Dijana Plaseska-Karanfilska

List of Publications by Year
in descending order

115 papers	4,257 citations	304701 22 h-index	138468 58 g-index
123 all docs	123 docs citations	123 times ranked	6526 citing authors

#	Article	IF	Citations
1	Rare germline copy number variants (CNVs) and breast cancer risk. Communications Biology, 2022, 5, 65.	4.4	6
2	Common variants in breast cancer risk loci predispose to distinct tumor subtypes. Breast Cancer Research, 2022, 24, 2.	5.0	15
3	Global change in hepatitis C virus prevalence and cascade of care between 2015 and 2020: a modelling study. The Lancet Gastroenterology and Hepatology, 2022, 7, 396-415.	8.1	237
4	AlstrÃ¶m Syndrome with Early Vision and Hearing Impairement. Prilozi - Makedonska Akademija Na Naukite I Umetnostite Oddelenie Za Medicinski Nauki, 2022, 43, 159-162.	0.5	0
5	De novo mutations in idiopathic male infertilityâ€"A pilot study. Andrology, 2021, 9, 212-220.	3.5	19
6	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. British Journal of Cancer, 2021, 124, 842-854.	6.4	5
7	Functional annotation of the 2q35 breast cancer risk locus implicates a structural variant in influencing activity of a long-range enhancer element. American Journal of Human Genetics, 2021, 108, 1190-1203.	6.2	6
8	Association of germline genetic variants with breast cancer-specific survival in patient subgroups defined by clinic-pathological variables related to tumor biology and type of systemic treatment. Breast Cancer Research, 2021, 23, 86.	5.0	7
9	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	6.4	9
10	Breast Cancer Risk Factors and Survival by Tumor Subtype: Pooled Analyses from the Breast Cancer Association Consortium. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 623-642.	2.5	19
11	Renal Hypouricemia 1: Rare Disorder as Common Disease in Eastern Slovakia Roma Population. Biomedicines, 2021, 9, 1607.	3.2	12
12	Current State of Compulsory Basic and Clinical Courses in Genetics for Medical Students at Medical Faculties in Balkan Countries With Slavic Languages. Frontiers in Genetics, 2021, 12, 793834.	2.3	0
13	Heterotopic ossifications and Charcot joints: Congenital insensitivity to pain with anhidrosis (CIPA) and a novel NTRK1 gene mutation. European Journal of Medical Genetics, 2020, 63, 103613.	1.3	3
14	Fine-mapping of 150 breast cancer risk regions identifies 191 likely target genes. Nature Genetics, 2020, 52, 56-73.	21.4	120
15	Association of TNF â€¤ (rs361525 and rs1800629) with susceptibility to cervical intraepithelial lesion and cervical carcinoma in women from Republic of North Macedonia. International Journal of Immunogenetics, 2020, 47, 522-528.	1.8	6
16	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	6.2	39
17	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
18	Germline HOXB13 mutations p.G84E and p.R217C do not confer an increased breast cancer risk. Scientific Reports, 2020, 10, 9688.	3.3	2

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19	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
20	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	3.7	11
21	Multilevel regression modeling for aneuploidy classification and physical separation of maternal cell contamination facilitates the QF-PCR based analysis of common fetal aneuploidies. PLoS ONE, 2019, 14, e0221227.	2.5	7
22	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
23	Two truncating variants in FANCC and breast cancer risk. Scientific Reports, 2019, 9, 12524.	3.3	5
24	Shared heritability and functional enrichment across six solid cancers. Nature Communications, 2019, 10, 431.	12.8	88
25	The spectrum of BRCA1 and BRCA2 pathogenic sequence variants in Middle Eastern, North African, and South European countries. Human Mutation, 2019, 40, e1-e23.	2.5	34
26	Severe digital malformations in a rare variant of fibrodysplasia ossificans progressiva. American Journal of Medical Genetics, Part A, 2019, 179, 1310-1314.	1.2	5
27	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
28	Evaluation of APOE Genotype and Vascular Risk Factors As Prognostic and Risk Factors for Alzheimerâ€™s Disease and Their Influence On Age of Symptoms Onset. Open Access Macedonian Journal of Medical Sciences, 2019, 7, 516-520.	0.2	2
29	Y-chromosome haplogroup architecture confers susceptibility to azoospermia factor c microrearrangements: a retrospective study. Croatian Medical Journal, 2019, 60, 273-283.	0.7	3
30	Cover Image, Volume 179A, Number 7, July 2019. , 2019, 179, .		2
31	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
32	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. International Journal of Epidemiology, 2019, 48, 795-806.	1.9	81
33	BRCA1 and BRCA2 germline variants in breast cancer patients from the Republic of Macedonia. Breast Cancer Research and Treatment, 2018, 168, 745-753.	2.5	8
34	TIMP3 Promoter Methylation Represents an Epigenetic Marker of BRCA1ness Breast Cancer Tumours. Pathology and Oncology Research, 2018, 24, 937-940.	1.9	13
35	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	21.4	184
36	Novel Founder Mutation in FANCA Gene (c.3446_3449dupCCCT) Among Romani Patients from the Balkan Region. Balkan Medical Journal, 2018, 35, 108-111.	0.8	6

