Aleksandra Jezela-Stanek

List of Publications by Year
in descending order

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80	835	567281 15	610901 24
papers	citations	h-index	g-index

89	89	89	1452
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
2	Congenital Disorders of Glycosylation from a Neurological Perspective. Brain Sciences, 2021, 11, 88.	2.3	53
3	Congenital disorder of glycosylphosphatidylinositol (GPI)-anchor biosynthesisâ€"The phenotype of two patients with novel mutations in the PIGN and PGAP2 genes. European Journal of Paediatric Neurology, 2016, 20, 462-473.	1.6	42
4	Infectious and Immunologic Phenotype of MECP2 Duplication Syndrome. Journal of Clinical Immunology, 2015, 35, 168-181.	3.8	35
5	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
6	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
7	Differences between predicted and established diagnoses of Smithâ€Lemliâ€Opitz syndrome in the Polish population: underdiagnosis or loss of affected fetuses?. Journal of Inherited Metabolic Disease, 2010, 33, 241-248.	3.6	29
8	DHCR7 mutations and genotype-phenotype correlation in 37 Polish patients with Smith-Lemli-Opitz syndrome. Clinical Genetics, 2004, 66, 517-524.	2.0	25
9	Post-Translational Modifications of Circulating Alpha-1-Antitrypsin Protein. International Journal of Molecular Sciences, 2020, 21, 9187.	4.1	25
10	Genetic causes of syndromic craniosynostoses. European Journal of Paediatric Neurology, 2013, 17, 221-224.	1.6	24
11	Fucosidosisâ€"Clinical Manifestation, Long-Term Outcomes, and Genetic Profileâ€"Review and Case Series. Genes, 2020, 11, 1383.	2.4	22
12	Clinical, biochemical and molecular phenotype of congenital disorders of glycosylation: long-term follow-up. Orphanet Journal of Rare Diseases, 2021, 16, 17.	2.7	22
13	Maternal urinary steroid profiles in prenatal diagnosis of Smith-Lemli-Opitz syndrome: first patient series comparing biochemical and molecular studies. Clinical Genetics, 2005, 69, 77-85.	2.0	19
14	Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. Journal of Neurology, 2019, 266, 2657-2664.	3.6	19
15	Angelman Syndrome Revisited. Neurologist, 2007, 13, 305-312.	0.7	17
16	Mild Smith-Lemli-Opitz syndrome: Further delineation of 5 Polish cases and review of the literature. European Journal of Medical Genetics, 2008, 51, 124-140.	1.3	15
17	The phenotypeâ€driven computational analysis yields clinical diagnosis for patients with atypical manifestations of known intellectual disability syndromes. Molecular Genetics & Genomic Medicine, 2020, 8, e1263.	1.2	15
18	Four novel RSK2 mutations in females with Coffinâ€"Lowry syndrome. European Journal of Medical Genetics, 2010, 53, 268-273.	1.3	14

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19	Additional data on the clinical phenotype of Helsmoortelâ€"Van der Aa syndrome associated with a novel truncating mutation in ADNP gene. American Journal of Medical Genetics, Part A, 2016, 170, 1647-1650.	1.2	14
20	Epilepsy in Mitochondrial Diseasesâ€"Current State of Knowledge on Aetiology and Treatment. Children, 2021, 8, 532.	1.5	14
21	Malan syndrome (Sotos syndrome 2) in two patients with 19p13.2 deletion encompassing NFIX gene and novel NFIX sequence variant. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2016, 160, 161-167.	0.6	13
22	A Novel PANK2 Gene Mutation: Clinical and Molecular Characteristics of Patientsâ€"Short Communication. Journal of Child Neurology, 2007, 22, 1256-1259.	1.4	11
23	Is diagnosing cardio-facio-cutaneous (CFC) syndrome still a challenge? Delineation of the phenotype in 15 Polish patients with proven mutations, including novel mutations in the BRAF gene. European Journal of Medical Genetics, 2015, 58, 14-20.	1.3	11
24	X-linked Î± thalassaemia/mental retardation syndrome: a case with gonadal dysgenesis, caused by a novel mutation in ATRX gene. Clinical Dysmorphology, 2009, 18, 168-171.	0.3	10
25	Neuropathophysiology, Genetic Profile, and Clinical Manifestation of Mucolipidosis IVâ€"A Review and Case Series. International Journal of Molecular Sciences, 2020, 21, 4564.	4.1	10
26	Cryptic X; Autosome Translocation in a Boyâ€"Delineation of the Phenotype. Pediatric Neurology, 2011, 44, 221-224.	2.1	9
27	Pulmonary involvement in selected lysosomal storage diseases and the impact of enzyme replacement therapy: A stateâ€ofâ€the art review. Clinical Respiratory Journal, 2020, 14, 422-429.	1.6	9
28	Nonimmune Hydrops Fetalisâ€"Prenatal Diagnosis, Genetic Investigation, Outcomes and Literature Review. Journal of Clinical Medicine, 2020, 9, 1789.	2.4	9
29	Beyond thelungs: Alpha-1 antitrypsinâ€™s potential role in human gestation. Advances in Clinical and Experimental Medicine, 2019, 28, 1257-1261.	1.4	9
30	Novel data on growth phenotype and causative genotypes in 29 patients with Morquio (Morquio-Brailsford) syndrome from Central-Eastern Europe. Journal of Applied Genetics, 2019, 60, 163-174.	1.9	8
31	The remarkable phenotypic variability of the p.Arg269HiS variant in the TRPV4 gene. Muscle and Nerve, 2019, 59, 129-133.	2.2	8
32	CHILD syndrome: clinical picture and diagnostic procedures. Journal of the European Academy of Dermatology and Venereology, 2007, 21, 070209222700152-???.	2.4	7
33	Trisomy 22pter-q12.3 presenting with hepatic dysfunction variability of cat-eye syndrome. Clinical Dysmorphology, 2009, 18, 13-17.	0.3	7
34	Minimal clinical findings in a patient with 15qter microdeletion syndrome: Delineation of the associated phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 922-926.	1.2	7
35	1.15â€‰Mb microdeletion in chromosome band 20p13 associated with moderate developmental delayâ€"Additional case and data's review. American Journal of Medical Genetics, Part A, 2013, 161, 172-178.	1.2	7
36	Novel COL12A1 variant as a cause of mild familial extracellular matrixâ€related myopathy. Clinical Genetics, 2019, 95, 736-738.	2.0	7

