

# Rafal Ploski

## List of Publications by Year in descending order

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Version: 2024-02-01

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	GJB2 Mutations and Degree of Hearing Loss: A Multicenter Study. <i>American Journal of Human Genetics</i> , 2005, 77, 945-957.	6.2	455
2	Correlation between Genetic and Geographic Structure in Europe. <i>Current Biology</i> , 2008, 18, 1241-1248.	3.9	449
3	Mutability of Y-Chromosomal Microsatellites: Rates, Characteristics, Molecular Bases, and Forensic Implications. <i>American Journal of Human Genetics</i> , 2010, 87, 341-353.	6.2	324
4	A global analysis of Y-chromosomal haplotype diversity for 23 STR loci. <i>Forensic Science International: Genetics</i> , 2014, 12, 12-23.	3.1	214
5	Cytotoxic T-Lymphocyte Associated Antigen 4 Gene Polymorphisms and Autoimmune Thyroid Disease: A Meta-Analysis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2007, 92, 3162-3170.	3.6	162
6	On the HLA-DQ(Î±1*0501, Î²1*0201)-associated susceptibility in celiac disease: A possible gene dosage effect of <i>DQB1*0201</i>. <i>Tissue Antigens</i> , 1993, 41, 173-177.	1.0	156
7	Toward Male Individualization with Rapidly Mutating Y-Chromosomal Short Tandem Repeats. <i>Human Mutation</i> , 2014, 35, 1021-1032.	2.5	151
8	Monogenic variants in dystonia: an exome-wide sequencing study. <i>Lancet Neurology</i> , The, 2020, 19, 908-918.	10.2	139
9	Significant genetic differentiation between Poland and Germany follows present-day political borders, as revealed by Y-chromosome analysis. <i>Human Genetics</i> , 2005, 117, 428-443.	3.8	123
10	Comprehensive mutation analysis of 17 Y-chromosomal short tandem repeat polymorphisms included in the AmpFISTRÂ® YfilerÂ® PCR amplification kit. <i>International Journal of Legal Medicine</i> , 2009, 123, 471-482.	2.2	121
11	Lymphoid tyrosine phosphatase (PTPN22/LYP) variant and Graves' disease in a Polish population: association and gene dose-dependent correlation with age of onset. <i>Clinical Endocrinology</i> , 2005, 62, 679-682.	2.4	117
12	Dilated Cardiomyopathy Due to BCL2-Associated Athanogene (BAG3) Mutations. <i>Journal of the American College of Cardiology</i> , 2018, 72, 2471-2481.	2.8	93
13	M34T and V37I mutations in <i>GJB2</i> associated hearing impairment: Evidence for pathogenicity and reduced penetrance. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2534-2543.	1.2	92
14	Homogeneity and distinctiveness of Polish paternal lineages revealed by Y chromosome microsatellite haplotype analysis. <i>Human Genetics</i> , 2002, 110, 592-600.	3.8	91
15	Does p.Q247X in TRIM63 Cause Human Hypertrophic Cardiomyopathy?. <i>Circulation Research</i> , 2014, 114, e2-5.	4.5	88
16	The peopling of Europe and the cautionary tale of Y chromosome lineage R-M269. <i>Proceedings of the Royal Society B: Biological Sciences</i> , 2012, 279, 884-892.	2.6	84
17	Linkage disequilibrium between TAP2 variants and HLA class II alleles; no primary association between TAP2 variants and insulin-dependent diabetes mellitus. <i>European Journal of Immunology</i> , 1993, 23, 1050-1056.	2.9	81
18	The BAG3 gene variants in Polish patients with dilated cardiomyopathy: four novel mutations and a genotype-phenotype correlation. <i>Journal of Translational Medicine</i> , 2014, 12, 192.	4.4	81

