocytosis of the aquaporin-2 water channel. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2006, 103, 18344-18349.	7.1	214
5	Mutations in the histone methyltransferase gene <i>KMT2B</i> cause complex early-onset dystonia. <i>Nature Genetics</i> , 2017, 49, 223-237.	21.4	186
6	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. <i>Genetics in Medicine</i> , 2017, 19, 667-675.	2.4	143
7	Best practice guidelines and recommendations on the molecular diagnosis of myotonic dystrophy types 1 and 2. <i>European Journal of Human Genetics</i> , 2012, 20, 1203-1208.	2.8	129
8	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.	3.2	124
9	Mutations in <i>VPS13D</i> lead to a new recessive ataxia with spasticity and mitochondrial defects. <i>Annals of Neurology</i> , 2018, 83, 1075-1088.	5.3	122
10	Loss-of-function mutations in <i>SCN4A</i> cause severe foetal hypokinesia or "classical" congenital myopathy. <i>Brain</i> , 2016, 139, 674-691.	7.6	100
11	De Novo Mutations in <i>PDE10A</i> Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. <i>American Journal of Human Genetics</i> , 2016, 98, 763-771.	6.2	96
12	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , 2018, 83, 1105-1124.	5.3	93
13	Clinical exome sequencing for cerebellar ataxia and spastic paraplegia uncovers novel gene-disease associations and unanticipated rare disorders. <i>European Journal of Human Genetics</i> , 2016, 24, 1460-1466.	2.8	89
14	De Novo Pathogenic Variants in <i>CACNA1E</i> Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	6.2	87
15	Loss of tubulin deglutamylase <i>CCP1</i> causes infantile-onset neurodegeneration. <i>EMBO Journal</i> , 2018, 37, .	7.8	86
16	Mutations in <i>ATP6V1E1</i> or <i>ATP6V1A</i> Cause Autosomal-Recessive Cutis Laxa. <i>American Journal of Human Genetics</i> , 2017, 100, 216-227.	6.2	82
17	Understanding the Psychosocial Effects of WES Test Results on Parents of Children with Rare Diseases. <i>Journal of Genetic Counseling</i> , 2016, 25, 1207-1214.	1.6	73
18	Loss-of-Function Variants in <i>HOPS</i> Complex Genes <i>VPS16</i> and <i>VPS41</i> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. <i>Annals of Neurology</i> , 2020, 88, 867-877.	5.3	70

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19	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. <i>Epilepsia</i> , 2019, 60, 155-164.	5.1	65
20	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. <i>European Journal of Human Genetics</i> , 2020, 28, 40-49.	2.8	65
21	Autosomal Recessive Cerebellar Ataxia Type 3 Due to <i>ANO10</i> Mutations. <i>JAMA Neurology</i> , 2014, 71, 1305.	9.0	57
22	Recurrent FXVD2 p.Gly41Arg mutation in patients with isolated dominant hypomagnesaemia. <i>Nephrology Dialysis Transplantation</i> , 2015, 30, 952-957.	0.7	51
23	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. <i>American Journal of Human Genetics</i> , 2018, 102, 995-1007.	6.2	49
24	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. <i>European Journal of Human Genetics</i> , 2021, 29, 1325-1331.	2.8	49
25	De novo BK channel variant causes epilepsy by affecting voltage gating but not Ca <sup>2+</sup> sensitivity. <i>European Journal of Human Genetics</i> , 2018, 26, 220-229.	2.8	47
26	Prevalence and mutation spectrum of skeletal muscle channelopathies in the Netherlands. <i>Neuromuscular Disorders</i> , 2018, 28, 402-407.	0.6	40
27	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. <i>Acta Neuropathologica</i> , 2020, 139, 415-442.	7.7	38
28	The Genetic Homogeneity of CAPOS Syndrome: Four New Patients With the c.2452G>A (p.Glu818Lys) Mutation in the ATP1A3 Gene. <i>Pediatric Neurology</i> , 2016, 59, 71-75.e1.	2.1	35
29	Making sense of missense variants in TTN-related congenital myopathies. <i>Acta Neuropathologica</i> , 2021, 141, 431-453.	7.7	34
30	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. <i>Journal of Neuromuscular Diseases</i> , 2019, 6, 241-258.	2.6	32
31	A novel <i>SLC2A1</i> mutation linking hemiplegic migraine with alternating hemiplegia of childhood. <i>Cephalalgia</i> , 2015, 35, 10-15.	3.9	28
32	Missorting of the Aquaporin-2 mutant E258K to multivesicular bodies/lysosomes in dominant NDI is associated with its monoubiquitination and increased phosphorylation by PKC but is due to the loss of E258. <i>Pflugers Archiv European Journal of Physiology</i> , 2008, 455, 1041-1054.	2.8	27
