

Borut Peterlin

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/1352779/publications.pdf>

Version: 2024-02-01

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	Maternal LINE-1 DNA Methylation in Early Spontaneous Preterm Birth. <i>Biological Research for Nursing</i> , 2022, 24, 85-93.	1.9	2
2	A case report of a novel GNB1 pathogenic variant and the response to deep brain stimulation. <i>Acta Neurologica Belgica</i> , 2022, , 1.	1.1	3
3	Syndromic male subfertility: A network view of genomeâ€“phenome associations. <i>Andrology</i> , 2022, 10, 720-732.	3.5	5
4	Axonal Polyneuropathy in 2 Brothers With a Homozygous Missense Variant in the First Catalytic Domain of <i>PCYT2</i> . <i>Neurology: Genetics</i> , 2022, 8, e658.	1.9	2
5	Editorial: The Importance of Genetic Literacy and Education in Medicine. <i>Frontiers in Genetics</i> , 2022, 13, 910530.	2.3	0
6	Biallelic <i>ATOH1</i> Gene Variant in Siblings With Pontocerebellar Hypoplasia, Developmental Delay, and Hearing Loss. <i>Neurology: Genetics</i> , 2022, 8, e677.	1.9	2
7	Family history tools for primary care: A systematic review. <i>European Journal of General Practice</i> , 2022, 28, 75-86.	2.0	7
8	Clinical exome sequencing in Serbian patients with movement disorders: Single centre experience. <i>Genetika</i> , 2022, 54, 395-409.	0.4	1
9	The Endometrial Transcriptome of Metabolic and Inflammatory Pathways During the Window of Implantation Is Deranged in Infertile Obese Polycystic Ovarian Syndrome Women. <i>Metabolic Syndrome and Related Disorders</i> , 2022, 20, 384-394.	1.3	4
10	De novo mutations in idiopathic male infertilityâ€“A pilot study. <i>Andrology</i> , 2021, 9, 212-220.	3.5	19
11	Improving diagnostics of rare genetic diseases with NGS approaches. <i>Journal of Community Genetics</i> , 2021, 12, 247-256.	1.2	25
12	Stationary and Progressive Phenotypes Caused by the p.G90D Mutation in Rhodopsin Gene. <i>International Journal of Molecular Sciences</i> , 2021, 22, 2133.	4.1	6
13	Cone Dystrophy Associated with a Novel Variant in the Terminal Codon of the RPGR-ORF15. <i>Genes</i> , 2021, 12, 499.	2.4	8
14	How to design a national genomic projectâ€“a systematic review of active projects. <i>Human Genomics</i> , 2021, 15, 20.	2.9	12
15	Whole-Genome Sequencing in Diagnostics of Selected Slovenian Undiagnosed Patients with Rare Disorders. <i>Life</i> , 2021, 11, 205.	2.4	5
16	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100024.	1.7	1
17	Diagnostic yield of exome sequencing in myopathies: Experience of a Slovenian tertiary centre. <i>PLoS ONE</i> , 2021, 16, e0252953.	2.5	7
18	Identification of women at risk for hereditary breast and ovarian cancer in a sample of 1000 Slovenian women: a comparison of guidelines. <i>BMC Cancer</i> , 2021, 21, 665.	2.6	0

#	ARTICLE	IF	CITATIONS
19	A Systematic Review of Parkinson's Disease Pharmacogenomics: Is There Time for Translation into the Clinics?. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7213.	4.1	5
20	MYH7-related disorders in two Bulgarian families: Novel variants in the same region associated with different clinical manifestation and disease penetrance. <i>Neuromuscular Disorders</i> , 2021, 31, 633-641.	0.6	2
21	Disease Progression in CNGA3 and CNGB3 Retinopathy; Characteristics of Slovenian Cohort and Proposed OCT Staging Based on Pooled Data from 126 Patients from 7 Studies. <i>Current Issues in Molecular Biology</i> , 2021, 43, 941-957.	2.4	4
22	Increasing Genomic Literacy Through National Genomic Projects. <i>Frontiers in Genetics</i> , 2021, 12, 693253.	2.3	12
23	Potential protective role of a NOD2 polymorphism in the susceptibility to multiple sclerosis is not associated with interferon therapy. <i>Biomedical Reports</i> , 2021, 15, 100.	2.0	0
24	Editorial: Prognostication of Heart Failure Evolution: From Circulating Biomarkers to Genetic Risk Predictive Score. <i>Frontiers in Cardiovascular Medicine</i> , 2021, 8, 687232.	2.4	1
25	Clinical and Histopathological Features of Gelsolin Amyloidosis Associated with a Novel GSN Variant p.Glu580Lys. <i>International Journal of Molecular Sciences</i> , 2021, 22, 1084.	4.1	8
26	Mucopolysaccharidosis III: Molecular basis and treatment. <i>Pediatric Endocrinology, Diabetes and Metabolism</i> , 2021, 27, 201-208.	0.7	7
27	Children with cavernous malformations of the central nervous system. <i>European Journal of Paediatric Neurology</i> , 2021, 35, 61-66.	1.6	2
28	MicroRNA-Target Interaction Regulatory Network in Alzheimer's Disease. <i>Journal of Personalized Medicine</i> , 2021, 11, 1275.	2.5	11
29	Current State of Compulsory Basic and Clinical Courses in Genetics for Medical Students at Medical Faculties in Balkan Countries With Slavic Languages. <i>Frontiers in Genetics</i> , 2021, 12, 793834.	2.3	0
30	Lack of association between C282Y and H63D polymorphisms in the hemochromatosis gene and risk of multiple sclerosis: A meta-analysis. <i>Biomedical Reports</i> , 2021, 16, 12.	2.0	2
31	Diagnostic and Clinical Utility of Clinical Exome Sequencing in Children With Moderate and Severe Global Developmental Delay / Intellectual Disability. <i>Journal of Child Neurology</i> , 2020, 35, 116-131.	1.4	22
32	Genetic testing offer for inherited neuromuscular diseases within the EURO-NMD reference network: A European survey study. <i>PLoS ONE</i> , 2020, 15, e0239329.	2.5	6
33	Telegenetics. <i>Current Opinion in Pediatrics</i> , 2020, 32, 739-741.	2.0	5
34	The Role of microRNAs in Heart Failure: A Systematic Review. <i>Frontiers in Cardiovascular Medicine</i> , 2020, 7, 161.	2.4	23
35	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 153-166.	2.6	18
36	Genetics-Related Activities in Everyday Practice of Family Physicians in Slovenia. <i>Public Health Genomics</i> , 2020, 23, 230-236.	1.0	1

