Ahmad N Abou Tayoun

List of Publications by Year
in descending order

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67 papers	2,764 citations	279798 23 h-index	214800 47 g-index
86 all docs	86 docs citations	86 times ranked	5252 citing authors

#	Article	IF	Citations
1	Recommendations for interpreting the loss of function PVS1 ACMG/AMP variant criterion. Human Mutation, 2018, 39, 1517-1524.	2.5	511
2	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	2.5	312
3	Recommendations for application of the functional evidence PS3/BS3 criterion using the ACMG/AMP sequence variant interpretation framework. Genome Medicine, 2020, 12, 3.	8.2	312
4	Understanding Genotypes and Phenotypes in Epileptic Encephalopathies. Molecular Syndromology, 2016, 7, 172-181.	0.8	97
5	Regulation of Angiotensin- Converting Enzyme 2 in Obesity: Implications for COVID-19. Frontiers in Physiology, 2020, 11, 555039.	2.8	86
6	MicroRNAs as diagnostic markers for pancreatic ductal adenocarcinoma and its precursor, pancreatic intraepithelial neoplasm. Cancer Genetics, 2013, 206, 217-221.	0.4	69
7	Host transcriptomic profiling of COVID-19 patients with mild, moderate, and severe clinical outcomes. Computational and Structural Biotechnology Journal, 2021, 19, 153-160.	4.1	69
8	Automated Clinical Exome Reanalysis Reveals Novel Diagnoses. Journal of Molecular Diagnostics, 2019, 21, 38-48.	2.8	68
9	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
10	Improving hearing loss gene testing: a systematic review of gene evidence toward more efficient next-generation sequencingâ€“based diagnostic testing and interpretation. Genetics in Medicine, 2016, 18, 545-553.	2.4	63
11	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452.	2.4	56
12	Prenatal DNA Sequencing: Clinical, Counseling, and Diagnostic Laboratory Considerations. Prenatal Diagnosis, 2018, 38, 26-32.	2.3	47
13	Ceramide kinase regulates phospholipase C and phosphatidylinositol 4, 5, bisphosphate in phototransduction. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 20063-20068.	7.1	45
14	Use of a Dynamic Genetic Testing Approach for Childhood-Onset Epilepsy. JAMA Network Open, 2019, 2, e192129.	5.9	45
15	A Comprehensive Assay for CFTR Mutational Analysis Using Next-Generation Sequencing. Clinical Chemistry, 2013, 59, 1481-1488.	3.2	44
16	Democratizing Molecular Diagnostics for the Developing World. American Journal of Clinical Pathology, 2014, 141, 17-24.	0.7	42
17	Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. Human Mutation, 2016, 37, 119-126.	2.5	37
18	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35

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19	Diagnosing Cornelia de Lange syndrome and related neurodevelopmental disorders using RNA sequencing. Genetics in Medicine, 2020, 22, 927-936.	2.4	34
20	A singleâ€center SCN8Aâ€related epilepsy cohort: clinical, genetic, and physiologic characterization. Annals of Clinical and Translational Neurology, 2019, 6, 1445-1455.	3.7	32
21	SARS-CoV-2 Whole Genome Amplification and Sequencing for Effective Population-Based Surveillance and Control of Viral Transmission. Clinical Chemistry, 2020, 66, 1450-1458.	3.2	31
22	The Drosophila SK Channel (dSK) Contributes to Photoreceptor Performance by Mediating Sensitivity Control at the First Visual Network. Journal of Neuroscience, 2011, 31, 13897-13910.	3.6	30
23	Sequencing-based diagnostics for pediatric genetic diseases: progress and potential. Expert Review of Molecular Diagnostics, 2016, 16, 987-999.	3.1	29
24	Multiple early introductions of SARS-CoV-2 into a global travel hub in the Middle East. Scientific Reports, 2020, 10, 17720.	3.3	28
25	AUDIOME: a tiered exome sequencingâ€“based comprehensive gene panel for the diagnosis of heterogeneous nonsyndromic sensorineural hearing loss. Genetics in Medicine, 2018, 20, 1600-1608.	2.4	27
26	Genetic variation in the Middle Eastâ€”an opportunity to advance the human genetics field. Genome Medicine, 2020, 12, 116.	8.2	27
27	Curating Clinically Relevant Transcripts for the Interpretation of Sequence Variants. Journal of Molecular Diagnostics, 2018, 20, 789-801.	2.8	25
28	Allele-Specific Droplet Digital PCR Combined with a Next-Generation Sequencing-Based Algorithm for Diagnostic Copy Number Analysis in Genes with High Homology: Proof of Concept Using Stereocilin. Clinical Chemistry, 2018, 64, 705-714.	3.2	24
29	A mutation update for the PCDH19 gene causing early-onset epilepsy in females with an unusual expression pattern. Human Mutation, 2019, 40, 243-257.	2.5	23
30	A multiplex PCR assay for the simultaneous detection of Chlamydia trachomatis, Neisseria gonorrhoeae, and Trichomonas vaginalis. Experimental and Molecular Pathology, 2015, 98, 214-218.	2.1	22
31	From Your Nose to Your Toes: A Review of Severe Acute Respiratory Syndrome Coronavirus 2 Pandemicâ€'Associated Pernio. Journal of Investigative Dermatology, 2021, 141, 2791-2796.	0.7	21
32	PCDH19 -related epilepsy in a male with Klinefelter syndrome: Additional evidence supporting PCDH19 cellular interference disease mechanism. Epilepsy Research, 2018, 145, 89-92.	1.6	20
33	Evaluating the impact of in silico predictors on clinical variant classification. Genetics in Medicine, 2022, 24, 924-930.	2.4	20
34	Genetic variant pathogenicity prediction trained using disease-specific clinical sequencing data sets. Genome Research, 2019, 29, 1144-1151.	5.5	19
35	Recurrent variants in OTOF are significant contributors to prelingual nonsydromic hearing loss in Saudi patients. Genetics in Medicine, 2018, 20, 536-544.	2.4	18
36	Considerations for whole exome sequencing unique to prenatal care. Human Genetics, 2020, 139, 1149-1159.	3.8	18

