

Anna Kostera-Pruszczyk

List of Publications by Year in descending order

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Version: 2024-02-01

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	Randomized Trial of Thymectomy in Myasthenia Gravis. <i>New England Journal of Medicine</i> , 2016, 375, 511-522.	27.0	695
2	The TREAT-NMD DMD Global Database: Analysis of More than 7,000 Duchenne Muscular Dystrophy Mutations. <i>Human Mutation</i> , 2015, 36, 395-402.	2.5	507
3	Safety, efficacy, and tolerability of efgartigimod in patients with generalised myasthenia gravis (ADAPT): a multicentre, randomised, placebo-controlled, phase 3 trial. <i>Lancet Neurology</i> , The, 2021, 20, 526-536.	10.2	194
4	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. <i>Lancet Neurology</i> , The, 2019, 18, 259-268.	10.2	139
5	Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database. <i>Journal of Neuromuscular Diseases</i> , 2017, 4, 293-306.	2.6	125
6	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. <i>Lancet Neurology</i> , The, 2022, 21, 42-52.	10.2	89
7	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. <i>Journal of Neurology</i> , 2014, 261, 152-163.	3.6	76
8	European ad-hoc consensus statement on gene replacement therapy for spinal muscular atrophy. <i>European Journal of Paediatric Neurology</i> , 2020, 28, 38-43.	1.6	74
9	Restrictive cardiomyopathy with atrioventricular conduction block resulting from a desmin mutation. <i>International Journal of Cardiology</i> , 2007, 117, 244-253.	1.7	66
10	Antititin antibody in early- and late-onset myasthenia gravis. <i>Acta Neurologica Scandinavica</i> , 2014, 130, 229-233.	2.1	60
11	Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. <i>Lancet Neurology</i> , The, 2021, 20, 1012-1026.	10.2	59
12	BAG3-related myopathy, polyneuropathy and cardiomyopathy with long QT syndrome. <i>Journal of Muscle Research and Cell Motility</i> , 2015, 36, 423-432.	2.0	57
13	European Cross-Sectional Survey of Current Care Practices for Duchenne Muscular Dystrophy Reveals Regional and Age-Dependent Differences. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 517-527.	2.6	55
14	Genome-Wide Association Study of Late-Onset Myasthenia Gravis: Confirmation of TNFRSF11A and Identification of ZBTB10 and Three Distinct HLA Associations. <i>Molecular Medicine</i> , 2015, 21, 769-781.	4.4	52
15	Amifampridine phosphate (Firdapse®) is effective and safe in a phase 3 clinical trial in LEMS. <i>Muscle and Nerve</i> , 2016, 53, 717-725.	2.2	51
16	Unaffected patients with a homozygous absence of the SMN1 gene. <i>European Journal of Human Genetics</i> , 2008, 16, 930-934.	2.8	50
17	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. <i>American Journal of Human Genetics</i> , 2018, 103, 817-825.	6.2	40
18	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. <i>Skeletal Muscle</i> , 2018, 8, 23.	4.2	40