#	ARTICLE	IF	CITATIONS
37	Development of an Algorithm for Determining of Genetic Risk at the Primary Healthcare Level – A New Tool for Primary Prevention: A Study Protocol. <i>Zdravstveno Varstvo</i> , 2020, 59, 27-32.	0.9	3
38	DNMT3B rs1569686 and rs2424913 gene polymorphisms are associated with positive family history of preterm birth and smoking status. <i>Croatian Medical Journal</i> , 2020, 61, 8-17.	0.7	8
39	Title is missing!. , 2020, 15, e0239329.		0
40	Title is missing!. , 2020, 15, e0239329.		0
41	Title is missing!. , 2020, 15, e0239329.		0
42	Title is missing!. , 2020, 15, e0239329.		0
43	Actionable Pharmacogenetic Variation in the Slovenian Genomic Database. <i>Frontiers in Pharmacology</i> , 2019, 10, 240.	3.5	10
44	Combination of QF-PCR and aCGH is an efficient diagnostic strategy for the detection of chromosome aberrations in recurrent miscarriage. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e980.	1.2	7
45	Vaginal Microbiome Signature Is Associated With Spontaneous Preterm Delivery. <i>Frontiers in Medicine</i> , 2019, 6, 201.	2.6	71
46	Multiple Sclerosis patients carry an increased burden of exceedingly rare genetic variants in the inflammasome regulatory genes. <i>Scientific Reports</i> , 2019, 9, 9171.	3.3	33
47	PEDIA: prioritization of exome data by image analysis. <i>Genetics in Medicine</i> , 2019, 21, 2807-2814.	2.4	58
48	The frequency of CNV s in a cohort population of consecutive fetuses with congenital anomalies after the termination of pregnancy. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e658.	1.2	3
49	Circadian clock genes and myocardial infarction in patients with type 2 diabetes mellitus. <i>Gene</i> , 2019, 701, 98-103.	2.2	19
50	Pharmacogenomics of Multiple Sclerosis: A Systematic Review. <i>Frontiers in Neurology</i> , 2019, 10, 134.	2.4	11
51	Circadian clock genes and circadian phenotypes in patients with myocardial infarction. <i>Advances in Medical Sciences</i> , 2019, 64, 224-229.	2.1	18
52	MiRNA as biomarker for uveitis - A systematic review of the literature. <i>Gene</i> , 2019, 696, 162-175.	2.2	16
53	The role of circadian rhythm in male reproduction. <i>Current Opinion in Endocrinology, Diabetes and Obesity</i> , 2019, 26, 313-316.	2.3	14
54	Rare missense TUBGCP5 gene variant in a patient with primary microcephaly. <i>European Journal of Medical Genetics</i> , 2019, 62, 103598.	1.3	22