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19	Genetic and ultrastructural studies in dilated cardiomyopathy patients: a large deletion in the lamin A/C gene is associated with cardiomyocyte nuclear envelope disruption. <i>Basic Research in Cardiology</i> , 2010, 105, 365-377.	5.9	79
20	Susceptibility genes in Graves' ophthalmopathy: searching for a needle in a haystack?. <i>Clinical Endocrinology</i> , 2007, 67, 3-19.	2.4	78
21	Hla class ii alleles and heterogeneity of juvenile rheumatoid arthritis.drbl*0101 may define a novel subset of the disease. <i>Arthritis and Rheumatism</i> , 1993, 36, 465-472.	6.7	70
22	Collagen synthesis disruption and downregulation of core elements of TGF- $\beta$ <sup>2</sup> , Hippo, and Wnt pathways in keratoconus corneas. <i>European Journal of Human Genetics</i> , 2017, 25, 582-590.	2.8	70
23	The Role of Recent Admixture in Forming the Contemporary West Eurasian Genomic Landscape. <i>Current Biology</i> , 2015, 25, 2518-2526.	3.9	68
24	Titin Truncating Variants in Dilated Cardiomyopathy – Prevalence and Genotype-Phenotype Correlations. <i>PLoS ONE</i> , 2017, 12, e0169007.	2.5	63
25	Association of CD40 Gene Polymorphism (C-1T) with Susceptibility and Phenotype of Graves' Disease. <i>Thyroid</i> , 2005, 15, 1119-1124.	4.5	59
26	Inverse association of the obesity predisposing <i>FTO</i> rs9939609 genotype with alcohol consumption and risk for alcohol dependence. <i>Addiction</i> , 2011, 106, 739-748.	3.3	59
27	Association to HLA-DRB1*08, HLA-DPB1*0301 and homozygosity for an HLA-linked proteasome gene in juvenile ankylosing spondylitis. <i>Human Immunology</i> , 1995, 44, 88-96.	2.4	50
28	Phenotypic variability of patients homozygous for the GJB2 mutation 35delG cannot be explained by the influence of one major modifier gene. <i>European Journal of Human Genetics</i> , 2009, 17, 517-524.	2.8	46
29	The Genetic Basis of Graves Disease. <i>Current Genomics</i> , 2011, 12, 542-563.	1.6	43
30	Association between Age at Diagnosis of Graves' Disease and Variants in Genes Involved in Immune Response. <i>PLoS ONE</i> , 2013, 8, e59349.	2.5	38
31	Evidence for troponin C ( <i>TNNC1</i> ) as a gene for autosomal recessive restrictive cardiomyopathy with fatal outcome in infancy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3241-3248.	1.2	37
32	Homozygous mutation in the Neurofascin gene affecting the glial isoform of Neurofascin causes severe neurodevelopment disorder with hypotonia, amimia and areflexia. <i>Human Molecular Genetics</i> , 2018, 27, 3669-3674.	2.9	34
33	iPSC-derived myelinoids to study myelin biology of humans. <i>Developmental Cell</i> , 2021, 56, 1346-1358.e6.	7.0	34
34	Polymorphism of human major histocompatibility complex-encoded transporter associated with antigen processing (TAP) genes and susceptibility to juvenile rheumatoid arthritis. <i>Human Immunology</i> , 1994, 39, 54-60.	2.4	33
35	Identification and characterization of novel rapidly mutating Y-chromosomal short tandem repeat markers. <i>Human Mutation</i> , 2020, 41, 1680-1696.	2.5	33
36	Association between Variants on Chromosome 4q25, 16q22 and 1q21 and Atrial Fibrillation in the Polish Population. <i>PLoS ONE</i> , 2011, 6, e21790.	2.5	31

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37	A Novel CDC42 Mutation in an 11-Year Old Child Manifesting as Syndromic Immunodeficiency, Autoinflammation, Hemophagocytic Lymphohistiocytosis, and Malignancy: A Case Report. <i>Frontiers in Immunology</i> , 2020, 11, 318.	4.8	31
38	Polymorphism of the oestrogen receptor beta gene ( ESR2 ) is associated with susceptibility to Gravesâ€™ disease. <i>Clinical Endocrinology</i> , 2008, 68, 429-434.	2.4	30
39	ZMZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 104, 319-330.	6.2	30
40	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. <i>Human Immunology</i> , 1995, 42, 343-347.	2.4	26
41	A gene in the telomeric HLA complex distinct from HLA-A is involved in predisposition to juvenile idiopathic arthritis. <i>Arthritis and Rheumatism</i> , 2002, 46, 1614-1619.	6.7	25
42	Variants of the Lamin A/C (LMNA) Gene in Non-Valvular Atrial Fibrillation Patients. <i>Molecular Diagnosis and Therapy</i> , 2012, 16, 99-107.	3.8	24
43	Further evidence for <i>GRIN2B</i> mutation as the cause of severe epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3265-3270.	1.2	22
44	Characterization of Ocular Surface Microbial Profiles Revealed Discrepancies between Conjunctival and Corneal Microbiota. <i>Pathogens</i> , 2021, 10, 405.	2.8	22
45	Effect of protein convertase subtilisin/kexin type 9 (PCSK9) 46L gene polymorphism on LDL cholesterol concentration in a Polish adult population. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 259-262.	1.1	21
46	Arrhythmogenic right ventricular cardiomyopathy in two pairs of monozygotic twins. <i>International Journal of Cardiology</i> , 2005, 105, 126-133.	1.7	20
47	Association between Polymorphisms in the TSHR Gene and Graves' Orbitopathy. <i>PLoS ONE</i> , 2014, 9, e102653.	2.5	20
48	Association between Tryptophan Hydroxylase 2 Gene Polymorphism and Completed Suicide. <i>Suicide and Life-Threatening Behavior</i> , 2010, 40, 553-560.	1.9	19
49	The 4q25, 1q21, and 16q22 polymorphisms and recurrence of atrial fibrillation after pulmonary vein isolation. <i>Archives of Medical Science</i> , 2016, 1, 38-44.	0.9	19
50	Phenotypic expansion in Zhuâ€™Tokitaâ€™Takenouchiâ€™Kim syndrome caused by de novo variants in the <i>SON</i> gene. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1432.	1.2	17
51	Novel de novo mutation affecting two adjacent aminoacids in the EED gene in a patient with Weaver syndrome. <i>Journal of Human Genetics</i> , 2018, 63, 517-520.	2.3	16
52	Multiple Differentially Methylated Regions Specific to Keratoconus Explain Known Keratoconus Linkage Loci. , 2019, 60, 1501.		15
53	Susceptibility to ovarian endometriosis in Polish population is not associated with HLA-DRB1 alleles. <i>Human Reproduction</i> , 2005, 20, 970-973.	0.9	13
54	Malan syndrome (Sotos syndrome 2) in two patients with 19p13.2 deletion encompassing NFIX gene and novel NFIX sequence variant. <i>Biomedical Papers of the Medical Faculty of the University Palacky&amp;#x0301;, Olomouc, Czechoslovakia</i> , 2016, 160, 161-167.	0.6	13

