Borut Peterlin

List of Publications by Year
in descending order

219 papers	5,498 citations	117625 34 h-index	114465 63 g-index
222 all docs	222 docs citations	222 times ranked	8815 citing authors

#	Article	IF	Citations
1	Y-Chromosomal Diversity in Europe Is Clinal and Influenced Primarily by Geography, Rather than by Language. American Journal of Human Genetics, 2000, 67, 1526-1543.	6.2	519
2	Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening. European Journal of Human Genetics, 2015, 23, 1438-1450.	2.8	260
3	Responsible implementation of expanded carrier screening. European Journal of Human Genetics, 2016, 24, e1-e12.	2.8	240
4	Using literature-based discovery to identify disease candidate genes. International Journal of Medical Informatics, 2005, 74, 289-298.	3.3	219
5	Mutations in C2orf37, Encoding a Nucleolar Protein, Cause Hypogonadism, Alopecia, Diabetes Mellitus, Mental Retardation, and Extrapyramidal Syndrome. American Journal of Human Genetics, 2008, 83, 684-691.	6.2	121
6	Systematic review and meta-analysis of genetic association studies in idiopathic recurrent spontaneous abortion. Fertility and Sterility, 2017, 107, 150-159.e2.	1.0	110
7	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq1 1 0.784314 rgBT /Over	2.8	109
8	Nonsense Mutations in Folliculin Presenting as Isolated Familial Spontaneous Pneumothorax in Adults. American Journal of Respiratory and Critical Care Medicine, 2005, 172, 39-44.	5.6	104
9	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
10	Therapeutic perspectives of epigenetically active nutrients. British Journal of Pharmacology, 2015, 172, 2756-2768.	5.4	99
11	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. American Journal of Human Genetics, 2018, 103, 995-1008.	6.2	92
12	Towards a European consensus for reporting incidental findings during clinical NGS testing. European Journal of Human Genetics, 2015, 23, 1601-1606.	2.8	85
13	Screening for Y chromosome microdeletions in 226 Slovenian subfertile men. Human Reproduction, 2002, 17, 17-24.	0.9	82
14	The gene for the familial form of incontinentia pigmenti (IP2) maps to the distal part of Xq28. Human Molecular Genetics, 1994, 3, 273-278.	2.9	71
15	Vaginal Microbiome Signature Is Associated With Spontaneous Preterm Delivery. Frontiers in Medicine, 2019, 6, 201.	2.6	71
16	Genetic polymorphisms in vasoactive genes and preeclampsia: A meta-analysis. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2007, 131, 115-126.	1.1	69
17	Mutations in SCN3A cause early infantile epileptic encephalopathy. Annals of Neurology, 2018, 83, 703-717.	5.3	69
18	Early sympathetic hyperactivity in Huntington's disease. European Journal of Neurology, 2004, 11, 842-848.	3.3	63

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19	Exploiting semantic relations for literature-based discovery. AMIA ... Annual Symposium proceedings, 2006, , 349-53.	0.2	59
20	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
21	Comprehensive use of extended exome analysis improves diagnostic yield in rare disease: a retrospective survey in 1,059 cases. Genetics in Medicine, 2018, 20, 303-312.	2.4	57
22	Local and genetic determinants of vascular endothelial growth factor expression in advanced proliferative diabetic retinopathy. Molecular Vision, 2008, 14, 1382-7.	1.1	54
23	Association of Vascular Endothelial Growth Factor Gene Polymorphism with Myocardial Infarction in Patients with Type 2 Diabetes. Cardiology, 2007, 107, 291-295.	1.4	53
24	Identification of rare genetic variation of NLRP1 gene in familial multiple sclerosis. Scientific Reports, 2017, 7, 3715.	3.3	53
25	Strong Association of Variants around FOXE1 and Orofacial Clefting. Journal of Dental Research, 2014, 93, 376-381.	5.2	51
26	De Novo Mutations in SLC25A24 Cause a Disorder Characterized by Early Aging, Bone Dysplasia, Characteristic Face, and Early Demise. American Journal of Human Genetics, 2017, 101, 844-855.	6.2	51
