Erik-jan Kamsteeg

List of Publications by Year
in descending order

84	3,908	186265	144013
papers	citations	28 h-index	57 g-index

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

#	Article	IF	Citations
1	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder with myoclonus. Brain, 2022, 145, 208-223.	7.6	15
2	The complexities of CACNA1A in clinical neurogenetics. Journal of Neurology, 2022, 269, 3094-3108.	3.6	16
3	Clinical exome sequencingâ€"Mistakes and caveats. Human Mutation, 2022, 43, 1041-1055.	2.5	20
4	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.	2.9	5
5	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.	2.5	12
6	DTYMK is essential for genome integrity and neuronal survival. Acta Neuropathologica, 2022, 143, 245-262.	7.7	11
7	Slow Channel Syndrome Revisited: 40 Years Clinical Follow-Up and Genetic Characterization of Two Cases. Journal of Neuromuscular Diseases, 2022, , 1-8.	2.6	0
8	Clinical presentation and longâ€term followâ€up of dopamine beta hydroxylase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 554-565.	3.6	13
9	Biallelic Variants in the COLGALT1 Gene Causes Severe Congenital Porencephaly. Neurology: Genetics, 2021, 7, e564.	1.9	9
10	Systematic analysis of short tandem repeats in 38,095 exomes provides an additional diagnostic yield. Genetics in Medicine, 2021, 23, 1569-1573.	2.4	21
11	Genotype-phenotype correlations of KIF5A stalk domain variants. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 561-570.	1.7	9
12	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
13	Spectrum of Clinical Features in X-Linked Myotubular Myopathy Carriers. Neurology, 2021, 97, e501-e512.	1.1	9
14	Pathogenic variants in TNNC2 cause congenital myopathy due to an impaired force response to calcium. Journal of Clinical Investigation, 2021, 131, .	8.2	11
15	Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. European Journal of Human Genetics, 2021, 29, 1325-1331.	2.8	49
16	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.	1.8	12
17	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. Clinical Genetics, 2021, 100, 692-702.	2.0	7
18	Making sense of missense variants in TTN-related congenital myopathies. Acta Neuropathologica, 2021, 141, 431-453.	7.7	34

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19	The etiology of rhabdomyolysis: an interaction between genetic susceptibility and external triggers. European Journal of Neurology, 2021, 28, 647-659.	3.3	26
20	Expanding Phenotype ofATP1A3- Related Disorders: A Case Series. Child Neurology Open, 2021, 8, 2329048X2110480.	1.1	3
21	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. European Journal of Human Genetics, 2020, 28, 40-49.	2.8	65
22	Mobility Characteristics of Children with Spastic Paraplegia Due to a Mutation in the KIF1A Gene. Neuropediatrics, 2020, 51, 146-153.	0.6	10
23	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.	7.7	38
24	A hereditary spastic paraplegia predominant phenotype caused by variants in the NEFL gene. Parkinsonism and Related Disorders, 2020, 80, 98-101.	2.2	4
25	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.	2.1	6
26	Recessive null-allele variants in MAG associated with spastic ataxia, nystagmus, neuropathy, and dystonia. Parkinsonism and Related Disorders, 2020, 77, 70-75.	2.2	3
27	HyperCKemia and rhabdomyolysis in the neuroleptic malignant and serotonin syndromes: A literature review. Neuromuscular Disorders, 2020, 30, 949-958.	0.6	9
28	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.	1.2	20
29	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.	5.3	70
30	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.	7.6	1
31	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.	2.9	19
32	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.	2.8	7
33	Autosomal dominant GCH1 mutations causing spastic paraplegia at disease onset. Parkinsonism and Related Disorders, 2020, 74, 12-15.	2.2	11
34	Cytidine Diphosphate-Ribitol Analysis for Diagnostics and Treatment Monitoring of Cytidine Diphosphate-l-Ribitol Pyrophosphorylase A Muscular Dystrophy. Clinical Chemistry, 2019, 65, 1295-1306.	3.2	11
35	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. American Journal of Human Genetics, 2019, 105, 907-920.	6.2	22
36	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. Journal of Neuromuscular Diseases, 2019, 6, 241-258.	2.6	32

