Agnieszka Pollak

List of Publications by Year
in descending order

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68 papers	1,253 citations	430754 18 h-index	395590 33 g-index
71 all docs	71 docs citations	71 times ranked	2782 citing authors

#	Article	IF	Citations
1	Postzygotic mosaicism of a novel PTPN11 mutation in monozygotic twins discordant for metachondromatosis. American Journal of Medical Genetics, Part A, 2022, , .	0.7	3
2	CD8+ T-Cell Exhaustion Phenotype in Chronic Hepatitis C Virus Infection Is Associated With Epitope Sequence Variation. Frontiers in Immunology, 2022, 13, 832206.	2.2	4
3	Severe Infantile Axonal Neuropathy with Respiratory Failure Caused by Novel Mutation in X-Linked LAS1L Gene. Genes, 2022, 13, 725.	1.0	1
4	De novo stop-loss variants in CLDN11 cause hypomyelinating leukodystrophy. Brain, 2021, 144, 411-419.	3.7	12
5	The Role of the Reanalysis of Genetic Test Results in the Diagnosis of Dysmorphic Syndrome Caused by Inherited Xq24 Deletion including the UBE2A and CXorf56 Genes. Genes, 2021, 12, 350.	1.0	4
6	Further Delineation of Phenotype and Genotype of Primary Microcephaly Syndrome with Cortical Malformations Associated with Mutations in the WDR62 Gene. Genes, 2021, 12, 594.	1.0	5
7	Biallelic variants in TMEM222 cause a new autosomal recessive neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 1246-1254.	1.1	5
8	18-Year-old patient with Klinefelter syndrome (47, XXY) and complete androgen insensitivity syndrome (CAIS) â€“ case report. Gynecological Endocrinology, 2021, 37, 572-575.	0.7	0
9	Prenatal Versus Postnatal Diagnosis of Meckelâ€“Gruber and Joubert Syndrome in Patients with TMEM67 Mutations. Genes, 2021, 12, 1078.	1.0	2
10	A recurrent de novo variant supports <scp>KCNC2</scp> involvement in the pathogenesis of developmental and epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2021, 185, 3384-3389.	0.7	15
11	Mosaic IL6ST variant inducing constitutive GP130 cytokine receptor signaling as a cause of neonatal onset immunodeficiency with autoinflammation and dysmorphy. Human Molecular Genetics, 2021, 30, 226-233.	1.4	8
12	Developmental delay with hypotrophy associated with homozygous functionally relevant REV3L variant. Journal of Molecular Medicine, 2021, 99, 415-423.	1.7	3
13	Celiac Disease in Conjunction with Hereditary Fructose Intolerance as a Rare Cause of Liver Steatosis with Mild Hypertransaminasemiaâ€”A Case Report. Pediatric Reports, 2021, 13, 589-593.	0.5	1
14	Sensorineural hearing loss in GSD type I patients. A newly recognized symptomatic association of potential clinical significance and unclear pathomechanism. International Journal of Pediatric Otorhinolaryngology, 2021, 151, 110970.	0.4	0
15	Results of Polish Adult Leukemia Study Group (PALG) project assessing TP53 mutations with next-generation sequencing technology in relapsed and refractory chronic lymphocytic leukemia patients â€” an 18-month update. Acta Haematologica Polonica, 2021, 52, 94-102.	0.1	0
16	Peritoneal dialysis in an adult patient with tetralogy of Fallot diagnosed with incomplete Alagille syndrome. BMC Medical Genetics, 2020, 21, 195.	2.1	0
17	Leukoencephalopathy with Calcifications and Cystsâ€”The First Polish Patient with Labrune Syndrome. Brain Sciences, 2020, 10, 869.	1.1	3
18	Targeted Next-Generation Sequencing in Diagnostic Approach to Monogenic Cholestatic Liver Disordersâ€”Single-Center Experience. Frontiers in Pediatrics, 2020, 8, 414.	0.9	17