27	Autonomic dysfunction in presymptomatic and early symptomatic Huntingtonâ€™s disease. Acta Neurologica Scandinavica, 2010, 121, 392-399.	2.1	49
28	Heart-hand syndrome of Slovenian type: a new kind of laminopathy. Journal of Medical Genetics, 2008, 45, 666-671.	3.2	47
29	Telegenetics: an Update on Availability and Use of Telemedicine in Clinical Genetics Service. Journal of Medical Systems, 2017, 41, 21.	3.6	46
30	Using Literature-based Discovery to Identify Novel Therapeutic Approaches. Cardiovascular and Hematological Agents in Medicinal Chemistry, 2013, 11, 14-24.	1.0	45
31	The challenges of the expanded availability of genomic information: an agenda-setting paper. Journal of Community Genetics, 2018, 9, 103-116.	1.2	45
32	Role of genetic polymorphisms in ACE and TNF-Î± gene in sarcoidosis: a meta-analysis. Journal of Human Genetics, 2007, 52, 836-847.	2.3	44
33	Phenotype-driven gene target definition in clinical genome-wide sequencing data interpretation. Genetics in Medicine, 2016, 18, 1102-1110.	2.4	41
34	Diagnostic exome sequencing of syndromic epilepsy patients in clinical practice. Clinical Genetics, 2018, 93, 1057-1062.	2.0	39
35	Fragile X premutation in women with sporadic premature ovarian failure in Slovenia. Human Reproduction, 2003, 18, 1637-1640.	0.9	36
36	Loss of heterozygosity at 11q23.1 and survival in breast cancer: Results of a large European study. Genes Chromosomes and Cancer, 1999, 25, 212-221.	2.8	34

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37	Copy number of DAZ genes in infertile men. Fertility and Sterility, 2005, 84, 1522-1525.	1.0	34
38	Association of circadian rhythm genes ARNTL/BMAL1 and CLOCK with multiple sclerosis. PLoS ONE, 2018, 13, e0190601.	2.5	34
39	Aldose reductase (AC)n gene polymorphism and susceptibility to diabetic retinopathy in Type 2 diabetes in Caucasians. Journal of Diabetes and Its Complications, 2005, 19, 70-73.	2.3	33
40	Matrix metalloproteinases 1, 2, 3 and 9 functional single-nucleotide polymorphisms in idiopathic recurrent spontaneous abortion. Reproductive BioMedicine Online, 2012, 24, 567-575.	2.4	33
41	Multiple Sclerosis patients carry an increased burden of exceedingly rare genetic variants in the inflammasome regulatory genes. Scientific Reports, 2019, 9, 9171.	3.3	33
42	Diagnostic outcomes of exome sequencing in patients with syndromic or non-syndromic hearing loss. PLoS ONE, 2018, 13, e0188578.	2.5	33
43	Genetic Variation in Circadian Rhythm Genes CLOCK and ARNTL as Risk Factor for Male Infertility. PLoS ONE, 2013, 8, e59220.	2.5	32
44	Involvement of CFTR Gene Alterations in Obstructive and Nonobstructive Infertility in Men. Genetic Testing and Molecular Biomarkers, 2001, 5, 243-247.	1.7	31
45	Factor V Leiden, prothrombin 20210G â†' A, methylenetetrahydrofolate reductase 677C â†' T and plasminogen activator inhibitor 4G/5G polymorphism in women with pregnancy-related venous thromboembolism. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2003, 111, 157-163.	1.1	30
46	The - 429 T/C and - 374 T/A Gene Polymorphisms of the Receptor of Advanced Glycation End Products Gene are not Risk Factors for Diabetic Retinopathy in Caucasians with Type 2 Diabetes. Klinische Monatsblatter Fur Augenheilkunde, 2003, 220, 873-876.	0.5	30
47	Histamine N-methyltransferase Thr105Ile polymorphism is associated with Parkinson's disease. Neurobiology of Aging, 2012, 33, 836.e1-836.e3.	3.1	29
48	Gly482Ser polymorphism of the peroxisome proliferator-activated receptor-Î³ coactivator-1 gene might be a risk factor for diabetic retinopathy in Slovene population (Caucasians) with type 2 diabetes and the Pro12Ala polymorphism of thePPARÎ³ gene is not. Diabetes/Metabolism Research and Reviews, 2005, 21, 470-474.	4.0	28
49	Gene expression changes in blood as a putative biomarker for Huntington's disease. Movement Disorders, 2009, 24, 2277-2281.	3.9	28