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55	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
56	Exome sequencing reveals mutations in <i>MFN2</i> and <i>GDAP1</i> in severe Charcot-Marie-Tooth disease. Journal of the Peripheral Nervous System, 2014, 19, 242-245.	3.1	12
57	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
58	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
59	LMNA mutations in Polish patients with dilated cardiomyopathy: prevalence, clinical characteristics, and in vitro studies. BMC Medical Genetics, 2013, 14, 55.	2.1	11
60	Differences in Gene-Gene Interactions in Graves' Disease Patients Stratified by Age of Onset. PLoS ONE, 2016, 11, e0150307.	2.5	11
61	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
62	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
63	STR data for the power plex-16 loci in a population from Central Poland. Forensic Science International, 2004, 139, 261-263.	2.2	9
64	Can Circulating Cardiac Biomarkers Be Helpful in the Assessment of LMNA Mutation Carriers?. Journal of Clinical Medicine, 2020, 9, 1443.	2.4	9
65	Variants of <i>ATP1A3</i> in residue 756 cause a separate phenotype of relapsing encephalopathy with cerebellar ataxia (RECA) - Report of two cases and literature review. Molecular Genetics & Genomic Medicine, 2021, 9, e1772.	1.2	9
66	Population genetics of 10 STR loci in a population of Central Poland. Forensic Science International, 2002, 130, 55-57.	2.2	8
67	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
68	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
69	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
70	Polish population data on 15 autosomal STRs of AmpFISTR NGM PCR kit. Forensic Science International: Genetics, 2014, 9, 142-149.	3.1	6
71	Co-occurrence of Jalili syndrome and muscular overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 2280-2283.	1.2	5
72	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

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73	A Neurodevelopmental Disorder With Dystonia and Chorea Resulting From Clustering <i>CAMK4</i> Variants. <i>Movement Disorders</i> , 2021, 36, 520-521.	3.9	5
74	Changes in Nuclear Gene Expression Related to Mitochondrial Function Affect Extracellular Matrix, Collagens, and Focal Adhesion in Keratoconus. <i>Translational Vision Science and Technology</i> , 2021, 10, 6.	2.2	5
75	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. <i>Endocrine Practice</i> , 2015, 21, 993-1000.	2.1	4
76	Perforin gene variation influences survival in childhood acute lymphoblastic leukemia. <i>Leukemia Research</i> , 2018, 65, 29-33.	0.8	4
77	The 9p21 polymorphism is linked with atrial fibrillation during acute phase of ST-segment elevation myocardial infarction. <i>Heart and Vessels</i> , 2016, 31, 1590-1594.	1.2	3
78	Evaluation of electrocardiographic parameters in patients with hearing loss genotyped for the connexin 26 gene ( <i>GJB2</i> ) mutations. <i>Brazilian Journal of Otorhinolaryngology</i> , 2017, 83, 176-182.	1.0	3
79	Puzzling outcome of the nationwide genetic survey of severe/moderate female haemophilia B in Poland. <i>Haemophilia</i> , 2019, 25, e373-e376.	2.1	3
80	Distribution of <i>TNFA</i> haplotypes in healthy Caucasians: Comment on the articles by Newton et al and Zeggini et al. <i>Arthritis and Rheumatism</i> , 2004, 50, 2034-2035.	6.7	2
81	Response to letter of Dr van Werkum et al.. <i>International Journal of Cardiology</i> , 2007, 119, 122-123.	1.7	1
82	Peritoneal dialysis in an adult patient with tetralogy of Fallot diagnosed with incomplete Alagille syndrome. <i>BMC Medical Genetics</i> , 2020, 21, 195.	2.1	0