Erik-jan Kamsteeg

List of Publications by Year
in descending order

84	3,908	185998 28	143772 57
papers	citations	h-index	g-index

85	85	85	8130
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	7.1	407
2	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
3	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	1.1	220
4	Short-chain ubiquitination mediates the regulated endocytosis of the aquaporin-2 water channel. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 18344-18349.	3.3	214
5	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	9.4	186
6	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	1.1	143
7	Best practice guidelines and recommendations on the molecular diagnosis of myotonic dystrophy types 1 and 2. European Journal of Human Genetics, 2012, 20, 1203-1208.	1.4	129
8	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.	1.5	124
9	Mutations in VPS13D lead to a new recessive ataxia with spasticity and mitochondrial defects. Annals of Neurology, 2018, 83, 1075-1088.	2.8	122
10	Loss-of-function mutations inSCN4Acause severe foetal hypokinesia or â€˜classicalâ€™ congenital myopathy. Brain, 2016, 139, 674-691.	3.7	100
11	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	2.6	96
12	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
13	Clinical exome sequencing for cerebellar ataxia and spastic paraplegia uncovers novel geneâ€“disease associations and unanticipated rare disorders. European Journal of Human Genetics, 2016, 24, 1460-1466.	1.4	89
14	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.	2.6	87
15	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
16	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	2.6	82
17	Understanding the Psychosocial Effects of WES Test Results on Parents of Children with Rare Diseases. Journal of Genetic Counseling, 2016, 25, 1207-1214.	0.9	73
18	Lossâ€ofâ€Function Variants in <scp>HOPS</scp> Complex Genes <scp>VPS16</scp> and <scp>VPS41</scp> Cause Early Onset Dystonia Associated with Lysosomal Abnormalities. Annals of Neurology, 2020, 88, 867-877.	2.8	70

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19	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. Epilepsia, 2019, 60, 155-164.	2.6	65
20	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. European Journal of Human Genetics, 2020, 28, 40-49.	1.4	65
21	Autosomal Recessive Cerebellar Ataxia Type 3 Due toANO10Mutations. JAMA Neurology, 2014, 71, 1305.	4.5	57
22	Recurrent FXYD2 p.Gly41Arg mutation in patients with isolated dominant hypomagnesaemia. Nephrology Dialysis Transplantation, 2015, 30, 952-957.	0.4	51
23	A Recurrent De Novo PACS2 Heterozygous Missense Variant Causes Neonatal-Onset Developmental Epileptic Encephalopathy, Facial Dysmorphism, and Cerebellar Dysgenesis. American Journal of Human Genetics, 2018, 102, 995-1007.	2.6	49
24	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	1.4	49
25	De novo BK channel variant causes epilepsy by affecting voltage gating but not Ca2+ sensitivity. European Journal of Human Genetics, 2018, 26, 220-229.	1.4	47
26	Prevalence and mutation spectrum of skeletal muscle channelopathies in the Netherlands. Neuromuscular Disorders, 2018, 28, 402-407.	0.3	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. Acta Neuropathologica, 2020, 139, 415-442.	3.9	38
28	The Genetic Homogeneity of CAPOS Syndrome: Four New Patients With the c.2452G>A (p.Glu818Lys) Mutation in the ATP1A3 Gene. Pediatric Neurology, 2016, 59, 71-75.e1.	1.0	35
29	Making sense of missense variants in TTN-related congenital myopathies. Acta Neuropathologica, 2021, 141, 431-453.	3.9	34
30	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. Journal of Neuromuscular Diseases, 2019, 6, 241-258.	1.1	32
31	A novel SLC2A1 mutation linking hemiplegic migraine with alternating hemiplegia of childhood. Cephalalgia, 2015, 35, 10-15.	1.8	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. Pflugers Archiv European Journal of Physiology, 2008, 455, 1041-1054.	1.3	27
33	Failure of ketogenic diet therapy in GLUT1 deficiency syndrome. European Journal of Paediatric Neurology, 2019, 23, 404-409.	0.7	26
34	The etiology of rhabdomyolysis: an interaction between genetic susceptibility and external triggers. European Journal of Neurology, 2021, 28, 647-659.	1.7	26
35	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. American Journal of Human Genetics, 2019, 105, 907-920.	2.6	22
36	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22

