

Bassam R Ali

List of Publications by Year in descending order

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144
papers

3,916
citations

172457

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161849

54
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149
all docs

149
docs citations

149
times ranked

6525
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	6.2	352
2	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	28.9	253
3	Mutations in DDHD2, Encoding an Intracellular Phospholipase A1, Cause a Recessive Form of Complex Hereditary Spastic Paraplegia. American Journal of Human Genetics, 2012, 91, 1073-1081.	6.2	159
4	Membrane Targeting of Rab GTPases Is Influenced by the Prenylation Motif. Molecular Biology of the Cell, 2003, 14, 1882-1899.	2.1	137
5	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	6.2	108
6	DNA methylation and repressive H3K9 and H3K27 trimethylation in the promoter regions of PD-1, CTLA-4, TIM-3, LAG-3, TIGIT, and PD-L1 genes in human primary breast cancer. Clinical Epigenetics, 2018, 10, 78.	4.1	103
7	Multiple regions contribute to membrane targeting of Rab GTPases. Journal of Cell Science, 2004, 117, 6401-6412.	2.0	100
8	Rab GTPases Containing a CAAX Motif Are Processed Post-geranylgeranylation by Proteolysis and Methylation. Journal of Biological Chemistry, 2007, 282, 1487-1497.	3.4	97
9	Preferential accumulation of regulatory T cells with highly immunosuppressive characteristics in breast tumor microenvironment. Oncotarget, 2017, 8, 33159-33171.	1.8	96
10	Dual inhibition of STAT1 and STAT3 activation downregulates expression of PD-L1 in human breast cancer cells. Expert Opinion on Therapeutic Targets, 2018, 22, 547-557.	3.4	90
11	A Homozygous Mutation in the Tight-Junction Protein JAM3 Causes Hemorrhagic Destruction of the Brain, Subependymal Calcification, and Congenital Cataracts. American Journal of Human Genetics, 2010, 87, 882-889.	6.2	87
12	Trafficking defects and loss of ligand binding are the underlying causes of all reported DDR2 missense mutations found in SMED-SL patients. Human Molecular Genetics, 2010, 19, 2239-2250.	2.9	77
13	Targeting of Rab GTPases to cellular membranes. Biochemical Society Transactions, 2005, 33, 652-656.	3.4	74
14	Intratumoral FoxP3+Helios+ Regulatory T Cells Upregulating Immunosuppressive Molecules Are Expanded in Human Colorectal Cancer. Frontiers in Immunology, 2017, 8, 619.	4.8	69
15	Mutations of a country: a mutation review of single gene disorders in the United Arab Emirates (UAE). Human Mutation, 2010, 31, 505-520.	2.5	67
16	ER-associated protein degradation is a common mechanism underpinning numerous monogenic diseases including Robinow syndrome. Human Molecular Genetics, 2005, 14, 2559-2569.	2.9	61
17	Pharmaceutical Chaperones and Proteostasis Regulators in the Therapy of Lysosomal Storage Disorders: Current Perspective and Future Promises. Frontiers in Pharmacology, 2017, 8, 448.	3.5	51
18	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	2.9	47