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37	Hyponatremic dehydration and metabolic alkalosis as dominant manifestation in cystic fibrosis infants with mild phenotype - a case series. Srpski Arhiv Za Celokupno Lekarstvo, 2018, 146, 581-583.	0.2	1
38	Molecular and immunohistochemical characteristics of complete hydatidiform moles. Balkan Journal of Medical Genetics, 2017, 20, 27-34.	0.5	6
39	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	27.8	1,099
40	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
41	First Cases of Hb Agrinio Described in Patients from the Republic of Macedonia. Hemoglobin, 2017, 41, 308-310.	0.8	2
42	Genetic variation in leptin and leptin receptor genes as a risk factor for idiopathic male infertility. Andrology, 2017, 5, 70-74.	3.5	8
43	Moleculary Confirmed, Cytogenetic Remission in a Case with Myelodysplastic Syndrome Treated with Azacitidne. Prilozi - Makedonska Akademija Na Naukite I Umetnostite Oddelenie Za Medicinski Nauki, 2017, 38, 157-162.	0.5	0
44	Influence of OASL gene polymorphisms on host response to interferon therapy in chronic hepatitis C virus patients. The EuroBiotech Journal, 2017, 1, 117-125.	1.0	0
45	Molecular and histological characteristics of early triploid and partial molar pregnancies. Polish Journal of Pathology, 2017, 2, 138-143.	0.3	1
46	Association of breast cancer risk with genetic variants showing differential allelic expression: Identification of a novel breast cancer susceptibility locus at 4q21. Oncotarget, 2016, 7, 80140-80163.	1.8	31
47	MicroRNA Profiling in Patients with Upper Tract Urothelial Carcinoma Associated with Balkan Endemic Nephropathy. BioMed Research International, 2016, 2016, 1-10.	1.9	12
48	Loss of Y Chromosome in Peripheral Blood of Colorectal and Prostate Cancer Patients. PLoS ONE, 2016, 11, e0146264.	2.5	79
49	Rare ATAD5 missense variants in breast and ovarian cancer patients. Cancer Letters, 2016, 376, 173-177.	7.2	21
50	Micro<scp>RNA</scp> expression profiles in testicular biopsies of patients with impaired spermatogenesis. Andrology, 2016, 4, 1020-1027.	3.5	39
51	Frameshift variant <scp>FANCL</scp>*c.1096_1099dupATTA is not associated with high breast cancer risk. Clinical Genetics, 2016, 90, 385-386.	2.0	3
52	Association of p53Pro72Arg (rs1042522) and MDM2309 (rs2279744) polymorphisms with risk for cervical intraepthelial lesions and cervical cancer development in Macedonian women. Makedonsko Farmacevtski Bilten, 2016, 62, 49-58.	0.0	0
53	LHX4 Gene Alterations: Patient Report and Review of the Literature. Pediatric Endocrinology Reviews, 2016, 13, 749-55.	1.2	8
54	Clinical Relevance of CHEK2 And NBN Mutations in the Macedonian Population. Balkan Journal of Medical Genetics, 2015, 18, 47-54.	0.5	0