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37	De novo SPAST mutations may cause a complex SPG4 phenotype. Brain, 2019, 142, e31-e31.	3.7	21
38	CAPN1 mutations: Expanding the CAPN1-related phenotype: From hereditary spastic paraparesis to spastic ataxia. European Journal of Medical Genetics, 2019, 62, 103605.	0.7	21
39	Systematic analysis of short tandem repeats in 38,095 exomes provides an additional diagnostic yield. Genetics in Medicine, 2021, 23, 1569-1573.	1.1	21
40	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	2.8	20
41	Functional impairments, fatigue and quality of life in RYR1-related myopathies: A questionnaire study. Neuromuscular Disorders, 2019, 29, 30-38.	0.3	20
42	Biallelic loss of function variants in <scp>SYT2</scp> cause a treatable congenital onset presynaptic myasthenic syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2272-2283.	0.7	20
43	Clinical exome sequencingâ€"Mistakes and caveats. Human Mutation, 2022, 43, 1041-1055.	1.1	20
44	GDAP2 mutations implicate susceptibility to cellular stress in a new form of cerebellar ataxia. Brain, 2018, 141, 2592-2604.	3.7	19
45	Biallelic variants in the RNA exosome gene EXOSC5 are associated with developmental delays, short stature, cerebellar hypoplasia and motor weakness. Human Molecular Genetics, 2020, 29, 2218-2239.	1.4	19
46	The movement disorder spectrum of SCA21 (ATX-TMEM240): 3 novel families and systematic review of the literature. Parkinsonism and Related Disorders, 2019, 62, 215-220.	1.1	18
47	A recurrent de novo DYNC1H1 tail domain mutation causes spinal muscular atrophy with lower extremity predominance, learning difficulties and mild brain abnormality. Neuromuscular Disorders, 2018, 28, 750-756.	0.3	16
48	The complexities of CACNA1A in clinical neurogenetics. Journal of Neurology, 2022, 269, 3094-3108.	1.8	16
49	A Novel TTBK2 De Novo Mutation in a Danish Family with Early-Onset Spinocerebellar Ataxia. Cerebellum, 2017, 16, 268-271.	1.4	15
50	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	1.4	15
51	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	3.7	15
52	â€œHuman Stress Syndromeâ€•and the Expanding Spectrum of RYR1-Related Myopathies. Cell Biochemistry and Biophysics, 2016, 74, 85-87.	0.9	14
53	Two Cases of Autosomal Recessive Congenital Ichthyosis due to CYP4F22 Mutations: Expanding the Genotype of Selfâ€Healing Collodion Baby. Pediatric Dermatology, 2016, 33, e48-51.	0.5	13
54	Clinical presentation and longâ€term followâ€up of dopamine beta hydroxylase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 554-565.	1.7	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. Human Mutation, 2009, 30, 1387-1396.	1.1	12
56	Lossâ€ofâ€function mutation in RUSC2 causes intellectual disability and secondary microcephaly. Developmental Medicine and Child Neurology, 2016, 58, 1317-1322.	1.1	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. BMC Neurology, 2021, 21, 313.	0.8	12
58	Referral Indications for Malignant Hyperthermia Susceptibility Diagnostics in Patients without Adverse Anesthetic Events in the Era of Next-generation Sequencing. Anesthesiology, 2022, 136, 940-953.	1.3	12
59	Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. Clinical Chemistry, 2019, 65, 1295-1306.	1.5	11
60	Autosomal dominant GCH1 mutations causing spastic paraplegia at disease onset. Parkinsonism and Related Disorders, 2020, 74, 12-15.	1.1	11
61	Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium. Journal of Clinical Investigation, 2021, 131, .	3.9	11
62	DTYMK is essential for genome integrity and neuronal survival. Acta Neuropathologica, 2022, 143, 245-262.	3.9	11
63	Mobility Characteristics of Children with Spastic Paraplegia Due to a Mutation in the KIF1A Gene. Neuropediatrics, 2020, 51, 146-153.	0.3	10
64	A brother and sister with intellectual disability and characteristic neuroimaging findings. European Journal of Paediatric Neurology, 2018, 22, 866-869.	0.7	9
65	Robust and accurate detection and sizing of repeats within the DMPK gene using a novel TP-PCR test. Scientific Reports, 2019, 9, 8280.	1.6	9
66	HyperCKemia and rhabdomyolysis in the neuroleptic malignant and serotonin syndromes: A literature review. Neuromuscular Disorders, 2020, 30, 949-958.	0.3	9
67	Biallelic Variants in the COLGALT1 Gene Causes Severe Congenital Porencephaly. Neurology: Genetics, 2021, 7, e564.	0.9	9
68	Genotype-phenotype correlations of KIF5A stalk domain variants. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 561-570.	1.1	9
69	Spectrum of Clinical Features in X-Linked Myotubular Myopathy Carriers. Neurology, 2021, 97, e501-e512.	1.5	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. Neuromuscular Disorders, 2017, 27, 175-182.	0.3	8
71	De novo variants in CAMTA1 cause a syndrome variably associated with spasticity, ataxia, and intellectual disability. European Journal of Human Genetics, 2020, 28, 763-769.	1.4	7
72	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. Clinical Genetics, 2021, 100, 692-702.	1.0	7