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19	Prevalence and Novel Mutations of Lysosomal Storage Disorders in United Arab Emirates. <i>JIMD Reports</i> , 2012, 10, 1-9.	1.5	44
20	Similar processes mediate glycopeptide export from the endoplasmic reticulum in mammalian cells and <i>Saccharomyces cerevisiae</i> . <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1997, 94, 6730-6734.	7.1	42
21	Success stories in genomic medicine from resource-limited countries. <i>Human Genomics</i> , 2015, 9, 11.	2.9	41
22	Characterization of the subunits in an apparently homogeneous subpopulation of <i>Clostridium thermocellum</i> cellulosomes. <i>Enzyme and Microbial Technology</i> , 1995, 17, 705-711.	3.2	40
23	A Novel Whole Exon Deletion in <i>WWOX</i> Gene Causes Early Epilepsy, Intellectual Disability and Optic Atrophy. <i>Journal of Molecular Neuroscience</i> , 2015, 56, 17-23.	2.3	40
24	A Novel Single-Nucleotide Deletion (c.1020delA) in <i>NSUN2</i> Causes Intellectual Disability in an Emirati Child. <i>Journal of Molecular Neuroscience</i> , 2015, 57, 393-399.	2.3	39
25	Novel Robinow syndrome causing mutations in the proximal region of the frizzled-like domain of <i>ROR2</i> are retained in the endoplasmic reticulum. <i>Human Genetics</i> , 2007, 122, 389-395.	3.8	38
26	Asparagine synthetase deficiency detected by whole exome sequencing causes congenital microcephaly, epileptic encephalopathy and psychomotor delay. <i>Metabolic Brain Disease</i> , 2015, 30, 687-694.	2.9	38
27	Defective cellular trafficking of missense <i>NPR-B</i> mutants is the major mechanism underlying acromesomelic dysplasia-type Maroteaux. <i>Human Molecular Genetics</i> , 2008, 18, 267-277.	2.9	36
28	Haploinsufficiency of the E3 ubiquitin-protein ligase gene <i>TRIP12</i> causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. <i>Human Genetics</i> , 2017, 136, 377-386.	3.8	36
29	Endoplasmic Reticulum Quality Control Is Involved in the Mechanism of Endoglin-Mediated Hereditary Haemorrhagic Telangiectasia. <i>PLoS ONE</i> , 2011, 6, e26206.	2.5	32
30	A missense founder mutation in <i>VLDLR</i> is associated with Dysequilibrium Syndrome without quadrupedal locomotion. <i>BMC Medical Genetics</i> , 2012, 13, 80.	2.1	31
31	Mutation spectrum of Joubert syndrome and related disorders among Arabs. <i>Human Genome Variation</i> , 2014, 1, 14020.	0.7	31
32	Delineation of the Clinical, Molecular and Cellular Aspects of Novel <i>JAM3</i> Mutations Underlying the Autosomal Recessive Hemorrhagic Destruction of the Brain, Subependymal Calcification, and Congenital Cataracts. <i>Human Mutation</i> , 2013, 34, 498-505.	2.5	30
33	A novel mutation in <i>DDR2</i> causing spondylo-meta-epiphyseal dysplasia with short limbs and abnormal calcifications (SMED-SL) results in defective intra-cellular trafficking. <i>BMC Medical Genetics</i> , 2014, 15, 42.	2.1	30
34	Toxicity and Pharmacogenomic Biomarkers in Breast Cancer Chemotherapy. <i>Frontiers in Pharmacology</i> , 2020, 11, 445.	3.5	30
35	Synergistic interaction of the cellulosome integrating protein (CipA) from <i>Clostridium thermocellum</i> with a cellulosomal endoglucanase. <i>FEBS Letters</i> , 1998, 422, 221-224.	2.8	29
36	A novel statin-mediated "prenylation block-and-release" assay provides insight into the membrane targeting mechanisms of small GTPases. <i>Biochemical and Biophysical Research Communications</i> , 2010, 397, 34-41.	2.1	29

