Anna Kostera-Pruszczyk

List of Publications by Year
in descending order

98 papers	3,315 citations	218677 26 h-index	161849 54 g-index
102 all docs	102 docs citations	102 times ranked	3956 citing authors

#	Article	IF	Citations
1	Myasthenia gravisâ€"treatment and severity in nationwide cohort. Acta Neurologica Scandinavica, 2022, 145, 471-478.	2.1	3
2	Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial. Lancet Neurology, The, 2022, 21, 42-52.	10.2	89
3	Quantitative magnetic resonance imaging measures as biomarkers of disease progression in boys with Duchenne muscular dystrophy: a phase 2 trial of domagrozumab. Journal of Neurology, 2022, 269, 4421-4435.	3.6	6
4	Safety, tolerability, and efficacy of a widely available nusinersen program for Polish children with Spinal Muscular Atrophy. European Journal of Paediatric Neurology, 2022, 39, 103-109.	1.6	3
5	Parametric analysis of pilot voice signals in Parkinson?s disease diagnostics. Journal of Automation Electronics and Electrical Engineering, 2022, 4, 21-28.	0.2	1
6	Analysis of hand and face images for the purpose of engineering support for Parkinson's disease diagnosis. Journal of Automation Electronics and Electrical Engineering, 2022, 4, 13-20.	0.2	0
7	Acute Autonomic Neuropathy as a Rare Cause of Severe Arterial Hypertension in a Child. International Journal of Pediatrics and Adolescent Medicine, 2021, 8, 121-124.	1.2	0
8	Response to letter: A decision for life â€“ Treatment decisions in newly diagnosed families with spinal muscular atrophy. European Journal of Paediatric Neurology, 2021, 30, 103-104.	1.6	1
9	Myasthenia Gravis in Poland: National Healthcare Database Epidemiological Study. Neuroepidemiology, 2021, 55, 62-69.	2.3	5
10	Comprehensive non-invasive assessment of electrocardiographic abnormalities and cardiac arrhythmias in patients with genetically confirmed mitochondrial diseases. Journal of Electrocardiology, 2021, 65, 136-142.	0.9	1
11	Observation of the natural course of type 3 spinal muscular atrophy: data from the polish registry of spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2021, 16, 150.	2.7	22
12	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
13	Floppy infant syndrome as a first manifestation of LMNA-related congenital muscular dystrophy. European Journal of Paediatric Neurology, 2021, 32, 115-121.	1.6	4
14	Facial onset sensory and motor neuronopathy syndrome â€" a rare variant of motor neurone disease. Neurologia I Neurochirurgia Polska, 2021, 55, 325-327.	1.2	3
15	Safety, efficacy, and tolerability of efgartigimod in patients with generalised myasthenia gravis (ADAPT): a multicentre, randomised, placebo-controlled, phase 3 trial. Lancet Neurology, The, 2021, 20, 526-536.	10.2	194
16	Pediatric CIDP: Diagnosis and Management. A Single-Center Experience. Frontiers in Neurology, 2021, 12, 667378.	2.4	8
17	Lower BAFF Levels in Myasthenic Patients Treated with Glucocorticoids. Archivum Immunologiae Et Therapiae Experimentalis, 2021, 69, 22.	2.3	1
18	Ultra-low radiation dose protocol for CT-guided intrathecal nusinersen injections for patients with spinal muscular atrophy and severe scoliosis. Neuroradiology, 2021, 63, 539-545.	2.2	8