50	Insertion/Deletion Plasminogen Activator Inhibitor 1 and Insertion/Deletion Angiotensin-Converting Enzyme Gene Polymorphisms in Diabetic Retinopathy in Type 2 Diabetes. Ophthalmologica, 2003, 217, 219-224.	1.9	27
51	Relation of CTG expansion and clinical variables to electrocardiogram conduction abnormalities and sudden death in patients with myotonic dystrophy. Neuromuscular Disorders, 2003, 13, 822-826.	0.6	27
52	Expression Signature as a Biomarker for Prenatal Diagnosis of Trisomy 21. PLoS ONE, 2013, 8, e74184.	2.5	27
53	Association between male infertility and genetic variability at the PON1/2 and GSTM1/T1 gene loci. Reproductive BioMedicine Online, 2011, 23, 105-110.	2.4	26
54	Improving diagnostics of rare genetic diseases with NGS approaches. Journal of Community Genetics, 2021, 12, 247-256.	1.2	25

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55	Improving literature based discovery support by genetic knowledge integration. Studies in Health Technology and Informatics, 2003, 95, 68-73.	0.3	25
56	Interaction Between Gene Polymorphisms of Renin-angiotensin System and Metabolic Risk Factors in Premature Myocardial Infarction. Angiology, 2001, 52, 247-252.	1.8	24
57	A hemochromatosis-causing mutation C282Y is a risk factor for proliferative diabetic retinopathy in Caucasians with type 2 diabetes. Journal of Human Genetics, 2003, 48, 646-649.	2.3	24
58	ORIGINAL ARTICLE: Genetic Predisposition to Idiopathic Recurrent Spontaneous Abortion: Contribution of Genetic Variations in IGFâ€2 and H19 Imprinted Genes. American Journal of Reproductive Immunology, 2008, 60, 111-117.	1.2	24
59	Association between genetic polymorphisms in cytokine genes and recurrent miscarriage â€“ a meta-analysis. Reproductive BioMedicine Online, 2009, 19, 406-414.	2.4	24
60	Genetic epidemiology of myotonic dystrophy in Istria, Croatia. Acta Neurologica Scandinavica, 1997, 95, 164-166.	2.1	23
61	BglII gene polymorphism of the Î±2Î²1 integrin gene is a risk factor for diabetic retinopathy in Caucasians with typeÂ2 diabetes. Journal of Human Genetics, 2003, 48, 457-460.	2.3	23
62	Genetic epidemiology of Duchenne and Becker muscular dystrophy in Slovenia. Clinical Genetics, 1997, 51, 94-97.	2.0	23
63	The Role of microRNAs in Heart Failure: A Systematic Review. Frontiers in Cardiovascular Medicine, 2020, 7, 161.	2.4	23
64	Mutations in the hemochromatosis gene (HFE) and multiple sclerosis. Neuroscience Letters, 2005, 383, 301-304.	2.1	22
65	Tumor Necrosis Factor-Î±-308 Gene Polymorphism in Croatian and Slovenian Multiple Sclerosis Patients. European Neurology, 2007, 57, 203-207.	1.4	22
66	Rare missense TUBGCP5 gene variant in a patient with primary microcephaly. European Journal of Medical Genetics, 2019, 62, 103598.	1.3	22
67	Diagnostic and Clinical Utility of Clinical Exome Sequencing in Children With Moderate and Severe Global Developmental Delay / Intellectual Disability. Journal of Child Neurology, 2020, 35, 116-131.	1.4	22
68	Sex Difference in the Effect of ACE-DD Genotype on the Risk of Premature Myocardial Infarction. Angiology, 2004, 55, 155-158.	1.8	21
69	Familial progressive sinoatrial and atrioventricular conduction disease of adult onset with sudden death, dilated cardiomyopathy, and brachydactyly. A new type of heart-hand syndrome?. Clinical Genetics, 2005, 68, 155-160.	2.0	21
70	Region with persistent high frequency of multiple sclerosis in Croatia and Slovenia. Journal of the Neurological Sciences, 2006, 247, 169-172.	0.6	21
71	Analysis of susceptibility loci for nonsyndromic orofacial clefting in a European trio sample. American Journal of Medical Genetics, Part A, 2013, 161, 2545-2549.	1.2	21
72	Genetic variations in circadian rhythm genes and susceptibility for myocardial infarction. Genetics and Molecular Biology, 2018, 41, 403-409.	1.3	21

