Rafal Ploski

List of Publications by Year in descending order

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82 papers	4,524 citations	136950 32 h-index	106344 65 g-index
82 all docs	82 docs citations	82 times ranked	6967 citing authors

#	Article	IF	Citations
1	A Neurodevelopmental Disorder With Dystonia and Chorea Resulting From Clustering <scp>CAMK4</scp> Variants. Movement Disorders, 2021, 36, 520-521.	3.9	5
2	Characterization of Ocular Surface Microbial Profiles Revealed Discrepancies between Conjunctival and Corneal Microbiota. Pathogens, 2021, 10, 405.	2.8	22
3	iPSC-derived myelinoids to study myelin biology of humans. Developmental Cell, 2021, 56, 1346-1358.e6.	7.0	34
4	Variants of ATP1A3 in residue 756 cause a separate phenotype of relapsing encephalopathy with cerebellar ataxia (RECA)â€"Report of two cases and literature review. Molecular Genetics & Genomic Medicine, 2021, 9, e1772.	1.2	9
5	Changes in Nuclear Gene Expression Related to Mitochondrial Function Affect Extracellular Matrix, Collagens, and Focal Adhesion in Keratoconus. Translational Vision Science and Technology, 2021, 10, 6.	2.2	5
6	Whole exome sequencing identifies a homozygous POLG2 missense variant in an adult patient presenting with optic atrophy, movement disorders, premature ovarian failure and mitochondrial DNA depletion. European Journal of Medical Genetics, 2020, 63, 103821.	1.3	5
7	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
8	Peritoneal dialysis in an adult patient with tetralogy of Fallot diagnosed with incomplete Alagille syndrome. BMC Medical Genetics, 2020, 21, 195.	2.1	0
9	Phenotypic expansion in Zhuâ€Tokitaâ€Takenouchiâ€Kim syndrome caused by de novo variants in the SON gene. Molecular Genetics & Genomic Medicine, 2020, 8, e1432.	1.2	17
10	Can Circulating Cardiac Biomarkers Be Helpful in the Assessment of LMNA Mutation Carriers?. Journal of Clinical Medicine, 2020, 9, 1443.	2.4	9
11	Identification and characterization of novel rapidly mutating Yâ€chromosomal short tandem repeat markers. Human Mutation, 2020, 41, 1680-1696.	2.5	33
12	Analysis of De Novo Mutations in Sporadic Cardiomyopathies Emphasizes Their Clinical Relevance and Points to Novel Candidate Genes. Journal of Clinical Medicine, 2020, 9, 370.	2.4	12
13	A Novel CDC42 Mutation in an 11-Year Old Child Manifesting as Syndromic Immunodeficiency, Autoinflammation, Hemophagocytic Lymphohistiocytosis, and Malignancy: A Case Report. Frontiers in Immunology, 2020, 11, 318.	4.8	31
14	Accumulation of sequence variants in genes of Wnt signaling and focal adhesion pathways in human corneas further explains their involvement in keratoconus. PeerJ, 2020, 8, e8982.	2.0	12
15	Puzzling outcome of the nationwide genetic survey of severe/moderate female haemophilia B in Poland. Haemophilia, 2019, 25, e373-e376.	2.1	3
16	Multiple Differentially Methylated Regions Specific to Keratoconus Explain Known Keratoconus Linkage Loci. , 2019, 60, 1501.		15
17	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	6.2	30
18	Novel de novo mutation affecting two adjacent aminoacids in the EED gene in a patient with Weaver syndrome. Journal of Human Genetics, 2018, 63, 517-520.	2.3	16