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55	CYP2D6 allele distribution in Macedonians, Albanians and Romanies in the Republic of Macedonia. Balkan Journal of Medical Genetics, 2015, 18, 49-58.	0.5	5
56	Identification of six novel F9 mutations among haemophilia B patients from Macedonia and Bulgaria. Haemophilia, 2015, 21, e144-6.	2.1	0
57	Hb G-Waimanalo [A1] orÎ±64(E13)Aspâ†'Asn (Î±1) (HBA1: c.193G>A) Observed in a Bulgarian Family. Hemoglobin, 2015, 39, 430-431.	0.8	1
58	HPV E6/E7 mRNA versus HPV DNA biomarker in cervical cancer screening of a group of Macedonian women. Journal of Medical Virology, 2015, 87, 1578-1586.	5.0	28
59	SNaPshot Assay for the Detection of the Most Common CFTR Mutations in Infertile Men. PLoS ONE, 2014, 9, e112498.	2.5	6
60	NGS NominatedCELA1,HSPG2, andKCNK5as Candidate Genes for Predisposition to Balkan Endemic Nephropathy. BioMed Research International, 2014, 2014, 1-7.	1.9	25
61	Fanconi Anemia Founder Mutation in Macedonian Patients. Acta Haematologica, 2014, 132, 15-21.	1.4	4
62	Study of Three Single Nucleotide Polymorphisms in the Slc6a14 Gene in Association with Male Infertility. Balkan Journal of Medical Genetics, 2014, 17, 61-66.	0.5	6
63	Fast, reliable and low cost user-developed protocol for detection, quantification and genotyping of hepatitis C virus. Journal of Virological Methods, 2014, 196, 104-112.	2.1	17
64	"Balkan journal of medical genetics"--facts, editorial policies, practices and challenges. Prilozi - Makedonska Akademija Na Naukite I Umetnostite Oddelenie Za Medicinski Nauki, 2014, 35, 89-93.	0.5	0
65	A Homozygous Deletion of the DPY19L2 Gene is a Cause of Globozoospermia in Men From the Republic of Macedonia. Balkan Journal of Medical Genetics, 2013, 16, 73-76.	0.5	17
66	Molecular characterization of cystinuria in south-eastern European countries. Urolithiasis, 2013, 41, 21-30.	2.0	9
67	Prevalence of hepatitis C virus genotypes in risk groups in the Republic of Macedonia: A 5 years survey. Journal of Medical Virology, 2013, 85, 2072-2078.	5.0	8
68	Detection of Thrombophilic Mutations Related to Spontaneous Abortions by a Multiplex SNaPshot Method. Genetic Testing and Molecular Biomarkers, 2012, 16, 259-264.	0.7	12
69	Genetic Variation of The BRCA1 and BRCA2 Genes in Macedonian Patients. Balkan Journal of Medical Genetics, 2012, 15, 81-85.	0.5	8
70	National Reference Centre for Genomics and Proteomics â€" Macprogen. Balkan Journal of Medical Genetics, 2012, 15, 9-12.	0.5	0
71	Study of the Hepatitis C Virus in the Republic of Macedonia. Balkan Journal of Medical Genetics, 2012, 15, 67-69.	0.5	3
72	Cag Repeat Number in the Androgen Receptor Gene and Prostate Cancer. Balkan Journal of Medical Genetics, 2012, 15, 31-36.	0.5	7

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73	Molecular Diagnostics of ÃŸ-Thalassemia. Balkan Journal of Medical Genetics, 2012, 15, 61-65.	0.5	3
74	Genetic Causes of Male Infertility. Balkan Journal of Medical Genetics, 2012, 15, 31-34.	0.5	35
75	Genetics of non Syndromic Hearing Loss in the Republic of Macedonia. Balkan Journal of Medical Genetics, 2012, 15, 57-59.	0.5	4
76	Association Study of Singleâ€Nucleotide Polymorphisms in FASLG, JMJDIA, LOC203413, TEX15, BRDT, OR2W3, INSR, and TAS2R38 Genes With Male Infertility. Journal of Andrology, 2012, 33, 675-683.	2.0	44
77	Proteomic analysis of seminal plasma in men with different spermatogenic impairment. Andrologia, 2012, 44, 256-264.	2.1	49
78	Efficient Detection of Mediterranean Î²-Thalassemia Mutations by Multiplex Single-Nucleotide Primer Extension. PLoS ONE, 2012, 7, e48167.	2.5	8
79	Human Seminal Plasma Proteome Study: a Search for Male Infertility Biomarkers. Balkan Journal of Medical Genetics, 2012, 15, 35-38.	0.5	7
80	MicroRNAs in Breast Cancer â€" Our Initial Results. Balkan Journal of Medical Genetics, 2012, 15, 87-89.	0.5	3
81	Rapid and non Invasive Prenatal Diagnosis. Balkan Journal of Medical Genetics, 2012, 15, 39-43.	0.5	0
82	Detection of the Most Common Genetic Causes of Male Infertility by Quantitative Fluorescent (QF)-PCR Analysis. , 2011, , .		4
83	Cystinuria AA (B): digenic inheritance with three mutations in two cystinuria genes. Journal of Genetics, 2011, 90, 157-159.	0.7	9
84	Professor Georgi D. Efremov (1932â€"2011). Acta Haematologica, 2011, 126, 151-151.	1.4	1
85	Quantitative Fluorescent-PCR Detection of Sex Chromosome Aneuploidies and AZF Deletions/Duplications. Genetic Testing and Molecular Biomarkers, 2008, 12, 595-605.	1.7	27
86	Prenatal Diagnosis of Spinal Muscular Atrophy in Macedonian Families. Genetic Testing and Molecular Biomarkers, 2008, 12, 391-393.	1.7	2
87	Hb Jambol: A New Hyperunstable Hemoglobin Causing Severe Hemolytic Anemia. Acta Haematologica, 2007, 117, 1-7.	1.4	11
88	A new familial mutation (R133G) in the SRY gene. Clinical Genetics, 2007, 71, 480-482.	2.0	6
89	Non-invasive fetal sex determination using real-time PCR. Journal of Maternal-Fetal and Neonatal Medicine, 2006, 19, 337-342.	1.5	9
90	AZF deletions in infertile men from the Republic of Macedonia. Prilozi / Makedonska Akademija Na Naukite I Umetnostite, Oddelenie Za Bioloiki I Medicinski Nauki = Contributions / Macedonian Academy of Sciences and Arts, Section of Biological and Medical Sciences, 2006, 27, 5-16.	0.2	2