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19	Targeted Next-Generation Sequencing Reveals Mutations in Non-coding Regions and Potential Regulatory Sequences of Calpain-3 Gene in Polish Limbâ€“Girdle Muscular Dystrophy Patients. Frontiers in Neuroscience, 2021, 15, 692482.	2.8	6
20	Spinal muscular atrophy: epidemiology and health burden in children â€" a Polish national healthcare database perspective before introduction of SMA-specific treatment. Neurologia I Neurochirurgia Polska, 2021, 55, 479-484.	1.2	2
21	Generation of DMBi002-A human induced pluripotent stem cell line from patient with Spinal muscular atrophy type 3. Stem Cell Research, 2021, 57, 102563.	0.7	0
22	Progressive External Ophthalmoplegia in Polish Patientsâ€"From Clinical Evaluation to Genetic Confirmation. Genes, 2021, 12, 54.	2.4	1
23	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
24	A Study on the Possible Diagnosis of Parkinsonâ€™s Disease on the Basis of Facial Image Analysis. Electronics (Switzerland), 2021, 10, 2832.	3.1	4
25	Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. Lancet Neurology, The, 2021, 20, 1012-1026.	10.2	59
26	Determinants of Quality of Life in Myasthenia Gravis Patients. Frontiers in Neurology, 2020, 11, 553626.	2.4	23
27	Randomized phase 2 trial and open-label extension of domagrozumab in Duchenne muscular dystrophy. Neuromuscular Disorders, 2020, 30, 492-502.	0.6	40
28	European ad-hoc consensus statement on gene replacement therapy for spinal muscular atrophy. European Journal of Paediatric Neurology, 2020, 28, 38-43.	1.6	74
29	Minimal manifestation status and prednisone withdrawal in the MGTX trial. Neurology, 2020, 95, e755-e766.	1.1	17
30	Lack of miR-378 attenuates muscular dystrophy in mdx mice. JCI Insight, 2020, 5, .	5.0	22
31	Spinal muscular atrophy â€" new therapies, new challenges. Neurologia I Neurochirurgia Polska, 2020, 54, 8-13.	1.2	6
32	Transthyretin-related familial amyloid polyneuropathy (ATTR-FAP) in Poland â€" genetic and clinical presentation. Neurologia I Neurochirurgia Polska, 2020, 54, 552-560.	1.2	1
33	Selected problems of image data preprocessing used to perform examination in Parkinsonâ€™s disease. , 2020, , .		1
34	Multimodal data acquisition set for objective assessment of Parkinsonâ€™s disease. , 2020, , .		4
35	The use of non-linear acoustic analysis to objectively evaluate the voice of people with Parkinson's disease. , 2020, , .		0
36	Spinal muscular atrophy with an overlapping syndrome â€" â€œdouble troubleâ€•or a potentially better outcome?. Neurologia I Neurochirurgia Polska, 2020, 54, 475-477.	1.2	1

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37	Gross Motor Function Disorders in Patients with Alternating Hemiplegia of Childhood. Medycyna Wieku Rozwojowego, 2020, 24, 24-32.	0.2	1
38	The needle EMG findings in myotonia congenita. Journal of Electromyography and Kinesiology, 2019, 49, 102362.	1.7	4
39	Malignant hyperthermia â€“ what do we know in 2019?. Anaesthesiology Intensive Therapy, 2019, 51, 169-177.	1.0	5
40	Long-term effect of thymectomy plus prednisone versus prednisone alone in patients with non-thymomatous myasthenia gravis: 2-year extension of the MGTX randomised trial. Lancet Neurology, The, 2019, 18, 259-268.	10.2	139
41	Screening for lateâ€onset Pompe disease in Poland. Acta Neurologica Scandinavica, 2019, 140, 239-243.	2.1	8
42	Motor Unit Number Index (MUNIX) as a biomarker of motor unit loss in post-polio syndrome versus needle EMG. Journal of Electromyography and Kinesiology, 2019, 46, 35-40.	1.7	8
43	Treatment outcome in juvenileâ€onset myasthenia gravis. Muscle and Nerve, 2019, 59, 549-554.	2.2	11
44	High incidence and clinical characteristics of fibromuscular dysplasia in patients with spontaneous cervical artery dissection: The ARCADIA-POL study. Vascular Medicine, 2019, 24, 112-119.	1.5	23
45	The remarkable phenotypic variability of the p.Arg269HiS variant in the TRPV4 gene. Muscle and Nerve, 2019, 59, 129-133.	2.2	8
46	The frequency of mitochondrial polymerase gamma related disorders in a large Polish population cohort. Mitochondrion, 2019, 47, 179-187.	3.4	4
47	Electromyographic findings in sporadic inclusion body myositis. Journal of Electromyography and Kinesiology, 2018, 39, 114-119.	1.7	10
48	Heme Oxygenase-1 Influences Satellite Cells and Progression of Duchenne Muscular Dystrophy in Mice. Antioxidants and Redox Signaling, 2018, 29, 128-148.	5.4	29
49	Evidence for a relatively high proportion of DM2 mutations in a large group of Polish patients. Neurologia I Neurochirurgia Polska, 2018, 52, 736-742.	1.2	0
50	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	6.2	40
51	Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease. Orphanet Journal of Rare Diseases, 2018, 13, 155.	2.7	19
52	Propafenone is not effective for severe ventricular arrhythmias in Andersen-Tawil syndrome. Archives of Medical Science, 2018, 1, 247-250.	0.9	2
53	Serum interleukin 15 levels in patients with seropositive myasthenia gravis do not correlate with disease severity. Neurologia I Neurochirurgia Polska, 2018, 52, 364-367.	1.2	0
54	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. Skeletal Muscle, 2018, 8, 23.	4.2	40