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19	Perforin gene variation influences survival in childhood acute lymphoblastic leukemia. Leukemia Research, 2018, 65, 29-33.	0.8	4
20	Dilated Cardiomyopathy DueÂtoÂBLC2-Associated AthanogeneÂ3Â(BAG3)ÂMutations. Journal of the American College of Cardiology, 2018, 72, 2471-2481.	2.8	93
21	Homozygous mutation in the Neurofascin gene affecting the glial isoform of Neurofascin causes severe neurodevelopment disorder with hypotonia, amimia and areflexia. Human Molecular Genetics, 2018, 27, 3669-3674.	2.9	34
22	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.	2.8	8
23	Evaluation of electrocardiographic parameters in patients with hearing loss genotyped for the connexin 26 gene (GJB2) mutations. Brazilian Journal of Otorhinolaryngology, 2017, 83, 176-182.	1.0	3
24	Collagen synthesis disruption and downregulation of core elements of TGF-Î², Hippo, and Wnt pathways in keratoconus corneas. European Journal of Human Genetics, 2017, 25, 582-590.	2.8	70
25	Coâ€œoccurrence of Jalili syndrome and muscular overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 2280-2283.	1.2	5
26	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.	1.2	7
27	Iterative Sequencing and Variant Screening (ISVS) as a novel pathogenic mutations search strategy - application for TMPRSS3 mutations screen. Scientific Reports, 2017, 7, 2543.	3.3	10
28	Titin Truncating Variants in Dilated Cardiomyopathy â€“ Prevalence and Genotype-Phenotype Correlations. PLoS ONE, 2017, 12, e0169007.	2.5	63
29	The 4q25, 1q21, and 16q22 polymorphisms and recurrence of atrial fibrillation after pulmonary vein isolation. Archives of Medical Science, 2016, 1, 38-44.	0.9	19
30	Differences in Gene-Gene Interactions in Gravesâ€™ Disease Patients Stratified by Age of Onset. PLoS ONE, 2016, 11, e0150307.	2.5	11
31	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.	1.2	37
32	Further evidence for GRIN2B mutation as the cause of severe epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2016, 170, 3265-3270.	1.2	22
33	The 9p21 polymorphism is linked with atrial fibrillation during acute phase of ST-segment elevation myocardial infarction. Heart and Vessels, 2016, 31, 1590-1594.	1.2	3
34	Haemophilia A and cardiovascular morbidity in a female SHAM syndrome carrier due to skewed X chromosome inactivation. European Journal of Medical Genetics, 2016, 59, 43-47.	1.3	11
35	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
36	HLA DQ2 Haplotype, Early Onset of Graves Disease, and Positive Family History of Autoimmune Disorders are Risk Factors for Developing Celiac Disease in Patients with Graves Disease. Endocrine Practice, 2015, 21, 993-1000.	2.1	4

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37	The Role of Recent Admixture in Forming the Contemporary West Eurasian Genomic Landscape. Current Biology, 2015, 25, 2518-2526.	3.9	68
38	Association between Polymorphisms in the TSHR Gene and Graves' Orbitopathy. PLoS ONE, 2014, 9, e102653.	2.5	20
39	Polymorphism of 9p21.3 Locus Is Associated with 5-Year Survival in High-Risk Patients with Myocardial Infarction. PLoS ONE, 2014, 9, e104635.	2.5	12
40	Does p.Q247X in TRIM63 Cause Human Hypertrophic Cardiomyopathy?. Circulation Research, 2014, 114, e2-5.	4.5	88
41	The BAG3 gene variants in Polish patients with dilated cardiomyopathy: four novel mutations and a genotype-phenotype correlation. Journal of Translational Medicine, 2014, 12, 192.	4.4	81
42	Polish population data on 15 autosomal STRs of AmpFlSTR NGM PCR kit. Forensic Science International: Genetics, 2014, 9, 142-149.	3.1	6
43	A global analysis of Y-chromosomal haplotype diversity for 23 STR loci. Forensic Science International: Genetics, 2014, 12, 12-23.	3.1	214
44	Toward Male Individualization with Rapidly Mutating Y-Chromosomal Short Tandem Repeats. Human Mutation, 2014, 35, 1021-1032.	2.5	151
45	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
46	LMNA mutations in Polish patients with dilated cardiomyopathy: prevalence, clinical characteristics, and in vitro studies. BMC Medical Genetics, 2013, 14, 55.	2.1	11
47	Association between Age at Diagnosis of Graves' Disease and Variants in Genes Involved in Immune Response. PLoS ONE, 2013, 8, e59349.	2.5	38
48	Variants of the Lamin A/C (LMNA) Gene in Non-Valvular Atrial Fibrillation Patients. Molecular Diagnosis and Therapy, 2012, 16, 99-107.	3.8	24
49	The peopling of Europe and the cautionary tale of Y chromosome lineage R-M269. Proceedings of the Royal Society B: Biological Sciences, 2012, 279, 884-892.	2.6	84
50	Association between Variants on Chromosome 4q25, 16q22 and 1q21 and Atrial Fibrillation in the Polish Population. PLoS ONE, 2011, 6, e21790.	2.5	31
51	The Genetic Basis of Graves Disease. Current Genomics, 2011, 12, 542-563.	1.6	43
52	Inverse association of the obesity predisposing FTO rs9939609 genotype with alcohol consumption and risk for alcohol dependence. Addiction, 2011, 106, 739-748.	3.3	59
53	Association between Tryptophan Hydroxylase 2 Gene Polymorphism and Completed Suicide. Suicide and Life-Threatening Behavior, 2010, 40, 553-560.	1.9	19
54	Mutability of Y-Chromosomal Microsatellites: Rates, Characteristics, Molecular Bases, and Forensic Implications. American Journal of Human Genetics, 2010, 87, 341-353.	6.2	324

