

AleÅ; Maver

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7097616/publications.pdf>

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	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
2	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
3	Vaginal Microbiome Signature Is Associated With Spontaneous Preterm Delivery. <i>Frontiers in Medicine</i> , 2019, 6, 201.	2.6	71
4	Mutations in <i>SCN3A</i> cause early infantile epileptic encephalopathy. <i>Annals of Neurology</i> , 2018, 83, 703-717.	5.3	69
5	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
6	Identification of rare genetic variation of NLRP1 gene in familial multiple sclerosis. <i>Scientific Reports</i> , 2017, 7, 3715.	3.3	53
7	De Novo Mutations in SLC25A24 Cause a Disorder Characterized by Early Aging, Bone Dysplasia, Characteristic Face, and Early Demise. <i>American Journal of Human Genetics</i> , 2017, 101, 844-855.	6.2	51
8	Role of genetic polymorphisms in ACE and TNF- α gene in sarcoidosis: a meta-analysis. <i>Journal of Human Genetics</i> , 2007, 52, 836-847.	2.3	44
9	Phenotype-driven gene target definition in clinical genome-wide sequencing data interpretation. <i>Genetics in Medicine</i> , 2016, 18, 1102-1110.	2.4	41
10	Association of circadian rhythm genes ARNTL/BMAL1 and CLOCK with multiple sclerosis. <i>PLoS ONE</i> , 2018, 13, e0190601.	2.5	34
11	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
12	Diagnostic outcomes of exome sequencing in patients with syndromic or non-syndromic hearing loss. <i>PLoS ONE</i> , 2018, 13, e0188578.	2.5	33
13	Genetic Variation in Circadian Rhythm Genes CLOCK and ARNTL as Risk Factor for Male Infertility. <i>PLoS ONE</i> , 2013, 8, e59220.	2.5	32
14	Expression Signature as a Biomarker for Prenatal Diagnosis of Trisomy 21. <i>PLoS ONE</i> , 2013, 8, e74184.	2.5	27
15	Improving diagnostics of rare genetic diseases with NGS approaches. <i>Journal of Community Genetics</i> , 2021, 12, 247-256.	1.2	25
16	Rare missense TUBGCP5 gene variant in a patient with primary microcephaly. <i>European Journal of Medical Genetics</i> , 2019, 62, 103598.	1.3	22
17	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
18	Third case of 8q23.3-q24.13 deletion in a patient with Langer-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

#	ARTICLE	IF	CITATIONS
19	Survival among children with "Lethal" congenital contracture syndrome 11 caused by novel mutations in the gliomedin gene (<i>GLDN</i>). <i>Human Mutation</i> , 2017, 38, 1477-1484.	2.5	19
20	De novo mutations in idiopathic male infertility" A pilot study. <i>Andrology</i> , 2021, 9, 212-220.	3.5	19
21	DNA Methylation Profiles in Whole Blood of Huntington's Disease Patients. <i>Frontiers in Neurology</i> , 2018, 9, 655.	2.4	18
22	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
23	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. <i>American Journal of Human Genetics</i> , 2020, 106, 830-845.	6.2	17
24	Positional integratomic approach in identification of genomic candidate regions for Parkinson's disease. <i>Bioinformatics</i> , 2011, 27, 1971-1978.	4.1	15
25	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
26	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
27	Actionable Pharmacogenetic Variation in the Slovenian Genomic Database. <i>Frontiers in Pharmacology</i> , 2019, 10, 240.	3.5	10
28	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
29	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	2.4	9
30	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
31	Transcriptome Profiling Uncovers Potential Common Mechanisms in Fetal Trisomies 18 and 21. <i>OMICS A Journal of Integrative Biology</i> , 2017, 21, 565-570.	2.0	8
32	Prevalence and genetic subtypes of congenital myasthenic syndromes in the pediatric population of Slovenia. <i>European Journal of Paediatric Neurology</i> , 2020, 26, 34-38.	1.6	8
33	Cone Dystrophy Associated with a Novel Variant in the Terminal Codon of the RPCR-ORF15. <i>Genes</i> , 2021, 12, 499.	2.4	8
34	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
35	Diagnostic yield of exome sequencing in myopathies: Experience of a Slovenian tertiary centre. <i>PLoS ONE</i> , 2021, 16, e0252953.	2.5	7
36	Polymorphisms in genes involved in folate metabolism as maternal risk factors for Down syndrome " meta-analysis. <i>Open Medicine (Poland)</i> , 2009, 4, 395-408.	1.3	6

#	ARTICLE	IF	CITATIONS
37	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
38	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
39	Whole-Genome Sequencing in Diagnostics of Selected Slovenian Undiagnosed Patients with Rare Disorders. Life, 2021, 11, 205.	2.4	5
40	Identification of Variants Associated With Rare Hematological Disorder Erythrocytosis Using Targeted Next-Generation Sequencing Analysis. Frontiers in Genetics, 2021, 12, 689868.	2.3	5
41	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
42	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
43	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
44	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
45	Children with cavernous malformations of the central nervous system. European Journal of Paediatric Neurology, 2021, 35, 61-66.	1.6	2
46	Axonal Polyneuropathy in 2 Brothers With a Homozygous Missense Variant in the First Catalytic Domain of <i>PCYT2</i> . Neurology: Genetics, 2022, 8, e658.	1.9	2
47	Biallelic <i>ATOH1</i> Gene Variant in Siblings With Pontocerebellar Hypoplasia, Developmental Delay, and Hearing Loss. Neurology: Genetics, 2022, 8, e677.	1.9	2
48	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
49	Clinical exome sequencing in Serbian patients with movement disorders: Single centre experience. Genetika, 2022, 54, 395-409.	0.4	1
50	Genome Profiling and Potential Biomarkers in Neurodegenerative Disorders. , 2011, , .		0
51	Diagnostic Testing in Epilepsy Genetics Clinical Practice. , 2018, , .		0
52	Genomic Testing for Prenatal Clinical Evaluation of Congenital Anomalies. , 2018, , .		0
53	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
54	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

#	ARTICLE	IF	CITATIONS
55	Title is missing!. , 2020, 15, e0239329.		0
56	Title is missing!. , 2020, 15, e0239329.		0
57	Title is missing!.. , 2020, 15, e0239329.		0
58	Title is missing!.. , 2020, 15, e0239329.		0
59	Front Cover, Volume 43, Issue 8. Human Mutation, 2022, 43, .	2.5	0
60	Human Mutation special issue on "Variant Effect Prediction". Human Mutation, 2022, 43, 973-975.	2.5	0