#	ARTICLE	IF	CITATIONS
55	MON-220 Androstendione Affects Endometrial Gene Expression Profile During Window of Implantation in Obese Infertile Women with PCOS. <i>Journal of the Endocrine Society</i> , 2019, 3, .	0.2	1
56	Mutations in <i>SCN3A</i> cause early infantile epileptic encephalopathy. <i>Annals of Neurology</i> , 2018, 83, 703-717.	5.3	69
57	Nonsyndromic cleft palate: An association study at GWAS candidate loci in a multiethnic sample. <i>Birth Defects Research</i> , 2018, 110, 871-882.	1.5	11
58	Sarcoidosis Related Novel Candidate Genes Identified by Multi-Omics Integrative Analyses. <i>OMICS A Journal of Integrative Biology</i> , 2018, 22, 322-331.	2.0	10
59	Transcriptomic Biomarkers for Huntington's Disease: Are Gene Expression Signatures in Whole Blood Reliable Biomarkers?. <i>OMICS A Journal of Integrative Biology</i> , 2018, 22, 283-294.	2.0	9
60	Diagnostic exome sequencing of syndromic epilepsy patients in clinical practice. <i>Clinical Genetics</i> , 2018, 93, 1057-1062.	2.0	39
61	Diagnostic efficacy and new variants in isolated and complex autism spectrum disorder using molecular karyotyping. <i>Journal of Applied Genetics</i> , 2018, 59, 179-185.	1.9	12
62	The challenges of the expanded availability of genomic information: an agenda-setting paper. <i>Journal of Community Genetics</i> , 2018, 9, 103-116.	1.2	45
63	Comprehensive use of extended exome analysis improves diagnostic yield in rare disease: a retrospective survey in 1,059 cases. <i>Genetics in Medicine</i> , 2018, 20, 303-312.	2.4	57
64	Association of Circadian Rhythm with Myocardial Infarction. <i>Acta Clinica Croatica</i> , 2018, 57, 480-486.	0.2	4
65	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. <i>American Journal of Human Genetics</i> , 2018, 103, 995-1008.	6.2	92
66	Diagnostic Testing in Epilepsy Genetics Clinical Practice. , 2018, , .		0
67	Association between angiotensin-converting enzyme gene insertion/deletion polymorphism and susceptibility to preterm birth: A case-control study and meta-analysis. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2018, 231, 122-128.	1.1	2
68	DNA Methylation Profiles in Whole Blood of Huntington's Disease Patients. <i>Frontiers in Neurology</i> , 2018, 9, 655.	2.4	18
69	Genetic variations in circadian rhythm genes and susceptibility for myocardial infarction. <i>Genetics and Molecular Biology</i> , 2018, 41, 403-409.	1.3	21
70	Microduplication in the 2p16.1p15 chromosomal region linked to developmental delay and intellectual disability. <i>Molecular Cytogenetics</i> , 2018, 11, 39.	0.9	4
71	Genetic variation in the CLOCK gene is associated with idiopathic recurrent spontaneous abortion. <i>PLoS ONE</i> , 2018, 13, e0196345.	2.5	16
72	Diagnostic outcomes of exome sequencing in patients with syndromic or non-syndromic hearing loss. <i>PLoS ONE</i> , 2018, 13, e0188578.	2.5	33

#	ARTICLE	IF	CITATIONS
73	Association of circadian rhythm genes ARNTL/BMAL1 and CLOCK with multiple sclerosis. PLoS ONE, 2018, 13, e0190601.	2.5	34
74	Screening for Rare Genetic Variants Associated with Atherosclerosis: Opportunity for Personalized Medicine. Current Vascular Pharmacology, 2018, 17, 25-28.	1.7	7
75	The lack of association between angiotensin-converting enzyme gene insertion/deletion polymorphism and nicotine dependence in multiple sclerosis. Brain and Behavior, 2017, 7, e00600.	2.2	3
76	Angiotensin-converting enzyme insertion/deletion gene polymorphism and interferon- β treatment response in multiple sclerosis patients. Pharmacogenetics and Genomics, 2017, 27, 232-235.	1.5	7
77	Identification of rare genetic variation of NLRP1 gene in familial multiple sclerosis. Scientific Reports, 2017, 7, 3715.	3.3	53
78	Telegenetics: an Update on Availability and Use of Telemedicine in Clinical Genetics Service. Journal of Medical Systems, 2017, 41, 21.	3.6	46
79	Functional single nucleotide polymorphisms of matrix metalloproteinase 7 and 12 genes in idiopathic recurrent spontaneous abortion. Journal of Assisted Reproduction and Genetics, 2017, 34, 365-371.	2.5	2
80	Genetic Interactions in Nonsyndromic Orofacial Clefts in Europe – EUROCRAN Study. Cleft Palate-Craniofacial Journal, 2017, 54, 623-630.	0.9	18
81	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
82	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
83	A Single Nucleotide Polymorphism of <i>DNMT3B</i> gene is a risk factor for recurrent spontaneous abortion. American Journal of Reproductive Immunology, 2017, 78, e12765.	1.2	8
84	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
85	Impact of prenatal screening on the prevalence of Down syndrome in Slovenia. PLoS ONE, 2017, 12, e0180348.	2.5	8
86	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
87	Clinical utility of array comparative genomic hybridisation in prenatal setting. BMC Medical Genetics, 2016, 17, 81.	2.1	16
88	Phenotype-driven gene target definition in clinical genome-wide sequencing data interpretation. Genetics in Medicine, 2016, 18, 1102-1110.	2.4	41
89	Angiotensin-converting enzyme insertion/deletion gene polymorphism in multiple sclerosis: a meta-analysis. Neurological Sciences, 2016, 37, 1955-1959.	1.9	4
90	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