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55	Genetic and ultrastructural studies in dilated cardiomyopathy patients: a large deletion in the lamin A/C gene is associated with cardiomyocyte nuclear envelope disruption. Basic Research in Cardiology, 2010, 105, 365-377.	5.9	79
56	Comprehensive mutation analysis of 17 Y-chromosomal short tandem repeat polymorphisms included in the AmpFlSTRÂ® YfilerÂ® PCR amplification kit. International Journal of Legal Medicine, 2009, 123, 471-482.	2.2	121
57	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.	2.8	46
58	An evaluation of the genetic-matched pair study design using genome-wide SNP data from the European population. European Journal of Human Genetics, 2009, 17, 967-975.	2.8	8
59	Polymorphism of the oestrogen receptor beta gene (ESR2) is associated with susceptibility to Gravesâ€™ disease. Clinical Endocrinology, 2008, 68, 429-434.	2.4	30
60	Correlation between Genetic and Geographic Structure in Europe. Current Biology, 2008, 18, 1241-1248.	3.9	449
61	Effect of protein convertase subtilisin/kexin type 9 (PCSK9) 46L gene polymorphism on LDL cholesterol concentration in a Polish adult population. Molecular Genetics and Metabolism, 2008, 94, 259-262.	1.1	21
62	Cytotoxic T-Lymphocyte Associated Antigen 4 Gene Polymorphisms and Autoimmune Thyroid Disease: A Meta-Analysis. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 3162-3170.	3.6	162
63	Response to letter of Dr van Werkum et al.. International Journal of Cardiology, 2007, 119, 122-123.	1.7	1
64	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.	1.2	92
65	Susceptibility genes in Graves? ophthalmopathy: searching for a needle in a haystack?. Clinical Endocrinology, 2007, 67, 3-19.	2.4	78
66	Lymphoid tyrosine phosphatase (PTPN22/LYP) variant and Graves' disease in a Polish population: association and gene dose-dependent correlation with age of onset. Clinical Endocrinology, 2005, 62, 679-682.	2.4	117
67	Significant genetic differentiation between Poland and Germany follows present-day political borders, as revealed by Y-chromosome analysis. Human Genetics, 2005, 117, 428-443.	3.8	123
68	Susceptibility to ovarian endometriosis in Polish population is not associated with HLA-DRB1 alleles. Human Reproduction, 2005, 20, 970-973.	0.9	13
69	GJB2 Mutations and Degree of Hearing Loss: A Multicenter Study. American Journal of Human Genetics, 2005, 77, 945-957.	6.2	455
70	Association of CD40 Gene Polymorphism (C-1T) with Susceptibility and Phenotype of Graves' Disease. Thyroid, 2005, 15, 1119-1124.	4.5	59
71	Arrhythmogenic right ventricular cardiomyopathy in two pairs of monozygotic twins. International Journal of Cardiology, 2005, 105, 126-133.	1.7	20
72	Distribution ofTNFA haplotypes in healthy Caucasians: Comment on the articles by Newton et al and Zeggini et al. Arthritis and Rheumatism, 2004, 50, 2034-2035.	6.7	2

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73	STR data for the power plex-16 loci in a population from Central Poland. Forensic Science International, 2004, 139, 261-263.	2.2	9
74	A gene in the telomeric HLA complex distinct from HLA-A is involved in predisposition to juvenile idiopathic arthritis. Arthritis and Rheumatism, 2002, 46, 1614-1619.	6.7	25
75	Homogeneity and distinctiveness of Polish paternal lineages revealed by Y chromosome microsatellite haplotype analysis. Human Genetics, 2002, 110, 592-600.	3.8	91
76	Population genetics of 10 STR loci in a population of Central Poland. Forensic Science International, 2002, 130, 55-57.	2.2	8
77	Interaction between HLA-DR and HLA-DP, and Between HLA and interleukin 1Î± in juvenile rheumatoid arthritis indicates heterogeneity of pathogenic mechanisms of the disease. Human Immunology, 1995, 42, 343-347.	2.4	26
78	Association to HLA-DRB1âˆ—08, HLA-DPB1âˆ—0301 and homozygosity for an HLA-linked proteasome gene in juvenile ankylosing spondylitis. Human Immunology, 1995, 44, 88-96.	2.4	50
79	Polymorphism of human major histocompatibility complex-encoded transporter associated with antigen processing (TAP) genes and susceptibility to juvenile rheumatoid arthritis. Human Immunology, 1994, 39, 54-60.	2.4	33
80	Hla class ii alleles and heterogeneity of juvenile rheumatoid arthritis.drb1*0101 may define a novel subset of the disease. Arthritis and Rheumatism, 1993, 36, 465-472.	6.7	70
81	Linkage disequilibrium between TAP2 variants and HLA class II alleles; no primary association between TAP2 variants and insulin-dependent diabetes mellitus. European Journal of Immunology, 1993, 23, 1050-1056.	2.9	81
82	On the HLAâ€ÐQ(Î±1*0501, Î²1*0201)â€associated susceptibility in celiac disease: A possible gene dosage effect of DQB1*0201. Tissue Antigens, 1993, 41, 173-177.	1.0	156