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37	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
38	Clinical evaluation and etiologic diagnosis of hearing loss: A clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2022, 24, 1392-1406.	2.4	18
39	Rapid and accurate interpretation of clinical exomes using Phenoxome: a computational phenotype-driven approach. European Journal of Human Genetics, 2019, 27, 612-620.	2.8	17
40	The Development and Validation of Clinical Exome-Based Panels Using ExomeSlicer. Journal of Molecular Diagnostics, 2018, 20, 643-652.	2.8	16
41	Development of a rapid clinical TPMT genotyping assay. Clinical Biochemistry, 2014, 47, 126-129.	1.9	15
42	Using large sequencing data sets to refine intragenic disease regions and prioritize clinical variant interpretation. Genetics in Medicine, 2017, 19, 496-504.	2.4	15
43	Utility of clinical exome sequencing in a complex Emirati pediatric cohort. Computational and Structural Biotechnology Journal, 2020, 18, 1020-1027.	4.1	14
44	Single-cell transcriptome identifies FCGR3B upregulated subtype of alveolar macrophages in patients with critical COVID-19. IScience, 2021, 24, 103030.	4.1	13
45	Genetic and Clinical Characteristics of Patients in the Middle East With Multisystem Inflammatory Syndrome in Children. JAMA Network Open, 2022, 5, e2214985.	5.9	13
46	Need for Automated Interactive Genomic Interpretation and Ongoing Reanalysis. JAMA Pediatrics, 2018, 172, 1113.	6.2	12
47	The genomic landscape of pediatric rheumatology disorders in the Middle East. Human Mutation, 2021, 42, e1-e14.	2.5	12
48	Roles of the Drosophila SK Channel (dSK) in Courtship Memory. PLoS ONE, 2012, 7, e34665.	2.5	12
49	Genomic medicine in the Middle East. Genome Medicine, 2021, 13, 184.	8.2	12
50	Association of Dromedary Camels and Camel Ticks with Reassortant Crimean-Congo Hemorrhagic Fever Virus, United Arab Emirates. Emerging Infectious Diseases, 2021, 27, 2471-2474.	4.3	11
51	VIPâ€HL: Semiâ€automated ACMG/AMP variant interpretation platform for genetic hearing loss. Human Mutation, 2021, 42, 1567-1575.	2.5	10
52	Utility of droplet digital PCR and NGS-based CNV clinical assays in hearing loss diagnostics: current status and future prospects. Expert Review of Molecular Diagnostics, 2021, 21, 213-221.	3.1	8
53	Genotypeâ€phenotype correlation identified a novel SARSâ€CoVâ€2 variant possibly linked to severe disease. Transboundary and Emerging Diseases, 2021, , .	3.0	8
54	A clinical PCR fragment analysis assay for TA repeat sizing in the UGT1A1 promoter region. Clinica Chimica Acta, 2013, 422, 1-4.	1.1	7

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55	Ca²⁺-Activated K⁺ Channels Reduce Network Excitability, Improving Adaptability and Energetics for Transmitting and Perceiving Sensory Information. Journal of Neuroscience, 2019, 39, 7132-7154.	3.6	7
56	Using Machine Learning to Identify True Somatic Variants from Next-Generation Sequencing. Clinical Chemistry, 2020, 66, 239-246.	3.2	7
57	MERSâ€CoV in sheep, goats, and cattle, United Arab Emirates, 2019: Virological and serological investigations reveal an accidental spillover from dromedaries. Transboundary and Emerging Diseases, 2022, 69, 3066-3072.	3.0	7
58	Middle Eastern Genetic Variation Improves Clinical Annotation of the Human Genome. Journal of Personalized Medicine, 2022, 12, 423.	2.5	7
59	Rapid whole genome sequencing of critically ill pediatric patients from genetically underrepresented populations. Genome Medicine, 2022, 14, .	8.2	7
60	A rapid RT-PCR assay for the detection of HIV-1 in human plasma specimens. Experimental and Molecular Pathology, 2014, 97, 111-115.	2.1	5
61	Case Report: CMV-Associated Congenital Nephrotic Syndrome. Frontiers in Pediatrics, 2020, 8, 580178.	1.9	5
62	Evaluating the thermostability of commercial fast real-time PCR master mixes. Experimental and Molecular Pathology, 2012, 93, 261-263.	2.1	4
63	Coexistence of a novel WISP3 pathogenic variant and an MEFV mutation in an Arabic family with progressive pseudorheumatoid dysplasia mimicking polyarticular juvenile idiopathic arthritis. Pediatric Rheumatology, 2020, 18, 69.	2.1	4
64	A Rare Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) Mutation Associated With Typical Cystic Fibrosis in an Arab Child. Cureus, 2021, 13, e13526.	0.5	2
65	A Simple Practical Guide to Genomic Diagnostics in a Pediatric Setting. Genes, 2021, 12, 818.	2.4	2
66	Detection of copy number variants and genes by chromosomal microarray in an Emirati neurodevelopmental disorders cohort. Neurogenetics, 2022, 23, 137-149.	1.4	0
67	Comprehensive Genomic Sequencingâ€"Based Screening for Hearing Loss in the Neonatal Intensive Care Settingâ€"Is It Time?. JAMA Network Open, 2022, 5, e2220992.	5.9	0