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37	Long Term Follow-Up of Polish Patients with Isovaleric Aciduria. Clinical and Molecular Delineation of Isovaleric Aciduria. Diagnostics, 2020, 10, 738.	2.6	7
38	Evidence of the milder phenotypic spectrum of c. 1582G >A PIGT variant: Delineation based on seven novel Polish patients. Clinical Genetics, 2020, 98, 468-476.	2.0	7
39	The first case of a patient with de novo partial distal 16q tetrasomy and a data's review. American Journal of Medical Genetics, Part A, 2014, 164, 2541-2550.	1.2	6
40	FGF12p.Gly112Ser variant as a cause of phenytoin/phenobarbital responsive epilepsy. Clinical Genetics, 2019, 96, 274-275.	2.0	6
41	Novel variant in HDAC8 gene resulting in the severe Cornelia de Lange phenotype. Clinical Dysmorphology, 2019, 28, 124-128.	0.3	6
42	Mild phenotype of glutaric aciduria type 1 in polish patients â€“ novel data from a group of 13 cases. Metabolic Brain Disease, 2019, 34, 641-649.	2.9	6
43	Possible effect of the HLA-DQ2/DQ8 polymorphism on autoimmune parameters and lymphocyte subpopulation in recurrent pregnancy losses. Journal of Reproductive Immunology, 2022, 149, 103467.	1.9	6
44	Skeletal and Bone Mineral Density Features, Genetic Profile in Congenital Disorders of Glycosylation: Review. Diagnostics, 2021, 11, 1438.	2.6	5
45	Infantile Alexander disease with late onset infantile spasms and hypsarrhythmia. Balkan Journal of Medical Genetics, 2019, 22, 77-82.	0.5	5
46	History and molecular characteristics of a patient with terminal deletion of 14q. Is this another syndrome with a striking phenotype?. Clinical Dysmorphology, 2012, 21, 97-100.	0.3	4
47	Vertical nystagmus as a feature of PIGN-related glycosylphosphatidylinositol biosynthesis defects. Clinical Neurology and Neurosurgery, 2020, 196, 106033.	1.4	4
48	The First Metabolome Analysis in Children with Epilepsy and ALG13-CDG Resulting from c.320A>G Variant. Children, 2021, 8, 251.	1.5	4
49	Diaphragmatic Hernia as a Prenatal Feature of Glycosylphosphatidylinositol Biosynthesis Defects and the Overlap With Fryns Syndrome â€“ Literature Review. Frontiers in Genetics, 2021, 12, 674722.	2.3	4
50	Case Report: Blepharophimosis and Ptosis as Leading Dysmorphic Features of Rare Congenital Malformation Syndrome With Developmental Delay â€“ New Cases With TRAF7 Variants. Frontiers in Medicine, 2021, 8, 708717.	2.6	4
51	GC-MS as a tool for reliable non-invasive prenatal diagnosis of Smith-Lemli-Opitz syndrome but essential also for other cholesterolopathies verification. Ginekologia Polska, 2020, 91, 287-293.	0.7	4
52	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
53	Tetraploidy in the era of molecular karyotyping â€“ What we need to remember. Pediatria Polska, 2013, 88, 467-471.	0.2	3
54	Proteins Structure Models in the Evaluation of Novel Variant (C.472_477del) in the MOCS2 Gene. Diagnostics, 2020, 10, 821.	2.6	3