#	ARTICLE	IF	CITATIONS
91	Reply to Sajantila and Budowle. <i>European Journal of Human Genetics</i> , 2016, 24, 330-330.	2.8	1
92	Responsible implementation of expanded carrier screening. <i>European Journal of Human Genetics</i> , 2016, 24, e1-e12.	2.8	240
93	Epigenetics and Bruxism: Possible Role of Epigenetics in the Etiology of Bruxism. <i>International Journal of Prosthodontics</i> , 2015, 28, 594-599.	1.7	15
94	The influence of cytokine gene polymorphisms on the risk of developing gastric cancer in patients with <i>Helicobacter pylori</i> infection. <i>Radiology and Oncology</i> , 2015, 49, 256-264.	1.7	14
95	Brachytelephalangic chondrodysplasia punctata caused by new small hemizygous deletion in a boy presenting with hearing loss. <i>Molecular Cytogenetics</i> , 2015, 8, 83.	0.9	4
96	Evaluating the SERCA2 and VEGF mRNAs as Potential Molecular Biomarkers of the Onset and Progression in Huntington's Disease. <i>PLoS ONE</i> , 2015, 10, e0125259.	2.5	18
97	Towards a European consensus for reporting incidental findings during clinical NGS testing. <i>European Journal of Human Genetics</i> , 2015, 23, 1601-1606.	2.8	85
98	The ~ 2549 insertion/deletion polymorphism in the promoter region of the VEGFA gene in couples with idiopathic recurrent spontaneous abortion. <i>Journal of Assisted Reproduction and Genetics</i> , 2015, 32, 1789-1794.	2.5	9
99	MMP-2 $\sim 1575G/A$ polymorphism modifies the onset of optic neuritis as a first presenting symptom in MS?. <i>Journal of Neuroimmunology</i> , 2015, 286, 13-15.	2.3	8
100	Therapeutic perspectives of epigenetically active nutrients. <i>British Journal of Pharmacology</i> , 2015, 172, 2756-2768.	5.4	99
101	Direct-to-consumer genetic testing in Slovenia: availability, ethical dilemmas and legislation. <i>Biochemia Medica</i> , 2015, 25, 84-89.	2.7	6
102	Cornelia de Lange syndrome caused by heterozygous deletions of chromosome 8q24: Comments on the article by Perez et al. [2012]. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1426-1427.	1.2	8
103	Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening. Summary and recommendations. <i>European Journal of Human Genetics</i> , 2015, , .	2.8	13
104	Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening. <i>European Journal of Human Genetics</i> , 2015, 23, 1438-1450.	2.8	260
105	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. <i>Molecular Human Reproduction</i> , 2015, 21, 466-478.	2.8	14
106	Family history based approach in risk prediction for Parkinson's disease: Additional contribution of familial associated disorders. <i>Genetika</i> , 2015, 47, 303-310.	0.4	0
107	Omics Technologies and Neovascular Ocular Disorders. <i>BioMed Research International</i> , 2014, 2014, 1-2.	1.9	1
108	Functional Polymorphisms of Matrix Metalloproteinases 1 and 9 Genes in Women with Spontaneous Preterm Birth. <i>Disease Markers</i> , 2014, 2014, 1-7.	1.3	13

#	ARTICLE	IF	CITATIONS
109	The Role of TPA I/D and PAI-1 4G/5G Polymorphisms in Multiple Sclerosis. <i>Disease Markers</i> , 2014, 2014, 1-8.	1.3	10
110	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. <i>Genome Biology</i> , 2014, 15, R53.	9.6	101
111	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) <i>Tj ETQq1 1 0.784314 rgBT /Over 2.8 109</i>	2.8	109
112	Strong Association of Variants around <i>FOXE1</i> and Orofacial Clefting. <i>Journal of Dental Research</i> , 2014, 93, 376-381.	5.2	51
113	GSTM1-null and GSTT1-null genotypes are associated with essential arterial hypertension in patients with type 2 diabetes. <i>Clinical Biochemistry</i> , 2014, 47, 574-577.	1.9	17
114	Genetic variation in tissue inhibitors of metalloproteinases as a risk factor for idiopathic recurrent spontaneous abortion. <i>Fertility and Sterility</i> , 2013, 99, 1923-1929.	1.0	7
115	Y chromosome azoospermia factor region microdeletions are not associated with idiopathic recurrent spontaneous abortion in a Slovenian population: association study and literature review. <i>Fertility and Sterility</i> , 2013, 99, 1663-1667.	1.0	9
116	Integration of Data from Omic Studies with the Literature-Based Discovery towards Identification of Novel Treatments for Neovascularization in Diabetic Retinopathy. <i>BioMed Research International</i> , 2013, 2013, 1-7.	1.9	5
117	Analysis of susceptibility loci for nonsyndromic orofacial clefting in a European trio sample. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2545-2549.	1.2	21
118	Correlates of Depression in the Slovenian Working Population. <i>Arhiv Za Higijenu Rada I Toksikologiju</i> , 2013, 64, 489-495.	0.7	7
119	Using Literature-based Discovery to Identify Novel Therapeutic Approaches. <i>Cardiovascular and Hematological Agents in Medicinal Chemistry</i> , 2013, 11, 14-24.	1.0	45
120	Genetic Variation in Circadian Rhythm Genes <i>CLOCK</i> and <i>ARNTL</i> as Risk Factor for Male Infertility. <i>PLoS ONE</i> , 2013, 8, e59220.	2.5	32
121	Family History as a Predictor for Disease Risk in Healthy Individuals: A Cross-Sectional Study in Slovenia. <i>PLoS ONE</i> , 2013, 8, e80333.	2.5	9
122	Expression Signature as a Biomarker for Prenatal Diagnosis of Trisomy 21. <i>PLoS ONE</i> , 2013, 8, e74184.	2.5	27
123	Detection of Thrombophilic Mutations Related to Spontaneous Abortions by a Multiplex SNaPshot Method. <i>Genetic Testing and Molecular Biomarkers</i> , 2012, 16, 259-264.	0.7	12
124	Matrix metalloproteinases 1, 2, 3 and 9 functional single-nucleotide polymorphisms in idiopathic recurrent spontaneous abortion. <i>Reproductive BioMedicine Online</i> , 2012, 24, 567-575.	2.4	33
125	The insulin-like growth factor 2 receptor gene Gly1619Arg polymorphism and idiopathic recurrent spontaneous abortion. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2012, 25, 429-431.	1.5	4
126	Histamine N-methyltransferase Thr105Ile polymorphism is associated with Parkinson's disease. <i>Neurobiology of Aging</i> , 2012, 33, 836.e1-836.e3.	3.1	29