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19	Phenotypic expansion in Zhuâ€Tokitaâ€Takenouchiâ€Kim syndrome caused by de novo variants in the SON gene. Molecular Genetics & Genomic Medicine, 2020, 8, e1432.	0.6	17
20	AP4B1-associated hereditary spastic paraplegia: expansion of phenotypic spectrum related to homozygous p.Thr387fs variant. Journal of Applied Genetics, 2020, 61, 213-218.	1.0	6
21	Breakpoint Mapping of Symptomatic Balanced Translocations Links the EPHA6, KLF13 and UBR3 Genes to Novel Disease Phenotype. Journal of Clinical Medicine, 2020, 9, 1245.	1.0	4
22	Maternally inherited diabetes and deafness (MIDD) syndrome with m.3243A>G mutation associated with renal failure â€" a case report. Clinical Diabetology, 2020, 9, 475-478.	0.2	0
23	First familial cases of type 2 congenital erythrocytosis (ECYT2) with a Chuvash pathogenic variant in VHL gene in Poland: example of the clinical utility of next-generation sequencing in diagnostics of orphan diseases. Acta Haematologica Polonica, 2020, 51, 220-225.	0.1	0
24	Overinterpretation of high throughput sequencing data in medical genetics: first evidence against TMPRSS3/GJB2 digenic inheritance of hearing loss. Journal of Translational Medicine, 2019, 17, 269.	1.8	11
25	Dominant ELOVL1 mutation causes neurological disorder with ichthyotic keratoderma, spasticity, hypomyelination and dysmorphic features. Journal of Medical Genetics, 2018, 55, 408-414.	1.5	41
26	Phenotypic consequences of gene disruption by a balanced de novo translocation involving SLC6A1 and NAA15. European Journal of Medical Genetics, 2018, 61, 596-601.	0.7	2
27	Clinical and molecular characteristics of newly reported mitochondrial disease entity caused by biallelic PARS2 mutations. Journal of Human Genetics, 2018, 63, 473-485.	1.1	19
28	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	2.6	40
29	Human Pegivirus in Patients with Encephalitis of Unclear Etiology, Poland. Emerging Infectious Diseases, 2018, 24, 1785-1794.	2.0	15
30	A 23â€year followâ€up of a male with Hajduâ€Cheney syndrome due to NOTCH2 mutation. American Journal of Medical Genetics, Part A, 2018, 176, 2382-2388.	0.7	3
31	Genetic Basis of Hearing Loss. , 2018, , .		0
32	Tinnitus in patients with hearing loss due to mitochondrial DNA pathogenic variants. European Archives of Oto-Rhino-Laryngology, 2018, 275, 1979-1985.	0.8	8
33	Developmental epileptic encephalopathy with hypomyelination and brain atrophy associated with PTPN23 variants affecting the assembly of UsnRNPs. European Journal of Human Genetics, 2018, 26, 1502-1511.	1.4	8
34	Next-generation sequencing analysis of new genotypes appearing during antiviral treatment of chronic hepatitis C reveals that these are selected from pre-existing minor strains. Journal of General Virology, 2018, 99, 1633-1642.	1.3	4
35	Novel neuro-audiological findings and further evidence for TWNK involvement in Perrault syndrome. Journal of Translational Medicine, 2017, 15, 25.	1.8	36
36	Homozygous truncating mutation in NRAP gene identified by whole exome sequencing in a patient with dilated cardiomyopathy. Scientific Reports, 2017, 7, 3362.	1.6	30

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37	Isolated Hearing Impairment Caused by SPATA5 Mutations in a Family with Variable Phenotypic Expression. Advances in Experimental Medicine and Biology, 2017, 980, 59-66.	0.8	9
38	Coexistence of mutations in keratin 10 (KRT10) and the mitochondrial genome in a patient with ichthyosis with confetti and Leber's hereditary optic neuropathy. American Journal of Medical Genetics, Part A, 2017, 173, 3093-3097.	0.7	7
39	Iterative Sequencing and Variant Screening (ISVS) as a novel pathogenic mutations search strategy - application for TMPRSS3 mutations screen. Scientific Reports, 2017, 7, 2543.	1.6	10
40	Advances in genetic hearing loss: CIB2 gene. European Archives of Oto-Rhino-Laryngology, 2017, 274, 1791-1795.	0.8	10
41	Application of nextâ€‘generation sequencing to identify mitochondrial mutations: Study on m.7511T>C in patients with hearing loss. Molecular Medicine Reports, 2017, 17, 1782-1790.	1.1	3
42	Pathology of mitochondria in MELAS syndrome: an ultrastructural study. Polish Journal of Pathology, 2017, 2, 173-181.	0.1	8
43	Titin Truncating Variants in Dilated Cardiomyopathy â€“ Prevalence and Genotype-Phenotype Correlations. PLoS ONE, 2017, 12, e0169007.	1.1	63
44	Whole exome sequencing identifies TRIOBP pathogenic variants as a cause of post-lingual bilateral moderate-to-severe sensorineural hearing loss. BMC Medical Genetics, 2017, 18, 142.	2.1	15
45	New perspective in diagnostics of mitochondrial disorders: two yearsâ€™ experience with whole-exome sequencing at a national paediatric centre. Journal of Translational Medicine, 2016, 14, 174.	1.8	176
46	Evidence for troponin C (TNNC1) as a gene for autosomal recessive restrictive cardiomyopathy with fatal outcome in infancy. American Journal of Medical Genetics, Part A, 2016, 170, 3241-3248.	0.7	37
47	Further evidence for GRIN2B mutation as the cause of severe epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2016, 170, 3265-3270.	0.7	22
48	Pathogenic p.Cys194Metfs*17 variant argues against TMPRSS3/GJB2 digenic inheritance of hearing loss. European Archives of Oto-Rhino-Laryngology, 2016, 273, 1327-1328.	0.8	2
49	Novel and De Novo Mutations Extend Association of POU3F4 with Distinct Clinical and Radiological Phenotype of Hearing Loss. PLoS ONE, 2016, 11, e0166618.	1.1	20
50	A note of caution on the diagnosis of Martin-Probst syndrome by the detection of the p.D59G mutation in the RAB40AL gene. European Journal of Pediatrics, 2015, 174, 693-696.	1.3	5
51	Next-generation sequencing (NGS) in the identification of encephalitis-causing viruses: Unexpected detection of human herpesvirus 1 while searching for RNA pathogens. Journal of Virological Methods, 2015, 226, 1-6.	1.0	54
52	Audio Profiles in Mitochondrial Deafness m.1555A>G and m.3243A>G Show Distinct Differences. Medical Science Monitor, 2015, 21, 694-700.	0.5	12
53	Nietypowy przebieg niewydolnoÅ›ci wielogruczoÅ‚owej z wspÃ³Å‚istniejÄ…cymi mutacjami genu AIRE u 18-letniej dziewczynki â€”12-letnia obserwacja. Endokrynologia Polska, 2015, 65, 514-518.	0.3	0
54	Does p.Q247X in TRIM63 Cause Human Hypertrophic Cardiomyopathy?. Circulation Research, 2014, 114, e2-5.	2.0	88