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55	A review of functional assessment scales in non-sitters with spinal muscular atrophy (SMA). , 2018, 27, 11-17.		0
56	Abnormal spontaneous activity in primary myopathic disorders. Muscle and Nerve, 2017, 56, 427-432.	2.2	6
57	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. Journal of Neuromuscular Diseases, 2017, 4, 293-306.	2.6	125
58	Mechanical thrombectomy in acute stroke â€“ Five years of experience in Poland. Neurologia I Neurochirurgia Polska, 2017, 51, 339-346.	1.2	11
59	Dopa-responsive dystonia or early-onset Parkinson disease â€“ Genotypeâ€“phenotype correlation. Neurologia I Neurochirurgia Polska, 2017, 51, 1-6.	1.2	13
60	Novel Col12A1 variant expands the clinical picture of congenital myopathies with extracellular matrix defects. Muscle and Nerve, 2017, 55, 277-281.	2.2	31
61	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. Orphanet Journal of Rare Diseases, 2017, 12, 173.	2.7	21
62	European Cross-Sectional Survey ofÂCurrent Care Practices for Duchenne Muscular Dystrophy Reveals Regional andÂAge-Dependent Differences. Journal of Neuromuscular Diseases, 2016, 3, 517-527.	2.6	55
63	Amifampridine phosphate (Firdapse^{Â®}) is effective and safe in a phase 3 clinical trial in LEMS. Muscle and Nerve, 2016, 53, 717-725.	2.2	51
64	Randomized Trial of Thymectomy in Myasthenia Gravis. New England Journal of Medicine, 2016, 375, 511-522.	27.0	695
65	Are electrophysiological criteria useful in distinguishing childhood demyelinating neuropathies?. Journal of the Peripheral Nervous System, 2016, 21, 22-26.	3.1	12
66	Prevalence and impact of autoimmune thyroid disease on myasthenia gravis course. Brain and Behavior, 2016, 6, e00537.	2.2	33
67	Motor unit number estimation as a complementary test to routine electromyography in the diagnosis of amyotrophic lateral sclerosis. Journal of Electromyography and Kinesiology, 2016, 26, 60-65.	1.7	7
68	Genome-Wide Association Study of Late-Onset Myasthenia Gravis: Confirmation of TNFRSF11A and Identification of ZBTB10 and Three Distinct HLA Associations. Molecular Medicine, 2015, 21, 769-781.	4.4	52
69	X-linked spinal muscular atrophy (SMAX2) caused by de novo c.1731C>T substitution in the UBA1 gene. Neuromuscular Disorders, 2015, 25, 661-666.	0.6	13
70	Andersenâ€“Tawil syndrome: Report of 3 novel mutations and high risk of symptomatic cardiac involvement. Muscle and Nerve, 2015, 51, 192-196.	2.2	16
71	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. Human Mutation, 2015, 36, 395-402.	2.5	507
72	The LITAF/SIMPLE I92V sequence variant results in an earlier age of onset of CMT1A/HNPP diseases. Neurogenetics, 2015, 16, 27-32.	1.4	20