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37	A mutation in KIF7 is responsible for the autosomal recessive syndrome of macrocephaly, multiple epiphyseal dysplasia and distinctive facial appearance. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 27.	2.7	29
38	Genomic Medicine Without Borders: Which Strategies Should Developing Countries Employ to Invest in Precision Medicine? A New "Fast-Second Winner" Strategy. <i>OMICS A Journal of Integrative Biology</i> , 2017, 21, 647-657.	2.0	29
39	Current opinion on the pharmacogenomics of paclitaxel-induced toxicity. <i>Expert Opinion on Drug Metabolism and Toxicology</i> , 2021, 17, 785-801.	3.3	29
40	ACE2 Nascence, trafficking, and SARS-CoV-2 pathogenesis: the saga continues. <i>Human Genomics</i> , 2021, 15, 8.	2.9	28
41	The farnesyltransferase inhibitor manumycin A is a novel trypanocide with a complex mode of action including major effects on mitochondria. <i>Molecular and Biochemical Parasitology</i> , 1999, 104, 67-80.	1.1	27
42	Is cystic fibrosis-related diabetes an apoptotic consequence of ER stress in pancreatic cells?. <i>Medical Hypotheses</i> , 2009, 72, 55-57.	1.5	27
43	Identification of New Alleles and the Determination of Alleles and Genotypes Frequencies at the CYP2D6 Gene in Emiratis. <i>PLoS ONE</i> , 2011, 6, e28943.	2.5	27
44	Is autosomal recessive Silver-Russel syndrome a separate entity or is it part of the 3M syndrome spectrum?. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1236-1245.	1.2	25
45	Pathological Crosstalk Between Oxidized LDL and ER Stress in Human Diseases: A Comprehensive Review. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 674103.	3.7	24
46	A novel de novo mutation in DYNC1H1 gene underlying malformation of cortical development and cataract. <i>Meta Gene</i> , 2016, 9, 124-127.	0.6	23
47	West syndrome, developmental and epileptic encephalopathy, and severe CNS disorder associated with <i>WWOX</i> mutations. <i>Epileptic Disorders</i> , 2018, 20, 401-412.	1.3	23
48	A null variant in <i>PUS3</i> confirms its involvement in intellectual disability and further delineates the associated neurodevelopmental disease. <i>Clinical Genetics</i> , 2018, 94, 586-587.	2.0	23
49	Defective cellular trafficking of the bone morphogenetic protein receptor type II by mutations underlying familial pulmonary arterial hypertension. <i>Gene</i> , 2015, 561, 148-156.	2.2	22
50	LINS, a modulator of the WNT signaling pathway, is involved in human cognition. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 87.	2.7	21
51	A novel aberrant splice site mutation in <i>COL27A1</i> is responsible for Steel syndrome and extension of the phenotype to include hearing loss. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1257-1263.	1.2	21
52	Genomic variants in the FTO gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients. <i>Human Genomics</i> , 2017, 11, 30.	2.9	21
53	Long-Term Effects of Pediatric Acute Lymphoblastic Leukemia Chemotherapy: Can Recent Findings Inform Old Strategies?. <i>Frontiers in Oncology</i> , 2021, 11, 710163.	2.8	21
54	Clinical and molecular analysis of UAE fibrochondrogenesis patients expands the phenotype and reveals two <i>COL11A1</i> homozygous null mutations. <i>Clinical Genetics</i> , 2012, 82, 147-156.	2.0	20