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91	Hemoglobin A2â€² (HbA2Îˆâ€²) in the Mauritanian population: first results of a preliminary survey. Annals of Hematology, 2002, 81, 386-388.	1.8	9
92	Differential Diagnosis of Hb EE and Hb E-Î²⁰-Thalassemia by Protein and DNA Analyses. Acta Haematologica, 2000, 103, 84-89.	1.4	7
93	Hb Saint Etienne or Hb Istanbul [Î²92(F8)Hisâ†'Gln] Found in An Argentinean Family. Hemoglobin, 2000, 24, 149-152.	0.8	3
94	HB old Dominion/Burton-upon-Trent or Î²143(H2l)Hisâ†'TYR, Found in a Diabetic Woman from Korea. Hemoglobin, 2000, 24, 323-326.	0.8	1
95	Hb Rambam [Î²69(E13)Glyâ†'Asp]/Î²⁰-Thalassemia [Codon 5 (-CT)] in a Family from Argentina. Hemoglobin, 2000, 24, 157-161.	0.8	2
96	A New Î² Chain Variant, Hb Vienna or Î²77(EF1)Hisâ†'Gln. Hemoglobin, 1998, 22, 391-395.	0.8	4
97	Hb Brockton [Î²l38(H16)Alaâ†'Pro] Observed in a Chinese Boy. Hemoglobin, 1998, 22, 397-400.	0.8	1
98	Molecular basis of cystic fibrosis in the Republic of Macedonia. Clinical Genetics, 1998, 54, 203-209.	2.0	11
99	A novel mutation in exon 12 (Y569C) of the CFTR gene identified in a patient of croatian origin. Human Mutation, 1996, 7, 374-375.	2.5	2
100	The Î³-Globin Gene Rearrangements in Newborns from the Repuc of Macedonia. Hemoglobin, 1996, 20, 401-414.	0.8	7
101	Two Rare Mutations [CD 30 (G->C) and CDs 36/37 (â^'T)] in a Turkish Thalassemia Major Patient from Bulgaria. Hemoglobin, 1994, 18, 359-364.	0.8	4
102	HB F-Macedonia-II [GÎ³104(G6)LYSâ†'ASN]: A New Î³ chain variant. Hemoglobin, 1994, 18, 373-382.	0.8	7
103	Hb F-macedonia-I or Î±₂^AÎ³₂2(NA2)HISâ†'GLN. Hemoglobin, 1994, 18, 241-245.	0.8	9
104	A Î²Â°â€thalassaernia due to a 1605 bp deletion of the 5â€˜Î²â€globin gene region. British Journal of Haematology, 1993, 85, 143-147.	2.5	29
105	HB Volga [Î²27(B9)ALAâ†'ASP]: Detection of a DE Novo Mutation by AVA II Digestion of PCR-Amplified DNA. Hemoglobin, 1993, 17, 209-215.	0.8	2
106	HB Hoshida [Î²43(CD2)GLUâ†'GLN] Observed in a Yugoslavian Family. Hemoglobin, 1991, 15, 541-543.	0.8	1
107	HB Yokohama [Î²31(B13)LEUâ†'PRO] Detected AS A de novo Mutation in a Yugoslavian Boy. Hemoglobin, 1991, 15, 469-476.	0.8	5
108	Hb F-Jiangsu, The First Î³ Chain Variant with a Valine -Methionine Substitution: Î±Î³₂134(H12) Valâ†'Met. Hemoglobin, 1990, 14, 177-183.	0.8	7

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109	Hb F-Brooklyn or Î±2GÎ³266(E10)LYSGLN. Hemoglobin, 1990, 14, 213-216.	0.8	7
110			