#	ARTICLE	IF	CITATIONS
127	Third case of 8q23.3â€”q24.13 deletion in a patient with Langerâ€”Giedion syndrome phenotype without <i>TRPS1</i> gene deletion. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 659-663.	1.2	20
128	Integrative â€”Omicâ€” Approach Towards Understanding the Nature of Human Diseases. <i>Balkan Journal of Medical Genetics</i> , 2012, 15, 45-50.	0.5	5
129	Association between male infertility and genetic variability at the PON1/2 and GSTM1/T1 gene loci. <i>Reproductive BioMedicine Online</i> , 2011, 23, 105-110.	2.4	26
130	A new case of rare proximal 3q13 interstitial deletion. <i>Open Medicine (Poland)</i> , 2011, 6, 625-630.	1.3	2
131	Angiotensin-Converting Enzyme Gene Polymorphism in Patients with Multiple Sclerosis from Bosnia and Herzegovina. <i>Genetic Testing and Molecular Biomarkers</i> , 2011, 15, 835-838.	0.7	5
132	Positional integratomic approach in identification of genomic candidate regions for Parkinson's disease. <i>Bioinformatics</i> , 2011, 27, 1971-1978.	4.1	15
133	Low frequency of HFE gene mutations in Croatian patients suspected of having hereditary hemochromatosis. <i>Medical Science Monitor</i> , 2011, 17, CR552-CR556.	1.1	2
134	Autonomic dysfunction in presymptomatic and early symptomatic Huntingtonâ€”s disease. <i>Acta Neurologica Scandinavica</i> , 2010, 121, 392-399.	2.1	49
135	SATB2 haploinsufficiency in patients with cleft palate. <i>Open Medicine (Poland)</i> , 2010, 5, 318-321.	1.3	0
136	Rasch-based high-dimensionality data reduction and class prediction with applications to microarray gene expression data. <i>Expert Systems With Applications</i> , 2010, 37, 5178-5185.	7.6	15
137	Chi-square-based Scoring Function for Categorization of MEDLINE Citations. <i>Methods of Information in Medicine</i> , 2010, 49, 371-378.	1.2	13
138	Interleukin 7 receptor alpha polymorphism rs6897932 and susceptibility to multiple sclerosis in the Western Balkans. <i>Multiple Sclerosis Journal</i> , 2010, 16, 533-536.	3.0	11
139	Combining Semantic Relations and DNA Microarray Data for Novel Hypotheses Generation. <i>Lecture Notes in Computer Science</i> , 2010, , 53-61.	1.3	15
140	Lack of association of immune-response-gene polymorphisms with susceptibility to sarcoidosis in Slovenian patients. <i>Genetics and Molecular Research</i> , 2010, 9, 58-68.	0.2	2
141	Interstitial deletion 2p11.2â€”p12: Further delineation. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2324-2326.	1.2	7
142	Gene expression changes in blood as a putative biomarker for Huntington's disease. <i>Movement Disorders</i> , 2009, 24, 2277-2281.	3.9	28
143	Epidemiology of Huntingtonâ€”s disease in Slovenia. <i>Acta Neurologica Scandinavica</i> , 2009, 119, 371-375.	2.1	18
144	Polymorphisms in genes involved in folate metabolism as maternal risk factors for Down syndrome â€” meta-analysis. <i>Open Medicine (Poland)</i> , 2009, 4, 395-408.	1.3	6

