AleÅ¡ Maver

List of Publications by Year
in descending order

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Version: 2024-02-01

60 papers	1,082 citations	430874 18 h-index	454955 30 g-index
60 all docs	60 docs citations	60 times ranked	2533 citing authors

#	Article	IF	Citations
1	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
2	Axonal Polyneuropathy in 2 Brothers With a Homozygous Missense Variant in the First Catalytic Domain of PCYT2. Neurology: Genetics, 2022, 8, e658.	1.9	2
3	Biallelic ATOH1 Gene Variant in Siblings With Pontocerebellar Hypoplasia, Developmental Delay, and Hearing Loss. Neurology: Genetics, 2022, 8, e677.	1.9	2
4	Clinical exome sequencing in Serbian patients with movement disorders: Single centre experience. Genetika, 2022, 54, 395-409.	0.4	1
5	Front Cover, Volume 43, Issue 8. Human Mutation, 2022, 43, .	2.5	0
6	Human Mutation special issue on â€œVariant Effect Prediction". Human Mutation, 2022, 43, 973-975.	2.5	0
7	The Endometrial Transcriptome of Metabolic and Inflammatory Pathways During the Window of Implantation Is Deranged in Infertile Obese Polycystic Ovarian Syndrome Women. Metabolic Syndrome and Related Disorders, 2022, 20, 384-394.	1.3	4
8	De novo mutations in idiopathic male infertilityâ€”A pilot study. Andrology, 2021, 9, 212-220.	3.5	19
9	Improving diagnostics of rare genetic diseases with NGS approaches. Journal of Community Genetics, 2021, 12, 247-256.	1.2	25
10	Cone Dystrophy Associated with a Novel Variant in the Terminal Codon of the RPGR-ORF15. Genes, 2021, 12, 499.	2.4	8
11	Whole-Genome Sequencing in Diagnostics of Selected Slovenian Undiagnosed Patients with Rare Disorders. Life, 2021, 11, 205.	2.4	5
12	Diagnostic yield of exome sequencing in myopathies: Experience of a Slovenian tertiary centre. PLoS ONE, 2021, 16, e0252953.	2.5	7
13	MYH7-related disorders in two Bulgarian families: Novel variants in the same region associated with different clinical manifestation and disease penetrance. Neuromuscular Disorders, 2021, 31, 633-641.	0.6	2
14	Identification of Variants Associated With Rare Hematological Disorder Erythrocytosis Using Targeted Next-Generation Sequencing Analysis. Frontiers in Genetics, 2021, 12, 689868.	2.3	5
15	Potential protective role of a NOD2 polymorphism in the susceptibility to multiple sclerosis is not associated with interferon therapy. Biomedical Reports, 2021, 15, 100.	2.0	0
16	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
17	Children with cavernous malformations of the central nervous system. European Journal of Paediatric Neurology, 2021, 35, 61-66.	1.6	2
18	Transcriptome study of receptive endometrium in overweight and obese women shows important expression differences in immune response and inflammatory pathways in women who do not conceive. PLoS ONE, 2021, 16, e0261873.	2.5	4

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19	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
20	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
21	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	2.6	18
22	Prevalence and genetic subtypes of congenital myasthenic syndromes in the pediatric population of Slovenia. European Journal of Paediatric Neurology, 2020, 26, 34-38.	1.6	8
23	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	6.2	17
24	Title is missing!. , 2020, 15, e0239329.		0
25	Title is missing!. , 2020, 15, e0239329.		0
26	Title is missing!. , 2020, 15, e0239329.		0
27	Title is missing!. , 2020, 15, e0239329.		0
28	Actionable Pharmacogenetic Variation in the Slovenian Genomic Database. Frontiers in Pharmacology, 2019, 10, 240.	3.5	10
29	Vaginal Microbiome Signature Is Associated With Spontaneous Preterm Delivery. Frontiers in Medicine, 2019, 6, 201.	2.6	71
30	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
31	Rare missense TUBGCP5 gene variant in a patient with primary microcephaly. European Journal of Medical Genetics, 2019, 62, 103598.	1.3	22
32	MON-220 Androstendione Affects Endometrial Gene Expression Profile During Window of Implantation in Obese Infertile Women with PCOS. Journal of the Endocrine Society, 2019, 3, .	0.2	1
33	Mutations in SCN3A cause early infantile epileptic encephalopathy. Annals of Neurology, 2018, 83, 703-717.	5.3	69
34	Sarcoidosis Related Novel Candidate Genes Identified by Multi-Omics Integrative Analyses. OMICS A Journal of Integrative Biology, 2018, 22, 322-331.	2.0	10
35	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
36	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

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37	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
38	Diagnostic Testing in Epilepsy Genetics Clinical Practice. , 2018, , .		0
39	DNA Methylation Profiles in Whole Blood of Huntington's Disease Patients. Frontiers in Neurology, 2018, 9, 655.	2.4	18
40	Genomic Testing for Prenatal Clinical Evaluation of Congenital Anomalies. , 2018, , .		0
41	Diagnostic outcomes of exome sequencing in patients with syndromic or non-syndromic hearing loss. PLoS ONE, 2018, 13, e0188578.	2.5	33
42	Association of circadian rhythm genes ARNTL/BMAL1 and CLOCK with multiple sclerosis. PLoS ONE, 2018, 13, e0190601.	2.5	34
43	Identification of rare genetic variation of NLRP1 gene in familial multiple sclerosis. Scientific Reports, 2017, 7, 3715.	3.3	53
44	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
45	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
46	Survival among children with â€œLethalâ€•congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (GLDN). Human Mutation, 2017, 38, 1477-1484.	2.5	19
47	Phenotype-driven gene target definition in clinical genome-wide sequencing data interpretation. Genetics in Medicine, 2016, 18, 1102-1110.	2.4	41
48	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
49	Family history based approach in risk prediction for Parkinson's disease: Additional contribution of familial associated disorders. Genetika, 2015, 47, 303-310.	0.4	0
50	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
51	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
52	Genetic Variation in Circadian Rhythm Genes CLOCK and ARNTL as Risk Factor for Male Infertility. PLoS ONE, 2013, 8, e59220.	2.5	32
53	Expression Signature as a Biomarker for Prenatal Diagnosis of Trisomy 21. PLoS ONE, 2013, 8, e74184.	2.5	27
54	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

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55	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
56	Genome Profiling and Potential Biomarkers in Neurodegenerative Disorders. , 2011, , .		0
57	Positional integratomic approach in identification of genomic candidate regions for Parkinson's disease. Bioinformatics, 2011, 27, 1971-1978.	4.1	15
58	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
59	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
60	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