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55	StÃ¼veâ€“Wiedemann syndrome and related bent bone dysplasias. <i>Clinical Genetics</i> , 2012, 82, 12-21.	2.0	20
56	Identification of the Cellular Mechanisms That Modulate Trafficking of Frizzled Family Receptor 4 (FZD4) Missense Mutants Associated With Familial Exudative Vitreoretinopathy. , 2014, 55, 3423.		20
57	Endoplasmic reticulum quality control of LDLR variants associated with familial hypercholesterolemia. <i>FEBS Open Bio</i> , 2019, 9, 1994-2005.	2.3	20
58	Prevalence of pharmacogenomic variants in 100 pharmacogenes among Southeast Asian populations under the collaboration of the Southeast Asian Pharmacogenomics Research Network (SEAPharm). <i>Human Genome Variation</i> , 2021, 8, 7.	0.7	19
59	Retention in the endoplasmic reticulum is the underlying mechanism of some hereditary haemorrhagic telangiectasia type 2 ALK1 missense mutations. <i>Molecular and Cellular Biochemistry</i> , 2013, 373, 247-257.	3.1	18
60	A homozygous splicing mutation in ELAC2 suggests phenotypic variability including intellectual disability with minimal cardiac involvement. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 139.	2.7	18
61	Cost-effectiveness analysis of pharmacogenomics-guided clopidogrel treatment in Spanish patients undergoing percutaneous coronary intervention. <i>Pharmacogenomics Journal</i> , 2019, 19, 438-445.	2.0	18
62	Glutathione S-transferase from oxadiazon treated chickpea. <i>Phytochemistry</i> , 1990, 29, 2431-2435.	2.9	17
63	Clinical and molecular analysis of isovaleric acidemia patients in the United Arab Emirates reveals remarkable phenotypes and four novel mutations in the IVD gene. <i>European Journal of Medical Genetics</i> , 2012, 55, 671-676.	1.3	17
64	A novel mutation in <i>PRG4</i> gene underlying camptodactylyâ€“arthropathyâ€“coxa varaâ€“pericarditis syndrome with the possible expansion of the phenotype to include congenital cataract. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2012, 94, 553-556.	1.6	17
65	A de novo mutation in the X-linked PAK3 gene is the underlying cause of intellectual disability and macrocephaly in monozygotic twins. <i>European Journal of Medical Genetics</i> , 2017, 60, 212-216.	1.3	17
66	Genomics and Pharmacogenomics Knowledge, Attitude and Practice of Pharmacists Working in United Arab Emirates: Findings from Focus Group Discussionsâ€“A Qualitative Study. <i>Journal of Personalized Medicine</i> , 2020, 10, 134.	2.5	17
67	Knowledge, Attitudes, and Perceived Barriers toward Genetic Testing and Pharmacogenomics among Healthcare Workers in the United Arab Emirates: A Cross-Sectional Study. <i>Journal of Personalized Medicine</i> , 2020, 10, 216.	2.5	17
68	Clinical and Molecular Analysis of a Novel COLQ Missense Mutation Causing Congenital Myasthenic Syndrome in a Syrian Family. <i>Pediatric Neurology</i> , 2014, 51, 165-169.	2.1	16
69	Clinical and molecular delineation of dysequilibrium syndrome type 2 and profound sensorineural hearing loss in an inbred Arab family. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 540-543.	1.2	16
70	Gonadal mosaicism in <i>ARID1B</i> gene causes intellectual disability and dysmorphic features in three siblings. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 156-161.	1.2	16
71	Genetic polymorphisms of cytochrome P450-1A2 (CYP1A2) among Emiratis. <i>PLoS ONE</i> , 2017, 12, e0183424.	2.5	16
72	Knowledge and Attitudes of Medical and Health Science Students in the United Arab Emirates toward Genomic Medicine and Pharmacogenomics: A Cross-Sectional Study. <i>Journal of Personalized Medicine</i> , 2020, 10, 191.	2.5	16