#	ARTICLE	IF	CITATIONS
145	Association between genetic polymorphisms in cytokine genes and recurrent miscarriage – a meta-analysis. <i>Reproductive BioMedicine Online</i> , 2009, 19, 406-414.	2.4	24
146	Semantic relations for interpreting DNA microarray data. <i>AMIA ... Annual Symposium proceedings</i> , 2009, 2009, 255-9.	0.2	2
147	Search for sarcoidosis candidate genes by integration of data from genomic, transcriptomic and proteomic studies. <i>Medical Science Monitor</i> , 2009, 15, SR22-8.	1.1	15
148	Distribution of HFE gene mutations in Slovenian patients with hereditary hemochromatosis. <i>Annals of Hematology</i> , 2008, 87, 667-669.	1.8	2
149	Genital anomalies in a patient with Treacher Collins syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2169-2171.	1.2	3
150	Mutations in C2orf37, Encoding a Nucleolar Protein, Cause Hypogonadism, Alopecia, Diabetes Mellitus, Mental Retardation, and Extrapramidal Syndrome. <i>American Journal of Human Genetics</i> , 2008, 83, 684-691.	6.2	121
151	ORIGINAL ARTICLE: Genetic Predisposition to Idiopathic Recurrent Spontaneous Abortion: Contribution of Genetic Variations in IGF2 and H19 Imprinted Genes. <i>American Journal of Reproductive Immunology</i> , 2008, 60, 111-117.	1.2	24
152	Meta analysis: Haplotype or single polymorphism analysis. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2008, 136, 260-261.	1.1	0
153	Incidence of the 35delG/GJB2 mutation in low-risk newborns. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2008, 21, 463-468.	1.5	5
154	Heart-hand syndrome of Slovenian type: a new kind of laminopathy. <i>Journal of Medical Genetics</i> , 2008, 45, 666-671.	3.2	47
155	PAI and TPA gene polymorphisms in multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2008, 14, 243-247.	3.0	13
156	Local and genetic determinants of vascular endothelial growth factor expression in advanced proliferative diabetic retinopathy. <i>Molecular Vision</i> , 2008, 14, 1382-7.	1.1	54
157	Tumor Necrosis Factor-308 Gene Polymorphism in Croatian and Slovenian Multiple Sclerosis Patients. <i>European Neurology</i> , 2007, 57, 203-207.	1.4	22
158	Genetic polymorphisms in vasoactive genes and preeclampsia: A meta-analysis. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2007, 131, 115-126.	1.1	69
159	Epidemiology of multiple sclerosis in western Herzegovina. <i>Clinical Neurology and Neurosurgery</i> , 2007, 109, 779-783.	1.4	15
160	Association of Vascular Endothelial Growth Factor Gene Polymorphism with Myocardial Infarction in Patients with Type 2 Diabetes. <i>Cardiology</i> , 2007, 107, 291-295.	1.4	53
161	Polymorphisms in the Interleukin-12/18 Genes and Recurrent Spontaneous Abortion. <i>American Journal of Reproductive Immunology</i> , 2007, 58, 403-408.	1.2	20
162	Analysis of the CAG repeat number in exon 1 of the androgen receptor gene in Slovene men with idiopathic azoospermia and oligoasthenoteratozoospermia. <i>Asian Journal of Andrology</i> , 2007, 9, 280-282.	1.6	7