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73	Y chromosome microdeletions in infertile men with cryptorchidism. Fertility and Sterility, 2003, 79, 1559-1565.	1.0	20
74	Angiotensin-converting enzyme I/D gene polymorphism and risk of multiple sclerosis. Acta Neurologica Scandinavica, 2006, 114, 374-377.	2.1	20
75	Polymorphisms in the Interleukin-12/18 Genes and Recurrent Spontaneous Abortion. American Journal of Reproductive Immunology, 2007, 58, 403-408.	1.2	20
76	Third case of 8q23.3â€q24.13 deletion in a patient with Langerâ€“Giedion syndrome phenotype without TRPS1 gene deletion. American Journal of Medical Genetics, Part A, 2012, 158A, 659-663.	1.2	20
77	Circadian clock genes and myocardial infarction in patients with type 2 diabetes mellitus. Gene, 2019, 701, 98-103.	2.2	19
78	De novo mutations in idiopathic male infertilityâ€”A pilot study. Andrology, 2021, 9, 212-220.	3.5	19
79	Prothrombotic gene polymorphisms and atherothrombotic cerebral infarction. Acta Neurologica Scandinavica, 2003, 108, 109-113.	2.1	18
80	Epidemiology of Huntingtonâ€™s disease in Slovenia. Acta Neurologica Scandinavica, 2009, 119, 371-375.	2.1	18
81	Evaluating the SERCA2 and VEGF mRNAs as Potential Molecular Biomarkers of the Onset and Progression in Huntingtonâ€™s Disease. PLoS ONE, 2015, 10, e0125259.	2.5	18
82	Genetic Interactions in Nonsyndromic Orofacial Clefts in Europeâ€”EUROCRAN Study. Cleft Palate-Craniofacial Journal, 2017, 54, 623-630.	0.9	18
83	DNA Methylation Profiles in Whole Blood of Huntington's Disease Patients. Frontiers in Neurology, 2018, 9, 655.	2.4	18
84	Circadian clock genes and circadian phenotypes in patients with myocardial infarction. Advances in Medical Sciences, 2019, 64, 224-229.	2.1	18
85	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	2.6	18
86	Hemochromatosis gene mutations in the Croatian and Slovenian populations. Clinical Genetics, 2003, 64, 444-446.	2.0	17
87	Human Y-specific STR haplotypes in the Western Croatian population sample. Forensic Science International, 2005, 149, 257-261.	2.2	17
88	Patients with primary cataract as a genetic pool of DMPK protomutation. Journal of Human Genetics, 2007, 52, 123-128.	2.3	17
89	GSTM1-null and GSTT1-null genotypes are associated with essential arterial hypertension in patients with type 2 diabetes. Clinical Biochemistry, 2014, 47, 574-577.	1.9	17
90	Clinical utility of array comparative genomic hybridisation in prenatal setting. BMC Medical Genetics, 2016, 17, 81.	2.1	16

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91	Genetic variation in the CLOCK gene is associated with idiopathic recurrent spontaneous abortion. PLoS ONE, 2018, 13, e0196345.	2.5	16
92	MiRNA as biomarker for uveitis - A systematic review of the literature. Gene, 2019, 696, 162-175.	2.2	16
93	Y microdeletions in the Istria county, Croatia. Asian Journal of Andrology, 2005, 7, 213-216.	1.6	15
94	No association of CCR5D32 gene mutation with multiple sclerosis in Croatian and Slovenian patients. Multiple Sclerosis Journal, 2006, 12, 360-362.	3.0	15
95	Association between the apolipoprotein B signal peptide gene insertion/deletion polymorphism and male infertility. Molecular Human Reproduction, 2006, 12, 777-779.	2.8	15
96	Epidemiology of multiple sclerosis in western Herzegovina. Clinical Neurology and Neurosurgery, 2007, 109, 779-783.	1.4	15
97	Rasch-based high-dimensionality data reduction and class prediction with applications to microarray gene expression data. Expert Systems With Applications, 2010, 37, 5178-5185.	7.6	15
98	Positional integratomic approach in identification of genomic candidate regions for Parkinson's disease. Bioinformatics, 2011, 27, 1971-1978.	4.1	15
99	Epigenetics and Bruxism: Possible Role of Epigenetics in the Etiology of Bruxism. International Journal of Prosthodontics, 2015, 28, 594-599.	1.7	15
100	Combining Semantic Relations and DNA Microarray Data for Novel Hypotheses Generation. Lecture Notes in Computer Science, 2010, , 53-61.	1.3	15