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19	Randomized phase 2 trial and open-label extension of domagrozumab in Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , 2020, 30, 492-502.	0.6	40
20	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.. <i>Acta Biochimica Polonica</i> , 2009, 56, .	0.5	37
21	Prevalence and impact of autoimmune thyroid disease on myasthenia gravis course. <i>Brain and Behavior</i> , 2016, 6, e00537.	2.2	33
22	Incidence of Spinal Muscular Atrophy in Poland – More Frequent than Predicted?. <i>Neuroepidemiology</i> , 2010, 34, 152-157.	2.3	32
23	Novel <i>Col12A1</i> variant expands the clinical picture of congenital myopathies with extracellular matrix defects. <i>Muscle and Nerve</i> , 2017, 55, 277-281.	2.2	31
24	Effect of Age and Gender on the Number of Motor Units in Healthy Subjects Estimated by the Multipoint Incremental MUNE Method. <i>Journal of Clinical Neurophysiology</i> , 2014, 31, 272-278.	1.7	29
25	Heme Oxygenase-1 Influences Satellite Cells and Progression of Duchenne Muscular Dystrophy in Mice. <i>Antioxidants and Redox Signaling</i> , 2018, 29, 128-148.	5.4	29
26	Juvenile seropositive myasthenia gravis with anti-MuSK antibody after thymectomy. <i>Journal of Neurology</i> , 2009, 256, 1780-1781.	3.6	28
27	<i>VAV1</i> and <i>BAFF</i> , via <i>NF-κB</i> pathway, are genetic risk factors for myasthenia gravis. <i>Annals of Clinical and Translational Neurology</i> , 2014, 1, 329-339.	3.7	27
28	Novel point mutations in survival motor neuron 1 gene expand the spectrum of phenotypes observed in spinal muscular atrophy patients. <i>Neuromuscular Disorders</i> , 2014, 24, 617-623.	0.6	27
29	Juvenile onset acid maltase deficiency presenting as a rigid spine syndrome. <i>Neuromuscular Disorders</i> , 2006, 16, 282-285.	0.6	23
30	High incidence and clinical characteristics of fibromuscular dysplasia in patients with spontaneous cervical artery dissection: The ARCADIA-POL study. <i>Vascular Medicine</i> , 2019, 24, 112-119.	1.5	23
31	Determinants of Quality of Life in Myasthenia Gravis Patients. <i>Frontiers in Neurology</i> , 2020, 11, 553626.	2.4	23
32	Observation of the natural course of type 3 spinal muscular atrophy: data from the polish registry of spinal muscular atrophy. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 150.	2.7	22
33	Lack of miR-378 attenuates muscular dystrophy in mdx mice. <i>JCI Insight</i> , 2020, 5, .	5.0	22
34	Clinical, electrophysiological, and molecular findings in early onset hereditary neuropathy with liability to pressure palsy. <i>Muscle and Nerve</i> , 2014, 50, 914-918.	2.2	21
35	Motor unit loss estimation by the multipoint incremental MUNE method in children with spinal muscular atrophy – A preliminary study. <i>Neuromuscular Disorders</i> , 2015, 25, 216-221.	0.6	21
36	Identification of GAA variants through whole exome sequencing targeted to a cohort of 606 patients with unexplained limb-girdle muscle weakness. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 173.	2.7	21

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37	The LITAF/SIMPLE I92V sequence variant results in an earlier age of onset of CMT1A/HNPP diseases. <i>Neurogenetics</i> , 2015, 16, 27-32.	1.4	20
38	Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 155.	2.7	19
39	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. <i>Acta Biochimica Polonica</i> , 2009, 56, 103-8.	0.5	18
40	Carpal Tunnel Syndrome in Children. <i>Journal of Child Neurology</i> , 2014, 29, 227-231.	1.4	17
41	Yeast model analysis of novel polymerase gamma variants found in patients with autosomal recessive mitochondrial disease. <i>Human Genetics</i> , 2015, 134, 951-966.	3.8	17
42	Minimal manifestation status and prednisone withdrawal in the MCTX trial. <i>Neurology</i> , 2020, 95, e755-e766.	1.1	17
43	Andersenâ€™Tawil syndrome: Report of 3 novel mutations and high risk of symptomatic cardiac involvement. <i>Muscle and Nerve</i> , 2015, 51, 192-196.	2.2	16
44	L239F founder mutation in GDAP1 is associated with a mild Charcotâ€™Marieâ€™Tooth type 4C4 (CMT4C4) phenotype. <i>Neurogenetics</i> , 2010, 11, 357-366.	1.4	15
45	A form of muscular dystrophy associated with pathogenic variants in JAG2. <i>American Journal of Human Genetics</i> , 2021, 108, 840-856.	6.2	15
46	MLPA based detection of mutations in the dystrophin gene of 180 Polish families with Duchenne/Becker muscular dystrophy. <i>Neurologia I Neurochirurgia Polska</i> , 2014, 48, 416-422.	1.2	14
47	X-linked spinal muscular atrophy (SMA2) caused by de novo c.1731C>T substitution in the UBA1 gene. <i>Neuromuscular Disorders</i> , 2015, 25, 661-666.	0.6	13
48	Dopa-responsive dystonia or early-onset Parkinson disease â€™ Genotypeâ€™phenotype correlation. <i>Neurologia I Neurochirurgia Polska</i> , 2017, 51, 1-6.	1.2	13
49	Exome sequencing reveals mutations in <i>MFN2</i> and <i>GDAP1</i> in severe Charcotâ€™Marieâ€™Tooth disease. <i>Journal of the Peripheral Nervous System</i> , 2014, 19, 242-245.	3.1	12
50	Are electrophysiological criteria useful in distinguishing childhood demyelinating neuropathies?. <i>Journal of the Peripheral Nervous System</i> , 2016, 21, 22-26.	3.1	12
51	Mechanical thrombectomy in acute stroke â€™ Five years of experience in Poland. <i>Neurologia I Neurochirurgia Polska</i> , 2017, 51, 339-346.	1.2	11
52	Treatment outcome in juvenileâ€™onset myasthenia gravis. <i>Muscle and Nerve</i> , 2019, 59, 549-554.	2.2	11
53	Electromyographic findings in sporadic inclusion body myositis. <i>Journal of Electromyography and Kinesiology</i> , 2018, 39, 114-119.	1.7	10
54	Screening for lateâ€™onset Pompe disease in Poland. <i>Acta Neurologica Scandinavica</i> , 2019, 140, 239-243.	2.1	8