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73	R58fs Mutation in the <i>HGD</i> Gene in a Family with Alkaptonuria in the UAE. <i>Annals of Human Genetics</i> , 2009, 73, 125-130.	0.8	15
74	Molecular and clinical analysis of Ellis-van Creveld syndrome in the United Arab Emirates. <i>BMC Medical Genetics</i> , 2010, 11, 33.	2.1	15
75	Variation in 100 relevant pharmacogenes among emiratis with insights from understudied populations. <i>Scientific Reports</i> , 2020, 10, 21310.	3.3	14
76	Pharmacogenomics in pediatric acute lymphoblastic leukemia: promises and limitations. <i>Pharmacogenomics</i> , 2017, 18, 687-699.	1.3	13
77	A Novel SLC1A4 Mutation (p.Y191*) Causes Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly (SPATCCM) With Seizure Disorder. <i>Child Neurology Open</i> , 2019, 6, 2329048X1988064.	1.1	13
78	Correlation of <i>SIN3A</i> genomic variants with β^2 -hemoglobinopathies disease severity and hydroxyurea treatment efficacy. <i>Pharmacogenomics</i> , 2016, 17, 1785-1793.	1.3	12
79	Endoplasmic reticulum retention of xylosyltransferase 1 (XYLT1) mutants underlying Desbuquois dysplasia type II. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1773-1781.	1.2	12
80	A <i>B3GALT6</i> variant in patient originally described as Al-Gazali syndrome and implicating the endoplasmic reticulum quality control in the mechanism of some <i>B3GALT6</i> opathy mutations. <i>Clinical Genetics</i> , 2018, 93, 1148-1158.	2.0	12
81	Compound heterozygous variants in the multiple PDZ domain protein (MPDZ) cause a case of mild non-progressive communicating hydrocephalus. <i>BMC Medical Genetics</i> , 2018, 19, 34.	2.1	12
82	Impaired trafficking of the very low density lipoprotein receptor caused by missense mutations associated with dysequilibrium syndrome. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2014, 1843, 2871-2877.	4.1	11
83	A recessive syndrome of intellectual disability, moderate overgrowth, and renal dysplasia predisposing to Wilms tumor is caused by a mutation in <i>FIBP</i> gene. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2111-2118.	1.2	11
84	FGF23 ^{S129F} mutant bypasses ER/Golgi to the circulation of hyperphosphatemic familial tumoral calcinosis patients. <i>Bone</i> , 2016, 93, 187-195.	2.9	11
85	Whole transcriptome analysis of human erythropoietic cells during ontogenesis suggests a role of VEGFA gene as modulator of fetal hemoglobin and pharmacogenomic biomarker of treatment response to hydroxyurea in β^2 -type hemoglobinopathy patients. <i>Human Genomics</i> , 2017, 11, 24.	2.9	11
86	Development of the pharmacogenomics and genomics literacy framework for pharmacists. <i>Human Genomics</i> , 2021, 15, 62.	2.9	11
87	Analysis of two Arab families reveals additional support for a DFNB2 nonsyndromic phenotype of MYO7A. <i>Molecular Biology Reports</i> , 2014, 41, 193-200.	2.3	10
88	Genomic variants in the <i>ASS1</i> gene, involved in the nitric oxide biosynthesis and signaling pathway, predict hydroxyurea treatment efficacy in compound sickle cell disease/ β^2 -thalassemia patients. <i>Pharmacogenomics</i> , 2016, 17, 393-403.	1.3	10
89	Degradation routes of trafficking-defective VLDLR mutants associated with Dysequilibrium syndrome. <i>Scientific Reports</i> , 2018, 8, 1583.	3.3	10
90	A recessive truncating variant in thrombospondin domain containing protein 1 gene <i>THSD1</i> is the underlying cause of nonimmune hydrops fetalis, congenital cardiac defects, and haemangiomas in four patients from a consanguineous family. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1996-2003.	1.2	10

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91	Evaluating the Role of MAST1 as an Intellectual Disability Disease Gene: Identification of a Novel De Novo Variant in a Patient with Developmental Disabilities. <i>Journal of Molecular Neuroscience</i> , 2020, 70, 320-327.	2.3	10
92	Endoplasmic Reticulum Associated Protein Degradation (ERAD) in the Pathology of Diseases Related to TGF β Signaling Pathway: Future Therapeutic Perspectives. <i>Frontiers in Molecular Biosciences</i> , 2020, 7, 575608.	3.5	10
93	Mendelian randomization in pharmacogenomics: The unforeseen potentials. <i>Biomedicine and Pharmacotherapy</i> , 2022, 150, 112952.	5.6	10
94	A progeroid syndrome with neonatal presentation and long survival maps to 19p13.3p13.2. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2013, 97, 456-462.	1.6	9
95	Novel genetic risk variants for pediatric celiac disease. <i>Human Genomics</i> , 2016, 10, 34.	2.9	9
96	Defect in phosphoinositide signalling through a homozygous variant in <i>PLCB3</i> causes a new form of spondylometaphyseal dysplasia with corneal dystrophy. <i>Journal of Medical Genetics</i> , 2018, 55, 122-130.	3.2	9
97	A novel mutation in ARG1 gene is responsible for arginase deficiency in an Asian family. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2009, 30, 1601-3.	1.1	9
98	Export of a misprocessed GPI-anchored protein from the endoplasmic reticulum in vitro in an ATP- and cytosol-dependent manner. <i>FEBS Letters</i> , 2000, 483, 32-36.	2.8	8
99	Novel mutations in ADAMTSL2 gene underlying geleophysic dysplasia in families from United Arab Emirates. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2013, 97, 764-769.	1.6	8
100	The mutational spectrum of the NF1 gene in neurofibromatosis type I patients from UAE. <i>Child's Nervous System</i> , 2014, 30, 1183-1189.	1.1	8
101	New Arab family with cerebral dysgenesis, neuropathy, ichthyosis and keratoderma syndrome suggests a possible founder effect for the c.223delG mutation. <i>Journal of Dermatology</i> , 2015, 42, 821-822.	1.2	8
102	Association of variants in <i>PTPN22</i> , <i>CTLA4</i> , <i>IL2RA</i> , and <i>INS</i> genes with type 1 diabetes in Emiratis. <i>Annals of Human Genetics</i> , 2021, 85, 48-57.	0.8	8
103	Stakeholders' Interest and Attitudes toward Genomic Medicine and Pharmacogenomics Implementation in the United Arab Emirates: A Qualitative Study. <i>Public Health Genomics</i> , 2021, 24, 99-109.	1.0	8
104	New and known mutations associated with inborn errors of metabolism in a heterogeneous Middle Eastern population. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2011, 32, 353-9.	1.1	8
105	Phenotypic and mutational spectrum of <i>ROR2</i> -related Robinow syndrome. <i>Human Mutation</i> , 2022, 43, 900-918.	2.5	8
106	A Microsomal GTPase Is Required for Glycopeptide Export from the Mammalian Endoplasmic Reticulum. <i>Journal of Biological Chemistry</i> , 2000, 275, 33222-33230.	3.4	7
107	Identification of Mutations Underlying 20 Inborn Errors of Metabolism in the United Arab Emirates Population. <i>Genetic Testing and Molecular Biomarkers</i> , 2012, 16, 366-371.	0.7	7
108	Determination of the CCR5 Δ 32 frequency in Emiratis and Tunisians and the screening of the CCR5 gene for novel alleles in Emiratis. <i>Gene</i> , 2013, 529, 113-118.	2.2	7