#	ARTICLE	IF	CITATIONS
163	Patients with primary cataract as a genetic pool of DMPK protomutation. <i>Journal of Human Genetics</i> , 2007, 52, 123-128.	2.3	17
164	Role of genetic polymorphisms in ACE and TNF- $\hat{\pm}$ gene in sarcoidosis: a meta-analysis. <i>Journal of Human Genetics</i> , 2007, 52, 836-847.	2.3	44
165	Literature Based Discovery Support System and Its Application to Disease Gene Identification. <i>Lecture Notes in Computer Science</i> , 2007, , 307-326.	1.3	3
166	Region with persistent high frequency of multiple sclerosis in Croatia and Slovenia. <i>Journal of the Neurological Sciences</i> , 2006, 247, 169-172.	0.6	21
167	Analysis of the hemochromatosis mutations C282Y and H63D in infertile men. <i>Fertility and Sterility</i> , 2006, 86, 1796-1798.	1.0	6
168	Angiotensin-converting enzyme I/D gene polymorphism and risk of multiple sclerosis. <i>Acta Neurologica Scandinavica</i> , 2006, 114, 374-377.	2.1	20
169	Preliminary analysis of AZFb region duplication by quantitative real-time PCR. <i>Human Reproduction</i> , 2006, 21, 753-754.	0.9	2
170	No association of CCR5D32 gene mutation with multiple sclerosis in Croatian and Slovenian patients. <i>Multiple Sclerosis Journal</i> , 2006, 12, 360-362.	3.0	15
171	Association between the apolipoprotein B signal peptide gene insertion/deletion polymorphism and male infertility. <i>Molecular Human Reproduction</i> , 2006, 12, 777-779.	2.8	15
172	Exploiting semantic relations for literature-based discovery. <i>AMIA ... Annual Symposium proceedings</i> , 2006, , 349-53.	0.2	59
173	Oestrogen receptor dinucleotide (TA) polymorphism affects total and LDL cholesterol but does not predict premature myocardial infarction in Caucasian men. <i>Acta Cardiologica</i> , 2005, 60, 307-309.	0.9	1
174	Human Y-specific STR haplotypes in the Western Croatian population sample. <i>Forensic Science International</i> , 2005, 149, 257-261.	2.2	17
175	Familial progressive sinoatrial and atrioventricular conduction disease of adult onset with sudden death, dilated cardiomyopathy, and brachydactyly. A new type of heart-hand syndrome?. <i>Clinical Genetics</i> , 2005, 68, 155-160.	2.0	21
176	Y microdeletions in the Istria county, Croatia. <i>Asian Journal of Andrology</i> , 2005, 7, 213-216.	1.6	15
177	Using literature-based discovery to identify disease candidate genes. <i>International Journal of Medical Informatics</i> , 2005, 74, 289-298.	3.3	219
178	Aldose reductase (AC)n gene polymorphism and susceptibility to diabetic retinopathy in Type 2 diabetes in Caucasians. <i>Journal of Diabetes and Its Complications</i> , 2005, 19, 70-73.	2.3	33
179	Gly482Ser polymorphism of the peroxisome proliferator-activated receptor- $\hat{\gamma}$ 3 coactivator-1 gene might be a risk factor for diabetic retinopathy in Slovene population (Caucasians) with type 2 diabetes and the Pro12Ala polymorphism of thePPAR $\hat{\gamma}$ 3 gene is not. <i>Diabetes/Metabolism Research and Reviews</i> , 2005, 21, 470-474.	4.0	28
180	Nonsense Mutations in Folliculin Presenting as Isolated Familial Spontaneous Pneumothorax in Adults. <i>American Journal of Respiratory and Critical Care Medicine</i> , 2005, 172, 39-44.	5.6	104

#	ARTICLE	IF	CITATIONS
181	Copy number of DAZ genes in infertile men. <i>Fertility and Sterility</i> , 2005, 84, 1522-1525.	1.0	34
182	Mutations in the hemochromatosis gene (HFE) and multiple sclerosis. <i>Neuroscience Letters</i> , 2005, 383, 301-304.	2.1	22
183	Genetic markers of restenosis after coronary angioplasty and after stent implantation. <i>Medical Science Monitor</i> , 2005, 11, RA127-35.	1.1	12
184	Sex Difference in the Effect of ACE-DD Genotype on the Risk of Premature Myocardial Infarction. <i>Angiology</i> , 2004, 55, 155-158.	1.8	21
185	CTG amplification in the DM1PK gene is not associated with idiopathic male subfertility. <i>Human Reproduction</i> , 2004, 19, 2084-2087.	0.9	10
186	Early sympathetic hyperactivity in Huntington's disease. <i>European Journal of Neurology</i> , 2004, 11, 842-848.	3.3	63
187	Apolipoprotein E gene polymorphism effects triglycerides but not CAD risk in Caucasian women younger than 65 years. <i>Annales De G�n�tologie</i> , 2004, 47, 147-153.	0.4	10
188	Pharmacogenomic Considerations of the Insertion / Deletion Gene Polymorphism of the Angiotensin I-Converting Enzyme and Coronary Artery Disease. <i>Current Vascular Pharmacology</i> , 2004, 2, 271-279.	1.7	6
189	BgIII gene polymorphism of the $\alpha_2\beta_1$ integrin gene is a risk factor for diabetic retinopathy in Caucasians with type 2 diabetes. <i>Journal of Human Genetics</i> , 2003, 48, 457-460.	2.3	23
190	A hemochromatosis-causing mutation C282Y is a risk factor for proliferative diabetic retinopathy in Caucasians with type 2 diabetes. <i>Journal of Human Genetics</i> , 2003, 48, 646-649.	2.3	24
191	Hemochromatosis gene mutations in the Croatian and Slovenian populations. <i>Clinical Genetics</i> , 2003, 64, 444-446.	2.0	17
192	Prothrombotic gene polymorphisms and atherothrombotic cerebral infarction. <i>Acta Neurologica Scandinavica</i> , 2003, 108, 109-113.	2.1	18
193	Y chromosome microdeletions in infertile men with cryptorchidism. <i>Fertility and Sterility</i> , 2003, 79, 1559-1565.	1.0	20
194	Insertion/Deletion Plasminogen Activator Inhibitor 1 and Insertion/Deletion Angiotensin-Converting Enzyme Gene Polymorphisms in Diabetic Retinopathy in Type 2 Diabetes. <i>Ophthalmologica</i> , 2003, 217, 219-224.	1.9	27
195	Factor V Leiden, prothrombin 20210G \rightarrow A, methylenetetrahydrofolate reductase 677C \rightarrow T and plasminogen activator inhibitor 4G/5G polymorphism in women with pregnancy-related venous thromboembolism. <i>European Journal of Obstetrics, Gynecology and Reproductive Biology</i> , 2003, 111, 157-163.	1.1	30
196	Relation of CTG expansion and clinical variables to electrocardiogram conduction abnormalities and sudden death in patients with myotonic dystrophy. <i>Neuromuscular Disorders</i> , 2003, 13, 822-826.	0.6	27
197	Estrogen Receptor Dinucleotide (TA) Polymorphism Does Not Predict Premature Myocardial Infarction in Caucasian Women. <i>Cardiology</i> , 2003, 99, 163-165.	1.4	9
198	Fragile X premutation in women with sporadic premature ovarian failure in Slovenia. <i>Human Reproduction</i> , 2003, 18, 1637-1640.	0.9	36