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55	Evidence AgainstRAB40ALBeing the Locus for Martin-Probst X-Linked Deafness-Intellectual Disability Syndrome. Human Mutation, 2014, 35, 1171-1174.	1.1	10
56	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.	1.4	12
57	MTHFR 677T Is a Strong Determinant of the Degree of Hearing Loss Among Polish Males with Postlingual Sensorineural Hearing Impairment. DNA and Cell Biology, 2012, 31, 1267-1273.	0.9	7
58	Transferrin isoelectric focusing and plasma lysosomal enzyme activities in the diagnosis and followâ€up of hereditary fructose intolerance. Clinica Chimica Acta, 2012, 413, 1714-1715.	0.5	4
59	Postlingual Hearing Loss as a Mitochondrial 3243A>G Mutation Phenotype. PLoS ONE, 2012, 7, e44054.	1.1	19
60	The contribution of the mitochondrial COI/tRNASer(UCN) gene mutations to non-syndromic and aminoglycoside-induced hearing loss in Polish patients. Molecular Genetics and Metabolism, 2011, 104, 153-159.	0.5	12
61	Novel keratin 14 hotspot mutation in Dowling-Meara type of epidermolysis bullosa simplex: Strategy to avoid KRT14 pseudogene amplification by a simple approach. Journal of Dermatological Science, 2010, 57, 69-70.	1.0	7
62	Mutation analysis of mitochondrial 12S rRNA gene in Polish patients with non-syndromic and aminoglycoside-induced hearing loss. Biochemical and Biophysical Research Communications, 2010, 395, 116-121.	1.0	47
63	Phenotypic variability of patients homozygous for the GJB2 mutation 35delG cannot be explained by the influence of one major modifier gene. European Journal of Human Genetics, 2009, 17, 517-524.	1.4	46
64	GJB2 and hearing impairment: promoter defects do not explain the excess of monoallelic mutations. Journal of Medical Genetics, 2008, 45, 607-608.	1.5	16
65	Genetics of Meesmann corneal dystrophy: a novel mutation in the keratin 3 gene in an asymptomatic family suggests genotype-phenotype correlation. Molecular Vision, 2008, 14, 1713-8.	1.1	22
66	M34T and V37I mutations in GJB2 associated hearing impairment: Evidence for pathogenicity and reduced penetrance. American Journal of Medical Genetics, Part A, 2007, 143A, 2534-2543.	0.7	92
67	Aldolase B mutations and prevalence of hereditary fructose intolerance in a Polish population. Molecular Genetics and Metabolism, 2006, 87, 376-378.	0.5	21
68	Molecular background of polyendocrinopathy-candidiasis-ectodermal dystrophy syndrome in a Polish population: novel AIRE mutations and an estimate of disease prevalence. Clinical Genetics, 2006, 70, 348-354.	1.0	69