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55	Motor Unit Number Index (MUNIX) as a biomarker of motor unit loss in post-polio syndrome versus needle EMG. <i>Journal of Electromyography and Kinesiology</i> , 2019, 46, 35-40.	1.7	8
56	The remarkable phenotypic variability of the p.Arg269His variant in the <i>TRPV4</i> gene. <i>Muscle and Nerve</i> , 2019, 59, 129-133.	2.2	8
57	Pediatric CIDP: Diagnosis and Management. A Single-Center Experience. <i>Frontiers in Neurology</i> , 2021, 12, 667378.	2.4	8
58	Ultra-low radiation dose protocol for CT-guided intrathecal nusinersen injections for patients with spinal muscular atrophy and severe scoliosis. <i>Neuroradiology</i> , 2021, 63, 539-545.	2.2	8
59	Motor unit number estimation as a complementary test to routine electromyography in the diagnosis of amyotrophic lateral sclerosis. <i>Journal of Electromyography and Kinesiology</i> , 2016, 26, 60-65.	1.7	7
60	Abnormal spontaneous activity in primary myopathic disorders. <i>Muscle and Nerve</i> , 2017, 56, 427-432.	2.2	6
61	Spinal muscular atrophy – new therapies, new challenges. <i>Neurologia i Neurochirurgia Polska</i> , 2020, 54, 8-13.	1.2	6
62	Targeted Next-Generation Sequencing Reveals Mutations in Non-coding Regions and Potential Regulatory Sequences of Calpain-3 Gene in Polish Limb Girdle Muscular Dystrophy Patients. <i>Frontiers in Neuroscience</i> , 2021, 15, 692482.	2.8	6
63	Quantitative magnetic resonance imaging measures as biomarkers of disease progression in boys with Duchenne muscular dystrophy: a phase 2 trial of domagrozumab. <i>Journal of Neurology</i> , 2022, 269, 4421-4435.	3.6	6
64	Malignant hyperthermia – what do we know in 2019?. <i>Anaesthesiology Intensive Therapy</i> , 2019, 51, 169-177.	1.0	5
65	Myasthenia Gravis in Poland: National Healthcare Database Epidemiological Study. <i>Neuroepidemiology</i> , 2021, 55, 62-69.	2.3	5
66	Accessory deep peroneal nerve - a clinically significant anomaly?. <i>Neurologia i Neurochirurgia Polska</i> , 2008, 42, 112-5.	1.2	5
67	Myofibrillar myopathy with congenital cataract and skeletal anomalies without mutations in the desmin, β -crystallin, myotilin, LMNA or SEPN1 genes. <i>Neuromuscular Disorders</i> , 2006, 16, 759-762.	0.6	4
68	Original article Charcot-Marie-Tooth type 1C disease coexisting with progressive multiple sclerosis: a study of an overlapping syndrome. <i>Folia Neuropathologica</i> , 2012, 4, 369-374.	1.2	4
69	The needle EMG findings in myotonia congenita. <i>Journal of Electromyography and Kinesiology</i> , 2019, 49, 102362.	1.7	4
70	The frequency of mitochondrial polymerase gamma related disorders in a large Polish population cohort. <i>Mitochondrion</i> , 2019, 47, 179-187.	3.4	4
71	Floppy infant syndrome as a first manifestation of LMNA-related congenital muscular dystrophy. <i>European Journal of Paediatric Neurology</i> , 2021, 32, 115-121.	1.6	4
72	Multimodal data acquisition set for objective assessment of Parkinson's disease. , 2020, , .		4