101	Search for sarcoidosis candidate genes by integration of data from genomic, transcriptomic and proteomic studies. Medical Science Monitor, 2009, 15, SR22-8.	1.1	15
102	The influence of cytokine gene polymorphisms on the risk of developing gastric cancer in patients with Helicobacter pylori infection. Radiology and Oncology, 2015, 49, 256-264.	1.7	14
103	A critical update on endothelial nitric oxide synthase gene variations in women with idiopathic recurrent spontaneous abortion: genetic association study, systematic review and meta-analyses. Molecular Human Reproduction, 2015, 21, 466-478.	2.8	14
104	Insertion/deletion polymorphism in intron 16 of ACE gene in idiopathic recurrent spontaneous abortion: case-control study, systematic review and meta-analysis. Reproductive BioMedicine Online, 2016, 32, 237-246.	2.4	14
105	The role of circadian rhythm in male reproduction. Current Opinion in Endocrinology, Diabetes and Obesity, 2019, 26, 313-316.	2.3	14
106	PAI and TPA gene polymorphisms in multiple sclerosis. Multiple Sclerosis Journal, 2008, 14, 243-247.	3.0	13
107	Chi-square-based Scoring Function for Categorization of MEDLINE Citations. Methods of Information in Medicine, 2010, 49, 371-378.	1.2	13
108	Functional Polymorphisms of Matrix Metalloproteinases 1 and 9 Genes in Women with Spontaneous Preterm Birth. Disease Markers, 2014, 2014, 1-7.	1.3	13

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109	Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening. Summary and recommendations. European Journal of Human Genetics, 2015, , .	2.8	13
110	Single signal of the Williams syndrome chromosome region 1 gene in hyperploidic bone marrow cells of acute lymphoblastic leukemia in a Williams syndrome patient. Medical and Pediatric Oncology, 2002, 38, 205-207.	1.0	12
111	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
112	Diagnostic efficacy and new variants in isolated and complex autism spectrum disorder using molecular karyotyping. Journal of Applied Genetics, 2018, 59, 179-185.	1.9	12
113	How to design a national genomic projectâ€”a systematic review of active projects. Human Genomics, 2021, 15, 20.	2.9	12
114	Increasing Genomic Literacy Through National Genomic Projects. Frontiers in Genetics, 2021, 12, 693253.	2.3	12
115	Genetic markers of restenosis after coronary angioplasty and after stent implantation. Medical Science Monitor, 2005, 11, RA127-35.	1.1	12
116	4G4G Genotype of PAI-1 Gene Promoter Polymorphism Is Not Associated with Myocardial Infarction in Caucasians with Type-2 Diabetes. Cardiology, 2003, 100, 157-158.	1.4	11
117	Interleukin 7 receptor alpha polymorphism rs6897932 and susceptibility to multiple sclerosis in the Western Balkans. Multiple Sclerosis Journal, 2010, 16, 533-536.	3.0	11
118	Nonsyndromic cleft palate: An association study at GWAS candidate loci in a multiethnic sample. Birth Defects Research, 2018, 110, 871-882.	1.5	11
119	Pharmacogenomics of Multiple Sclerosis: A Systematic Review. Frontiers in Neurology, 2019, 10, 134.	2.4	11
120	MicroRNA-Target Interaction Regulatory Network in Alzheimerâ€™s Disease. Journal of Personalized Medicine, 2021, 11, 1275.	2.5	11
121	CTG amplification in the DM1PK gene is not associated with idiopathic male subfertility. Human Reproduction, 2004, 19, 2084-2087.	0.9	10
122	Apolipoprotein E gene polymorphism effects triglycerides but not CAD risk in Caucasian women younger than 65Âyears. Annales De GÃ©nÃ©tique, 2004, 47, 147-153.	0.4	10
123	The Role of TPA I/D and PAI-1 4G/5G Polymorphisms in Multiple Sclerosis. Disease Markers, 2014, 2014, 1-8.	1.3	10
124	Sarcoidosis Related Novel Candidate Genes Identified by Multi-Omics Integrative Analyses. OMICS A Journal of Integrative Biology, 2018, 22, 322-331.	2.0	10
125	Actionable Pharmacogenetic Variation in the Slovenian Genomic Database. Frontiers in Pharmacology, 2019, 10, 240.	3.5	10