#	ARTICLE	IF	CITATIONS
199	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. <i>Klinische Monatsblätter Für Augenheilkunde</i> , 2003, 220, 873-876.	0.5	30
200	4G4G Genotype of PAI-1 Gene Promoter Polymorphism Is Not Associated with Myocardial Infarction in Caucasians with Type-2 Diabetes. <i>Cardiology</i> , 2003, 100, 157-158.	1.4	11
201	Fibrinogen Polymorphisms <i>Taq</i> I, <i>Hae</i> III and <i>Bcl</i> I Are Not Associated with a Higher Risk of Deep Vein Thrombosis. <i>Pathophysiology of Haemostasis and Thrombosis: International Journal on Haemostasis and Thrombosis Research</i> , 2003, 33, 164-169.	0.3	4
202	Improving literature based discovery support by genetic knowledge integration. <i>Studies in Health Technology and Informatics</i> , 2003, 95, 68-73.	0.3	25
203	Screening for Y chromosome microdeletions in 226 Slovenian subfertile men. <i>Human Reproduction</i> , 2002, 17, 17-24.	0.9	82
204	Single signal of the Williams syndrome chromosome region 1 gene in hyperpliodic bone marrow cells of acute lymphoblastic leukemia in a Williams syndrome patient. <i>Medical and Pediatric Oncology</i> , 2002, 38, 205-207.	1.0	12
205	Interaction Between Gene Polymorphisms of Renin-angiotensin System and Metabolic Risk Factors in Premature Myocardial Infarction. <i>Angiology</i> , 2001, 52, 247-252.	1.8	24
206	Involvement of CFTR Gene Alterations in Obstructive and Nonobstructive Infertility in Men. <i>Genetic Testing and Molecular Biomarkers</i> , 2001, 5, 243-247.	1.7	31
207	The Str mouse as a model for Incontinentia pigmenti. <i>Pflugers Archiv European Journal of Physiology</i> , 2000, 440, R053-R054.	2.8	1
208	Angiotensin-converting enzyme gene polymorphism as a cardiovascular risk factor in children. <i>Pflugers Archiv European Journal of Physiology</i> , 2000, 439, r034-r035.	2.8	1
209	DD Genotype of the angiotensin - converting enzyme gene and stroke in Slovenian population. <i>Pflugers Archiv European Journal of Physiology</i> , 2000, 439, r038-r039.	2.8	6
210	Deletion/insertion polymorphism in the angiotension - converting enzyme gene as a risk factor in the Slovenian patients with coronary heart disease. <i>Pflugers Archiv European Journal of Physiology</i> , 2000, 439, r040-r041.	2.8	5
211	Y-Chromosomal Diversity in Europe Is Clinal and Influenced Primarily by Geography, Rather than by Language. <i>American Journal of Human Genetics</i> , 2000, 67, 1526-1543.	6.2	519
212	Deletion/insertion polymorphism in the angiotension " converting enzyme gene as a risk factor in the Slovenian patients with coronary heart disease. <i>Pflugers Archiv European Journal of Physiology</i> , 2000, 439, R40-R41.	2.8	1
213	DD Genotype of the angiotensin " converting enzyme gene and stroke in Slovenian population. <i>Pflugers Archiv European Journal of Physiology</i> , 2000, 439, R38-R39.	2.8	0
214	Loss of heterozygosity at 11q23.1 and survival in breast cancer: Results of a large European study. <i>Genes Chromosomes and Cancer</i> , 1999, 25, 212-221.	2.8	34
215	Bilateral Macular Dysplasia in Fragile X Syndrome. <i>Optometry and Vision Science</i> , 1998, 75, 856-859.	1.2	2
216	Rapid DNA-based prenatal diagnosis by genetic linkage in three families with Alport's syndrome. <i>American Journal of Kidney Diseases</i> , 1997, 30, 174-179.	1.9	9

#	ARTICLE	IF	CITATIONS
217	Genetic epidemiology of myotonic dystrophy in Istria, Croatia. <i>Acta Neurologica Scandinavica</i> , 1997, 95, 164-166.	2.1	23
218	Genetic epidemiology of Duchenne and Becker muscular dystrophy in Slovenia. <i>Clinical Genetics</i> , 1997, 51, 94-97.	2.0	23
219	The gene for the familial form of incontinentia pigmenti (IP2) maps to the distal part of Xq28. <i>Human Molecular Genetics</i> , 1994, 3, 273-278.	2.9	71