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73	KIF16B is a candidate gene for a novel autosomalâ€recessive intellectual disability syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1602-1609.	0.7	6
74	The Phenotypic Spectrum of PNKP-Associated Disease and the Absence of Immunodeficiency and Cancer Predisposition in a Dutch Cohort. Pediatric Neurology, 2020, 113, 26-32.	1.0	6
75	Dominant Centronuclear Myopathy with Early Childhood Onset due to a Novel Mutation in BIN1. Journal of Neuromuscular Diseases, 2017, 4, 349-355.	1.1	5
76	Complicated hereditary spastic paraplegia due to ATP13A2 mutations: whatâ€™s in a name?. Brain, 2017, 140, e73-e73.	3.7	5
77	Benign nocturnal alternating hemiplegia of childhood: A clinical and nomenclatural reappraisal. European Journal of Paediatric Neurology, 2018, 22, 1110-1117.	0.7	5
78	De novo variants in EMC1 lead to neurodevelopmental delay and cerebellar degeneration and affect glial function in Drosophila. Human Molecular Genetics, 2022, 31, 3231-3244.	1.4	5
79	A hereditary spastic paraplegia predominant phenotype caused by variants in the NEFL gene. Parkinsonism and Related Disorders, 2020, 80, 98-101.	1.1	4
80	Recessive null-allele variants in MAG associated with spastic ataxia, nystagmus, neuropathy, and dystonia. Parkinsonism and Related Disorders, 2020, 77, 70-75.	1.1	3
81	Expanding Phenotype ofATP1A3- Related Disorders: A Case Series. Child Neurology Open, 2021, 8, 2329048X2110480.	0.5	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. Brain, 2020, 143, e51-e51.	3.7	1
83	Paroxysmal Kinesigenic Dyskinesia: First Molecularly Confirmed Case from Africa. Tremor and Other Hyperkinetic Movements, 2019, 10, .	1.1	0
84	Slow Channel Syndrome Revisited: 40 Years Clinical Follow-Up and Genetic Characterization of Two Cases. Journal of Neuromuscular Diseases, 2022, , 1-8.	1.1	0