126	Rapid DNA-based prenatal diagnosis by genetic linkage in three families with Alport's syndrome. American Journal of Kidney Diseases, 1997, 30, 174-179.	1.9	9

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127	Estrogen Receptor Dinucleotide (TA) Polymorphism Does Not Predict Premature Myocardial Infarction in Caucasian Women. Cardiology, 2003, 99, 163-165.	1.4	9
128	Y chromosome azoospermia factor region microdeletions are not associated with idiopathic recurrent spontaneous abortion in a Slovenian population: association study and literature review. Fertility and Sterility, 2013, 99, 1663-1667.	1.0	9
129	Family History as a Predictor for Disease Risk in Healthy Individuals: A Cross-Sectional Study in Slovenia. PLoS ONE, 2013, 8, e80333.	2.5	9
130	The â^'2549 insertion/deletion polymorphism in the promoter region of the VEGFA gene in couples with idiopathic recurrent spontaneous abortion. Journal of Assisted Reproduction and Genetics, 2015, 32, 1789-1794.	2.5	9
131	Transcriptomic Biomarkers for Huntington's Disease: Are Gene Expression Signatures in Whole Blood Reliable Biomarkers?. OMICS A Journal of Integrative Biology, 2018, 22, 283-294.	2.0	9
132	MMP-2 â^'1575G/A polymorphism modifies the onset of optic neuritis as a first presenting symptom in MS?. Journal of Neuroimmunology, 2015, 286, 13-15.	2.3	8
133	Cornelia de Lange syndrome caused by heterozygous deletions of chromosome 8q24: Comments on the article by Pereza et al. [2012]. American Journal of Medical Genetics, Part A, 2015, 167, 1426-1427.	1.2	8
134	Transcriptome Profiling Uncovers Potential Common Mechanisms in Fetal Trisomies 18 and 21. OMICS A Journal of Integrative Biology, 2017, 21, 565-570.	2.0	8
135	A Single Nucleotide Polymorphism of <scp>DNA</scp> methyltransferase 3B gene is a risk factor for recurrent spontaneous abortion. American Journal of Reproductive Immunology, 2017, 78, e12765.	1.2	8
136	Impact of prenatal screening on the prevalence of Down syndrome in Slovenia. PLoS ONE, 2017, 12, e0180348.	2.5	8
137	Cone Dystrophy Associated with a Novel Variant in the Terminal Codon of the RPGR-ORF15. Genes, 2021, 12, 499.	2.4	8
138	Clinical and Histopathological Features of Gelsolin Amyloidosis Associated with a Novel GSN Variant p.Glu580Lys. International Journal of Molecular Sciences, 2021, 22, 1084.	4.1	8
139	DNMT3B rs1569686 and rs2424913 gene polymorphisms are associated with positive family history of preterm birth and smoking status. Croatian Medical Journal, 2020, 61, 8-17.	0.7	8
140	Analysis of the CAG repeat number in exon 1 of the androgen receptor gene in Slovene men with idiopathic azoospermia and oligoasthenoteratozoospermia. Asian Journal of Andrology, 2007, 9, 280-282.	1.6	7
141	Interstitial deletion 2p11.2â€"p12: Further delineation. American Journal of Medical Genetics, Part A, 2009, 149A, 2324-2326.	1.2	7
142	Genetic variation in tissue inhibitors of metalloproteinases as a risk factorÂfor idiopathic recurrent spontaneous abortion. Fertility and Sterility, 2013, 99, 1923-1929.	1.0	7
143	Correlates of Depression in the Slovenian Working Population. Arhiv Za Higijenu Rada I Toksikologiju, 2013, 64, 489-495.	0.7	7
144	Angiotensin-converting enzyme insertion/deletion gene polymorphism and interferon-Î² treatment response in multiple sclerosis patients. Pharmacogenetics and Genomics, 2017, 27, 232-235.	1.5	7

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145	Combination of QFâ€PCR and aCGH is an efficient diagnostic strategy for the detection of chromosome aberrations in recurrent miscarriage. Molecular Genetics & Genomic Medicine, 2019, 7, e980.	1.2	7
146	Diagnostic yield of exome sequencing in myopathies: Experience of a Slovenian tertiary centre. PLoS ONE, 2021, 16, e0252953.	2.5	7
147	Screening for Rare Genetic Variants Associated with Atherosclerosis: Opportunity for Personalized Medicine. Current Vascular Pharmacology, 2018, 17, 25-28.	1.7	7