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73	Yeast model analysis of novel polymerase gamma variants found in patients with autosomal recessive mitochondrial disease. Human Genetics, 2015, 134, 951-966.	3.8	17
74	Motor unit loss estimation by the multipoint incremental MUNE method in children with spinal muscular atrophy â€“ A preliminary study. Neuromuscular Disorders, 2015, 25, 216-221.	0.6	21
75	BAG3-related myopathy, polyneuropathy and cardiomyopathy with long QT syndrome. Journal of Muscle Research and Cell Motility, 2015, 36, 423-432.	2.0	57
76	Clinical, electrophysiological, and molecular findings in early onset hereditary neuropathy with liability to pressure palsy. Muscle and Nerve, 2014, 50, 914-918.	2.2	21
77	Carpal Tunnel Syndrome in Children. Journal of Child Neurology, 2014, 29, 227-231.	1.4	17
78	<scp>VAV</scp>1 and <scp>BAFF</scp>, via <scp>NF</scp>ÎºB pathway, are genetic risk factors for myasthenia gravis. Annals of Clinical and Translational Neurology, 2014, 1, 329-339.	3.7	27
79	MLPA based detection of mutations in the dystrophin gene of 180 Polish families with Duchenne/Becker muscular dystrophy. Neurologia I Neurochirurgia Polska, 2014, 48, 416-422.	1.2	14
80	Effect of Age and Gender on the Number of Motor Units in Healthy Subjects Estimated by the Multipoint Incremental MUNE Method. Journal of Clinical Neurophysiology, 2014, 31, 272-278.	1.7	29
81	Novel point mutations in survival motor neuron 1 gene expand the spectrum of phenotypes observed in spinal muscular atrophy patients. Neuromuscular Disorders, 2014, 24, 617-623.	0.6	27
82	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. Journal of Neurology, 2014, 261, 152-163.	3.6	76
83	Antititin antibody in early- and late-onset myasthenia gravis. Acta Neurologica Scandinavica, 2014, 130, 229-233.	2.1	60
84	Exome sequencing reveals mutations in <scp>MFN2</scp> and <scp>GDAP1</scp> in severe Charcotâ€“Marieâ€“Tooth disease. Journal of the Peripheral Nervous System, 2014, 19, 242-245.	3.1	12
85	Original article Charcot-Marie-Tooth type 1C disease coexisting with progressive multiple sclerosis: a study of an overlapping syndrome. Folia Neuropathologica, 2012, 4, 369-374.	1.2	4
86	L239F founder mutation in GDAP1 is associated with a mild Charcotâ€“Marieâ€“Tooth type 4C4 (CMT4C4) phenotype. Neurogenetics, 2010, 11, 357-366.	1.4	15
87	Incidence of Spinal Muscular Atrophy in Poland â€“ More Frequent than Predicted?. Neuroepidemiology, 2010, 34, 152-157.	2.3	32
88	Juvenile seropositive myasthenia gravis with anti-MuSK antibody after thymectomy. Journal of Neurology, 2009, 256, 1780-1781.	3.6	28
89	Phenotype modifiers of spinal muscular atrophy: the number of SMN2 gene copies, deletion in the NAIP gene and probably gender influence the course of the disease.. Acta Biochimica Polonica, 2009, 56, .	0.5	37
90	Phenotype modifiers of spinal muscular atrophy: the number of SMN2 gene copies, deletion in the NAIP gene and probably gender influence the course of the disease. Acta Biochimica Polonica, 2009, 56, 103-8.	0.5	18

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91	Oculopharyngeal muscular dystrophy: phenotypic and genotypic characteristics of 9 Polish patients. Neurologia I Neurochirurgia Polska, 2009, 43, 113-20.	1.2	0
92	Unaffected patients with a homozygous absence of the SMN1 gene. European Journal of Human Genetics, 2008, 16, 930-934.	2.8	50
93	Accessory deep peroneal nerve - a clinically significant anomaly?. Neurologia I Neurochirurgia Polska, 2008, 42, 112-5.	1.2	5
94	Restrictive cardiomyopathy with atrioventricular conduction block resulting from a desmin mutation. International Journal of Cardiology, 2007, 117, 244-253.	1.7	66
95	Multi-minicore myopathy: a clinical and histopathological study of 17 cases. Folia Neuropathologica, 2007, 45, 56-65.	1.2	0
96	Juvenile onset acid maltase deficiency presenting as a rigid spine syndrome. Neuromuscular Disorders, 2006, 16, 282-285.	0.6	23
97	Myofibrillar myopathy with congenital cataract and skeletal anomalies without mutations in the desmin, Î±B-crystallin, myotilin, LMNA or SEPN1 genes. Neuromuscular Disorders, 2006, 16, 759-762.	0.6	4
98	Application of a rapid non-invasive technique in the molecular diagnosis of spinal muscular atrophy (SMA). Neurologia I Neurochirurgia Polska, 2005, 39, 89-94.	1.2	1