33	Failure of ketogenic diet therapy in GLUT1 deficiency syndrome. <i>European Journal of Paediatric Neurology</i> , 2019, 23, 404-409.	1.6	26
34	The etiology of rhabdomyolysis: an interaction between genetic susceptibility and external triggers. <i>European Journal of Neurology</i> , 2021, 28, 647-659.	3.3	26
35	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. <i>American Journal of Human Genetics</i> , 2019, 105, 907-920.	6.2	22
36	Biallelic variants in <i>HPDL</i> cause pure and complicated hereditary spastic paraplegia. <i>Brain</i> , 2021, 144, 1422-1434.	7.6	22

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37	De novo SPAST mutations may cause a complex SPG4 phenotype. <i>Brain</i> , 2019, 142, e31-e31.	7.6	21
38	CAPN1 mutations: Expanding the CAPN1-related phenotype: From hereditary spastic paraparesis to spastic ataxia. <i>European Journal of Medical Genetics</i> , 2019, 62, 103605.	1.3	21
39	Systematic analysis of short tandem repeats in 38,095 exomes provides an additional diagnostic yield. <i>Genetics in Medicine</i> , 2021, 23, 1569-1573.	2.4	21
40	The epilepsy phenotypic spectrum associated with a recurrent <i>CUX2</i> variant. <i>Annals of Neurology</i> , 2018, 83, 926-934.	5.3	20
41	Functional impairments, fatigue and quality of life in RYR1-related myopathies: A questionnaire study. <i>Neuromuscular Disorders</i> , 2019, 29, 30-38.	0.6	20
42	Biallelic loss of function variants in <i>SYT2</i> cause a treatable congenital onset presynaptic myasthenic syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2272-2283.	1.2	20
43	Clinical exome sequencing—Mistakes and caveats. <i>Human Mutation</i> , 2022, 43, 1041-1055.	2.5	20
44	GDAP2 mutations implicate susceptibility to cellular stress in a new form of cerebellar ataxia. <i>Brain</i> , 2018, 141, 2592-2604.	7.6	19
45	Biallelic variants in the RNA exosome gene EXOSC5 are associated with developmental delays, short stature, cerebellar hypoplasia and motor weakness. <i>Human Molecular Genetics</i> , 2020, 29, 2218-2239.	2.9	19
46	The movement disorder spectrum of SCA21 (ATX-TMEM240): 3 novel families and systematic review of the literature. <i>Parkinsonism and Related Disorders</i> , 2019, 62, 215-220.	2.2	18
47	A recurrent de novo DYNC1H1 tail domain mutation causes spinal muscular atrophy with lower extremity predominance, learning difficulties and mild brain abnormality. <i>Neuromuscular Disorders</i> , 2018, 28, 750-756.	0.6	16
48	The complexities of CACNA1A in clinical neurogenetics. <i>Journal of Neurology</i> , 2022, 269, 3094-3108.	3.6	16
49	A Novel TTBK2 De Novo Mutation in a Danish Family with Early-Onset Spinocerebellar Ataxia. <i>Cerebellum</i> , 2017, 16, 268-271.	2.5	15
50	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. <i>European Journal of Human Genetics</i> , 2017, 25, 771-774.	2.8	15
51	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. <i>Brain</i> , 2022, 145, 208-223.	7.6	15
52	Human Stress Syndrome and the Expanding Spectrum of RYR1-Related Myopathies. <i>Cell Biochemistry and Biophysics</i> , 2016, 74, 85-87.	1.8	14
53	Two Cases of Autosomal Recessive Congenital Ichthyosis due to <i>CYP4F22</i> Mutations: Expanding the Genotype of Self-Healing Collodion Baby. <i>Pediatric Dermatology</i> , 2016, 33, e48-51.	0.9	13
54	Clinical presentation and long-term follow-up of dopamine beta hydroxylase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 554-565.	3.6	13

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55	Repulsion between Lys258 and upstream arginines explains the missorting of the AQP2 mutant p.Glu258Lys in nephrogenic diabetes insipidus. <i>Human Mutation</i> , 2009, 30, 1387-1396.	2.5	12
56	Loss of function mutation in <i>RUSC2</i> causes intellectual disability and secondary microcephaly. <i>Developmental Medicine and Child Neurology</i> , 2016, 58, 1317-1322.	2.1	12
57	Natural history, outcome measures and trial readiness in LAMA2-related muscular dystrophy and SELENON-related myopathy in children and adults: protocol of the LAST STRONG study. <i>BMC Neurology</i> , 2021, 21, 313.	1.8	12
58	Referral Indications for Malignant Hyperthermia Susceptibility Diagnostics in Patients without Adverse Anesthetic Events in the Era of Next-generation Sequencing. <i>Anesthesiology</i> , 2022, 136, 940-953.	2.5	12