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73	A Study on the Possible Diagnosis of Parkinson's Disease on the Basis of Facial Image Analysis. <i>Electronics (Switzerland)</i> , 2021, 10, 2832.	3.1	4
74	Facial onset sensory and motor neuronopathy syndrome – a rare variant of motor neurone disease. <i>Neurologia I Neurochirurgia Polska</i> , 2021, 55, 325-327.	1.2	3
75	Myasthenia gravis – treatment and severity in nationwide cohort. <i>Acta Neurologica Scandinavica</i> , 2022, 145, 471-478.	2.1	3
76	Safety, tolerability, and efficacy of a widely available nusinersen program for Polish children with Spinal Muscular Atrophy. <i>European Journal of Paediatric Neurology</i> , 2022, 39, 103-109.	1.6	3
77	Propafenone is not effective for severe ventricular arrhythmias in Andersen-Tawil syndrome. <i>Archives of Medical Science</i> , 2018, 1, 247-250.	0.9	2
78	Spinal muscular atrophy: epidemiology and health burden in children – a Polish national healthcare database perspective before introduction of SMA-specific treatment. <i>Neurologia I Neurochirurgia Polska</i> , 2021, 55, 479-484.	1.2	2
79	Response to letter: A decision for life – Treatment decisions in newly diagnosed families with spinal muscular atrophy. <i>European Journal of Paediatric Neurology</i> , 2021, 30, 103-104.	1.6	1
80	Comprehensive non-invasive assessment of electrocardiographic abnormalities and cardiac arrhythmias in patients with genetically confirmed mitochondrial diseases. <i>Journal of Electrocardiology</i> , 2021, 65, 136-142.	0.9	1
81	Lower BAFF Levels in Myasthenic Patients Treated with Glucocorticoids. <i>Archivum Immunologiae Et Therapiae Experimentalis</i> , 2021, 69, 22.	2.3	1
82	Transthyretin-related familial amyloid polyneuropathy (ATTR-FAP) in Poland – genetic and clinical presentation. <i>Neurologia I Neurochirurgia Polska</i> , 2020, 54, 552-560.	1.2	1
83	Progressive External Ophthalmoplegia in Polish Patients – From Clinical Evaluation to Genetic Confirmation. <i>Genes</i> , 2021, 12, 54.	2.4	1
84	Selected problems of image data preprocessing used to perform examination in Parkinson's disease. , 2020, , .		1
85	Spinal muscular atrophy with an overlapping syndrome – double trouble or a potentially better outcome?. <i>Neurologia I Neurochirurgia Polska</i> , 2020, 54, 475-477.	1.2	1
86	Application of a rapid non-invasive technique in the molecular diagnosis of spinal muscular atrophy (SMA). <i>Neurologia I Neurochirurgia Polska</i> , 2005, 39, 89-94.	1.2	1
87	Gross Motor Function Disorders in Patients with Alternating Hemiplegia of Childhood. <i>Medycyna Wiekii Rozwojowego</i> , 2020, 24, 24-32.	0.2	1
88	Parametric analysis of pilot voice signals in Parkinson's disease diagnostics. <i>Journal of Automation Electronics and Electrical Engineering</i> , 2022, 4, 21-28.	0.2	1
89	Evidence for a relatively high proportion of DM2 mutations in a large group of Polish patients. <i>Neurologia I Neurochirurgia Polska</i> , 2018, 52, 736-742.	1.2	0
90	Serum interleukin 15 levels in patients with seropositive myasthenia gravis do not correlate with disease severity. <i>Neurologia I Neurochirurgia Polska</i> , 2018, 52, 364-367.	1.2	0

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91	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
92	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
93	A review of functional assessment scales in non-sitters with spinal muscular atrophy (SMA)., 2018, 27, 11-17.		0
94	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects. , 2021, 52, .		0
95	The use of non-linear acoustic analysis to objectively evaluate the voice of people with Parkinson's disease. , 2020, , .		0
96	Multi-minicore myopathy: a clinical and histopathological study of 17 cases. Folia Neuropathologica, 2007, 45, 56-65.	1.2	0
97	Oculopharyngeal muscular dystrophy: phenotypic and genotypic characteristics of 9 Polish patients. Neurologia I Neurochirurgia Polska, 2009, 43, 113-20.	1.2	0
98	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