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37	Robust and accurate detection and sizing of repeats within the DMPK gene using a novel TP-PCR test. Scientific Reports, 2019, 9, 8280.	3.3	9
38	De novo SPAST mutations may cause a complex SPG4 phenotype. Brain, 2019, 142, e31-e31.	7.6	21
39	Failure of ketogenic diet therapy in GLUT1 deficiency syndrome. European Journal of Paediatric Neurology, 2019, 23, 404-409.	1.6	26
40	Diagnostic exome sequencing in 100 consecutive patients with both epilepsy and intellectual disability. Epilepsia, 2019, 60, 155-164.	5.1	65
41	CAPN1 mutations: Expanding the CAPN1-related phenotype: From hereditary spastic paraparesis to spastic ataxia. European Journal of Medical Genetics, 2019, 62, 103605.	1.3	21
42	Functional impairments, fatigue and quality of life in RYR1-related myopathies: A questionnaire study. Neuromuscular Disorders, 2019, 29, 30-38.	0.6	20
43	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.	2.2	18
44	Paroxysmal Kinesigenic Dyskinesia: First Molecularly Confirmed Case from Africa. Tremor and Other Hyperkinetic Movements, 2019, 10, .	2.0	0
45	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.	6.2	49
46	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
47	Mutations in VPS13D lead to a new recessive ataxia with spasticity and mitochondrial defects. Annals of Neurology, 2018, 83, 1075-1088.	5.3	122
48	The epilepsy phenotypic spectrum associated with a recurrent CUX2 variant. Annals of Neurology, 2018, 83, 926-934.	5.3	20
49	De novo BK channel variant causes epilepsy by affecting voltage gating but not Ca2+ sensitivity. European Journal of Human Genetics, 2018, 26, 220-229.	2.8	47
50	Prevalence and mutation spectrum of skeletal muscle channelopathies in the Netherlands. Neuromuscular Disorders, 2018, 28, 402-407.	0.6	40
51	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
52	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.	6.2	87
53	A brother and sister with intellectual disability and characteristic neuroimaging findings. European Journal of Paediatric Neurology, 2018, 22, 866-869.	1.6	9
54	GDAP2 mutations implicate susceptibility to cellular stress in a new form of cerebellar ataxia. Brain, 2018, 141, 2592-2604.	7.6	19

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55	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.	0.6	16
56	KIF16B is a candidate gene for a novel autosomalâ€recessive intellectual disability syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1602-1609.	1.2	6
57	Benign nocturnal alternating hemiplegia of childhood: A clinical and nomenclatural reappraisal. European Journal of Paediatric Neurology, 2018, 22, 1110-1117.	1.6	5
58	A Novel TTBK2 De Novo Mutation in a Danish Family with Early-Onset Spinocerebellar Ataxia. Cerebellum, 2017, 16, 268-271.	2.5	15
59	Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal-Recessive Cutis Laxa. American Journal of Human Genetics, 2017, 100, 216-227.	6.2	82
60	Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome. European Journal of Human Genetics, 2017, 25, 771-774.	2.8	15
61	A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology. Genetics in Medicine, 2017, 19, 1055-1063.	2.4	220
62	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
63	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
64	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.	0.6	8
65	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	2.4	143
66	Dominant Centronuclear Myopathy with Early Childhood Onset due to a Novel Mutation in BIN1. Journal of Neuromuscular Diseases, 2017, 4, 349-355.	2.6	5
67	Complicated hereditary spastic paraplegia due to ATP13A2 mutations: whatâ€™s in a name?. Brain, 2017, 140, e73-e73.	7.6	5
68	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.	2.8	89
69	De Novo Mutations in PDE10A Cause Childhood-Onset Chorea with Bilateral Striatal Lesions. American Journal of Human Genetics, 2016, 98, 763-771.	6.2	96
70	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.	2.1	35
71	Understanding the Psychosocial Effects of WES Test Results on Parents of Children with Rare Diseases. Journal of Genetic Counseling, 2016, 25, 1207-1214.	1.6	73
72	Lossâ€ofâ€function mutation in RUSC2 causes intellectual disability and secondary microcephaly. Developmental Medicine and Child Neurology, 2016, 58, 1317-1322.	2.1	12

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73	Meta-analysis of 2,104 trios provides support for 10 new genes for intellectual disability. Nature Neuroscience, 2016, 19, 1194-1196.	14.8	407
74	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.	0.9	13
75	Loss-of-function mutations inSCN4Acause severe foetal hypokinesia or â€˜classicalâ€™ congenital myopathy. Brain, 2016, 139, 674-691.	7.6	100
76	â€œHuman Stress Syndromeâ€•and the Expanding Spectrum of RYR1-Related Myopathies. Cell Biochemistry and Biophysics, 2016, 74, 85-87.	1.8	14
77	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
78	Recurrent FXYD2 p.Gly41Arg mutation in patients with isolated dominant hypomagnesaemia. Nephrology Dialysis Transplantation, 2015, 30, 952-957.	0.7	51
79	A novel SLC2A1 mutation linking hemiplegic migraine with alternating hemiplegia of childhood. Cephalalgia, 2015, 35, 10-15.	3.9	28
80	Autosomal Recessive Cerebellar Ataxia Type 3 Due toANO10Mutations. JAMA Neurology, 2014, 71, 1305.	9.0	57
81	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.	2.8	129
82	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.	2.5	12
83	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.	2.8	27
84	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.	7.1	214