59	Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. <i>Clinical Chemistry</i> , 2019, 65, 1295-1306.	3.2	11
60	Autosomal dominant GCH1 mutations causing spastic paraplegia at disease onset. <i>Parkinsonism and Related Disorders</i> , 2020, 74, 12-15.	2.2	11
61	Pathogenic variants in <i>TNNC2</i> cause congenital myopathy due to an impaired force response to calcium. <i>Journal of Clinical Investigation</i> , 2021, 131, .	8.2	11
62	DTYMK is essential for genome integrity and neuronal survival. <i>Acta Neuropathologica</i> , 2022, 143, 245-262.	7.7	11
63	Mobility Characteristics of Children with Spastic Paraplegia Due to a Mutation in the <i>KIF1A</i> Gene. <i>Neuropediatrics</i> , 2020, 51, 146-153.	0.6	10
64	A brother and sister with intellectual disability and characteristic neuroimaging findings. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 866-869.	1.6	9
65	Robust and accurate detection and sizing of repeats within the <i>DMPK</i> gene using a novel TP-PCR test. <i>Scientific Reports</i> , 2019, 9, 8280.	3.3	9
66	HyperCKemia and rhabdomyolysis in the neuroleptic malignant and serotonin syndromes: A literature review. <i>Neuromuscular Disorders</i> , 2020, 30, 949-958.	0.6	9
67	Biallelic Variants in the <i>COLGALT1</i> Gene Causes Severe Congenital Porencephaly. <i>Neurology: Genetics</i> , 2021, 7, e564.	1.9	9
68	Genotype-phenotype correlations of <i>KIF5A</i> stalk domain variants. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021, 22, 561-570.	1.7	9
69	Spectrum of Clinical Features in X-Linked Myotubular Myopathy Carriers. <i>Neurology</i> , 2021, 97, e501-e512.	1.1	9
70	A novel Ile1455Thr variant in the skeletal muscle sodium channel alpha-subunit in a patient with a severe adult-onset proximal myopathy with electrical myotonia and a patient with mild paramyotonia phenotype. <i>Neuromuscular Disorders</i> , 2017, 27, 175-182.	0.6	8
71	De novo variants in <i>CAMTA1</i> cause a syndrome variably associated with spasticity, ataxia, and intellectual disability. <i>European Journal of Human Genetics</i> , 2020, 28, 763-769.	2.8	7
72	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. <i>Clinical Genetics</i> , 2021, 100, 692-702.	2.0	7

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73	<i>KIF16B</i> is a candidate gene for a novel autosomal recessive intellectual disability syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1602-1609.	1.2	6
74	The Phenotypic Spectrum of PNKP-Associated Disease and the Absence of Immunodeficiency and Cancer Predisposition in a Dutch Cohort. <i>Pediatric Neurology</i> , 2020, 113, 26-32.	2.1	6
75	Dominant Centronuclear Myopathy with Early Childhood Onset due to a Novel Mutation in BIN1. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 349-355.	2.6	5
76	Complicated hereditary spastic paraplegia due to ATP13A2 mutations: what's in a name?. <i>Brain</i> , 2017, 140, e73-e73.	7.6	5
77	Benign nocturnal alternating hemiplegia of childhood: A clinical and nomenclatural reappraisal. <i>European Journal of Paediatric Neurology</i> , 2018, 22, 1110-1117.	1.6	5
78	De novo variants in <i>EMC1</i> lead to neurodevelopmental delay and cerebellar degeneration and affect glial function in <i>Drosophila</i> . <i>Human Molecular Genetics</i> , 2022, 31, 3231-3244.	2.9	5
79	A hereditary spastic paraplegia predominant phenotype caused by variants in the NEFL gene. <i>Parkinsonism and Related Disorders</i> , 2020, 80, 98-101.	2.2	4
80	Recessive null-allele variants in MAG associated with spastic ataxia, nystagmus, neuropathy, and dystonia. <i>Parkinsonism and Related Disorders</i> , 2020, 77, 70-75.	2.2	3
81	Expanding Phenotype of <i>ATP1A3</i> - Related Disorders: A Case Series. <i>Child Neurology Open</i> , 2021, 8, 2329048X2110480.	1.1	3
82	Reply: A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia; and Novel GDAP2 pathogenic variants cause autosomal recessive spinocerebellar ataxia-27 (SCAR27) in a Chinese family. <i>Brain</i> , 2020, 143, e51-e51.	7.6	1
83	Paroxysmal Kinesigenic Dyskinesia: First Molecularly Confirmed Case from Africa. <i>Tremor and Other Hyperkinetic Movements</i> , 2019, 10, .	2.0	0
84	Slow Channel Syndrome Revisited: 40 Years Clinical Follow-Up and Genetic Characterization of Two Cases. <i>Journal of Neuromuscular Diseases</i> , 2022, , 1-8.	2.6	0