Khalid A Fakhro

List of Publications by Year
in descending order

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42 papers	1,369 citations	471371 17 h-index	377752 34 g-index
44 all docs	44 docs citations	44 times ranked	2414 citing authors

#	Article	IF	Citations
1	Rare copy number variations in congenital heart disease patients identify unique genes in left-right patterning. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 2915-2920.	3.3	226
2	Point-of-care whole-exome sequencing of idiopathic male infertility. Genetics in Medicine, 2018, 20, 1365-1373.	1.1	105
3	Role of NAD+ in regulating cellular and metabolic signaling pathways. Molecular Metabolism, 2021, 49, 101195.	3.0	104
4	The Qatar genome: a population-specific tool for precision medicine in the Middle East. Human Genome Variation, 2016, 3, 16016.	0.4	103
5	GA4GH: International policies and standards for data sharing across genomic research and healthcare. Cell Genomics, 2021, 1, 100029.	3.0	94
6	Indigenous Arabs are descendants of the earliest split from ancient Eurasian populations. Genome Research, 2016, 26, 151-162.	2.4	89
7	Whole-exome sequencing identifies common and rare variant metabolic QTLs in a Middle Eastern population. Nature Communications, 2018, 9, 333.	5.8	63
8	Genetics of structural and functional brain changes in autism spectrum disorder. Translational Psychiatry, 2020, 10, 229.	2.4	63
9	Exome Sequencing Identifies Potential Risk Variants for Mendelian Disorders at High Prevalence in Qatar. Human Mutation, 2014, 35, 105-116.	1.1	43
10	Diagnosis and treatment of type 1 diabetes at the dawn of the personalized medicine era. Journal of Translational Medicine, 2021, 19, 137.	1.8	41
11	Genetic variations influence brain changes in patients with attention-deficit hyperactivity disorder. Translational Psychiatry, 2021, 11, 349.	2.4	37
12	A systematic review on the genetics of male infertility in the era of next-generation sequencing. Arab Journal of Urology Arab Association of Urology, 2018, 16, 53-64.	0.7	36
13	Epigenetics and Cardiovascular Disease in Diabetes. Current Diabetes Reports, 2015, 15, 108.	1.7	32
14	Identification of mutation resistance coldspots for targeting the SARSâ€CoV2 main protease. IUBMB Life, 2021, 73, 670-675.	1.5	30
15	Type 2 Diabetes Risk Allele Loci in the Qatari Population. PLoS ONE, 2016, 11, e0156834.	1.1	30
16	Metabolic and Metabo-Clinical Signatures of Type 2 Diabetes, Obesity, Retinopathy, and Dyslipidemia. Diabetes, 2022, 71, 184-205.	0.3	29
17	Qatar genome: Insights on genomics from the Middle East. Human Mutation, 2022, 43, 499-510.	1.1	29
18	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	3.7	20

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19	A population study of clinically actionable genetic variation affecting drug response from the Middle East. Npj Genomic Medicine, 2022, 7, 10.	1.7	20
20	Thousands of Qatari genomes inform human migration history and improve imputation of Arab haplotypes. Nature Communications, 2021, 12, 5929.	5.8	18
21	Two hits in one: whole genome sequencing unveils LIG4 syndrome and urofacial syndrome in a case report of a child with complex phenotype. BMC Medical Genetics, 2016, 17, 84.	2.1	17
22	Point of Care Exome Sequencing Reveals Allelic and Phenotypic Heterogeneity Underlying Mendelian disease in Qatar. Human Molecular Genetics, 2019, 28, 3970-3981.	1.4	16
23	PGAP3 Associated with Hyperphosphatasia with Mental Retardation Plays a Novel Role in Brain Morphogenesis and Neuronal Wiring at Early Development. Cells, 2020, 9, 1782.	1.8	14
24	Actionable genomic variants in 6045 participants from the Qatar Genome Program. Human Mutation, 2021, 42, 1584-1601.	1.1	13
25	Willingness to participate in genome testing: a survey of public attitudes from Qatar. Journal of Human Genetics, 2020, 65, 1067-1073.	1.1	12
26	Single-Nucleotide Variations of the Human Nuclear Hormone Receptor Genes in 60,000 Individuals. Journal of the Endocrine Society, 2018, 2, 77-90.	0.1	11
27	Copy number variations in the genome of the Qatari population. BMC Genomics, 2015, 16, 834.	1.2	9
28	Hypertrophic cardiomyopathy-linked variants of cardiac myosin-binding protein C3 display altered molecular properties and actin interaction. Biochemical Journal, 2018, 475, 3933-3948.	1.7	8
29	Ethnic-specific association of amylase gene copy number with adiposity traits in a large Middle Eastern biobank. Npj Genomic Medicine, 2021, 6, 8.	1.7	8
30	Brain microstructural changes support cognitive deficits in HIV uninfected children born to HIV infected mothers. Brain, Behavior, & Immunity - Health, 2020, 2, 100039.	1.3	7
31	Genomics of Autism. Advances in Neurobiology, 2020, 24, 83-96.	1.3	7
32	Reading between the (Genetic) Lines: How Epigenetics is Unlocking Novel Therapies for Type 1 Diabetes. Cells, 2020, 9, 2403.	1.8	6
33	Functional characterization of human myosinâ€binding protein C3 variants associated with hypertrophic cardiomyopathy reveals exonâ€specific cardiac phenotypes in zebrafish model. Journal of Cellular Physiology, 2020, 235, 7870-7888.	2.0	6
34	A map of copy number variations in the Tunisian population: a valuable tool for medical genomics in North Africa. Npj Genomic Medicine, 2021, 6, 3.	1.7	5
35	The QChip1 knowledgebase and microarray for precision medicine in Qatar. Npj Genomic Medicine, 2022, 7, 3.	1.7	4
36	Homozygous duplication identified by whole genome sequencing causes LRBA deficiency. Npj Genomic Medicine, 2021, 6, 96.	1.7	3

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37	Clinical, Genetic and Functional Characterization of a Novel AVPR2 Missense Mutation in a Woman with X-Linked Recessive Nephrogenic Diabetes Insipidus. Journal of Personalized Medicine, 2022, 12, 118.	1.1	2
38	Analysis of incidental findings in Qatar genome participants reveals novel functional variants in LMNA and DSP. Human Molecular Genetics, 2022, , .	1.4	2
39	A recessive variant in SIM2 in a child with complex craniofacial anomalies and global developmental delay. European Journal of Medical Genetics, 2022, 65, 104455.	0.7	2
40	Patterns and distribution of de novo mutations in multiplex Middle Eastern families. Journal of Human Genetics, 2022, 67, 579-588.	1.1	2
41	A de novo start-loss in EFTUD2 associated with mandibulofacial dysostosis with microcephaly: case report. Journal of Physical Education and Sports Management, 2022, 8, a006206.	0.5	2
42	Severe loss-of-function mutations affect critical genes and pathways in the Qatari population. , 2013, , .		0