148	Mucopolysaccharidosis III: Molecular basis and treatment. Pediatric Endocrinology, Diabetes and Metabolism, 2021, 27, 201-208.	0.7	7
149	Family history tools for primary care: A systematic review. European Journal of General Practice, 2022, 28, 75-86.	2.0	7
150	DD Genotype of the angiotensin - converting enzyme gene and stroke in Slovenian population. Pflugers Archiv European Journal of Physiology, 2000, 439, r038-r039.	2.8	6
151	Analysis of the hemochromatosis mutations C282Y and H63D in infertile men. Fertility and Sterility, 2006, 86, 1796-1798.	1.0	6
152	Polymorphisms in genes involved in folate metabolism as maternal risk factors for Down syndrome â€" meta-analysis. Open Medicine (Poland), 2009, 4, 395-408.	1.3	6
153	Direct-to-consumer genetic testing in Slovenia: availability, ethical dilemmas and legislation. Biochemia Medica, 2015, 25, 84-89.	2.7	6
154	Genetic testing offer for inherited neuromuscular diseases within the EURO-NMD reference network: A European survey study. PLoS ONE, 2020, 15, e0239329.	2.5	6
155	Stationary and Progressive Phenotypes Caused by the p.G90D Mutation in Rhodopsin Gene. International Journal of Molecular Sciences, 2021, 22, 2133.	4.1	6
156	Pharmacogenomic Considerations of the Insertion / Deletion Gene Polymorphism of the Angiotensin I-Converting Enzyme and Coronary Artery Disease. Current Vascular Pharmacology, 2004, 2, 271-279.	1.7	6
157	Deletion/insertion polymorphism in the angiotension - converting enzyme gene as a risk factor in the Slovenian patients with coronary heart disease. Pflugers Archiv European Journal of Physiology, 2000, 439, r040-r041.	2.8	5
158	Incidence of the 35delG/GJB2 mutation in low-risk newborns. Journal of Maternal-Fetal and Neonatal Medicine, 2008, 21, 463-468.	1.5	5
159	Angiotensin-Converting Enzyme Gene Polymorphism in Patients with Multiple Sclerosis from Bosnia and Herzegovina. Genetic Testing and Molecular Biomarkers, 2011, 15, 835-838.	0.7	5
160	Integration of Data from Omic Studies with the Literature-Based Discovery towards Identification of Novel Treatments for Neovascularization in Diabetic Retinopathy. BioMed Research International, 2013, 2013, 1-7.	1.9	5
161	Genetic variation in leptin and leptin receptor genes is a risk factor for idiopathic recurrent spontaneous abortion. Croatian Medical Journal, 2016, 57, 566-571.	0.7	5
162	Telegenetics. Current Opinion in Pediatrics, 2020, 32, 739-741.	2.0	5

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163	Whole-Genome Sequencing in Diagnostics of Selected Slovenian Undiagnosed Patients with Rare Disorders. Life, 2021, 11, 205.	2.4	5
164	A Systematic Review of Parkinsonâ€™s Disease Pharmacogenomics: Is There Time for Translation into the Clinics?. International Journal of Molecular Sciences, 2021, 22, 7213.	4.1	5
165	Integrative â€˜Omicâ€™ Approach Towards Understanding the Nature of Human Diseases. Balkan Journal of Medical Genetics, 2012, 15, 45-50.	0.5	5
166	Syndromic male subfertility: A network view of genomeâ€“phenome associations. Andrology, 2022, 10, 720-732.	3.5	5
167	Fibrinogen Polymorphisms Taq I, Hae III and Bcl I Are Not Associated with a Higher Risk of Deep Vein Thrombosis. Pathophysiology of Haemostasis and Thrombosis: International Journal on Haemostasis and Thrombosis Research, 2003, 33, 164-169.	0.3	4
168	The insulin-like growth factor 2 receptor gene Gly1619Arg polymorphism and idiopathic recurrent spontaneous abortion. Journal of Maternal-Fetal and Neonatal Medicine, 2012, 25, 429-431.	1.5	4
169	Brachytelephalangic chondrodysplasia punctata caused by new small hemizygous deletion in a boy presenting with hearing loss. Molecular Cytogenetics, 2015, 8, 83.	0.9	4
170	Angiotensin-converting enzyme insertion/deletion gene polymorphism in multiple sclerosis: a meta-analysis. Neurological Sciences, 2016, 37, 1955-1959.	1.9	4
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