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55	Molecular Background and Disease Prevalence of Biotinidase Deficiency in a Polish Populationâ€"Data Based on the National Newborn Screening Programme. Genes, 2022, 13, 802.	2.4	3
56	Alpha-1 Antitrypsin Z Variant (AAT PI*Z) as a Risk Factor for Intrahepatic Cholestasis of Pregnancy. Frontiers in Genetics, 2021, 12, 720465.	2.3	2
57	Trends in prenatal diagnosis of non-specific multiple malformations disorders with reference to the own experience and research study on Smith-Lemli-Opitz syndrome. Ginekologia Polska, 2015, 86, 598-602.	0.7	2
58	Oculocutaneous albinism in a patient with 17p13.2-pter duplication - a review on the molecular syndromology of 17p13 duplication. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2015, 159, 333-337.	0.6	2
59	The neuropathological findings of developmental and epileptic encephalopathy-43 (DEE43) and delineation of a the molecular spectrum of novel case. Seizure: the Journal of the British Epilepsy Association, 2021, 93, 75-80.	2.0	2
60	Clinical expression of Holt-Oram syndrome on the basis of own clinical experience considering prenatal diagnosis. Ginekologia Polska, 2016, 87, 706-710.	0.7	2
61	Spectrum of Neurological Symptoms in Glycosylphosphatidylinositol Biosynthesis Defects: Systematic Review. Frontiers in Neurology, 2021, 12, 758899.	2.4	2
62	Novel pathogenic variant in the HRAS gene with lethal outcome and a broad phenotypic spectrum among Polish patients with Costello syndrome. Clinical Dysmorphology, 2017, 26, 83-90.	0.3	1
63	Is leucodystrophy really a feature of PIGTâ€CDG?. Neuropathology and Applied Neurobiology, 2020, 46, 615-616.	3.2	1
64	Anthropometric characteristics of 65 Polish Smith-Lemli-Opitz patients. Journal of Applied Genetics, 2021, 62, 469-475.	1.9	1
65	The genetic basis of classical galactosaemia in Polish patients. Orphanet Journal of Rare Diseases, 2021, 16, 239.	2.7	1
66	Do Not Miss the (Genetic) Diagnosis of Gaucher Syndrome: A Narrative Review on Diagnostic Clues and Management in Severe Prenatal and Perinatal-Lethal Sporadic Cases. Journal of Clinical Medicine, 2021, 10, 4890.	2.4	1
67	Diverse clinical outcome of Hunter syndrome in patients with chromosomal aberration encompassing entire and partial IDS deletions: what is important for early diagnosis and counseling?. Clinical Dysmorphology, 2021, 30, 76-82.	0.3	1
68	Polish activity within Orphanet Europe--state of art of database and services. Medycyna Wieku Rozwojowego, 2015, 19, 536-41.	0.2	1
69	Structural Analysis of the Effect of Asn107Ser Mutation on Alg13 Activity and Alg13-Alg14 Complex Formation and Expanding the Phenotypic Variability of ALG13-CDG. Biomolecules, 2022, 12, 398.	4.0	1
70	Interstitial Lung Disease in Rare Congenital Syndromes. Medycyna Wieku Rozwojowego, 2020, 24, 47-52.	0.2	1
71	WspÃ³Å‚praca genetyka klinicznego i biologa molekularnego â€“ wczoraj i dziÅ›. Pediatria Polska, 2015, 90, 171-180.	0.2	0
72	Broad clinical spectrum observed in patients with scapuloperoneal spinal muscular atrophy (SPSMA) caused by an c.806Gâ€‰›â€‰A (p. Arg269His) mutation in the TRPV4 gene. Neuromuscular Disorders, 2017, 27, S135.	0.6	0

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73	Genetics of fetal growth restrictionâ€"Isolated is not syndromic. Prenatal Diagnosis, 2021, 41, 6-7.	2.3	0
74	Review of neurological aspects in a 3-month-old boy with Ehlers-Danlos syndrome (EDS) â€“ case report. , 2018, 27, 75-78.		0
75	Genetic counselling. , 2019, , 143-149.		0
76	How does terminal 21q22 deletion really manifest? Delineation based on prenatal diagnosis and literature review. Taiwanese Journal of Obstetrics and Gynecology, 2021, 60, 1121-1125.	1.3	0
77	Noninvasive prenatal testing of aneuploidies: where are we now?. Revista Brasileira De Ginecologia E Obstetricia, 2014, 36, 383-6.	0.8	0
78	Editorial: Inherited Protein Glycosylation Defects in Humans. Frontiers in Genetics, 2022, 13, 851438.	2.3	0
79	Dear Readers and Authors. Medycyna Wieku Rozwojowego, 2021, 24, 1.	0.2	0
80	Editorial. Medycyna Wieku Rozwojowego, 2021, 25, 1.	0.2	0