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109	Role of Genomic Biomarkers in Increasing Fetal Hemoglobin Levels Upon Hydroxyurea Therapy and in β^2 -Thalassemia Intermedia: A Validation Cohort Study. <i>Hemoglobin</i> , 2019, 43, 27-33.	0.8	7
110	Synthesis of 3-arsonoalanine and its action on aspartate aminotransferase and aspartate ammonia-lyase. Comparison with arsenical analogues of malate and fumarate. <i>FEBS Journal</i> , 1993, 215, 161-166.	0.2	6
111	Improved plasma membrane expression of the trafficking defective P344R mutant of muscle, skeletal, receptor tyrosine kinase (MuSK) causing congenital myasthenic syndrome. <i>International Journal of Biochemistry and Cell Biology</i> , 2015, 60, 119-129.	2.8	6
112	Key Pharmacogenomic Considerations for Sickle Cell Disease Patients. <i>OMICS A Journal of Integrative Biology</i> , 2017, 21, 314-322.	2.0	6
113	Studies on N-Acetyltransferase (NAT2) Genotype Relationships in Emiratis: Confirmation of the Existence of Phenotype Variation among Slow Acetylators. <i>Annals of Human Genetics</i> , 2017, 81, 190-196.	0.8	6
114	<i>VKORC1</i> variants as significant predictors of warfarin dose in Emiratis.	0.7	6
115	Further Delineation of the Microcephaly-Micromelia Syndrome Associated with Loss-of-Function Variants in DONSON. <i>Molecular Syndromology</i> , 2019, 10, 171-176.	0.8	6
116	Proteostasis Regulation in the Endoplasmic Reticulum: An Emerging Theme in the Molecular Pathology and Therapeutic Management of Familial Hypercholesterolemia. <i>Frontiers in Genetics</i> , 2020, 11, 570355.	2.3	6
117	The pharmacological chaperone N-n-butyl-deoxygalactonojirimycin enhances β -galactosidase processing and activity in fibroblasts of a patient with infantile GM1-gangliosidosis. <i>Human Genetics</i> , 2020, 139, 657-673.	3.8	6
118	Clinical implementation of drug metabolizing gene-based therapeutic interventions worldwide. <i>Human Genetics</i> , 2022, 141, 1137-1157.	3.8	6
119	Role of Ceramides in the Molecular Pathogenesis and Potential Therapeutic Strategies of Cardiometabolic Diseases: What we Know so Far. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 816301.	3.7	6
120	Anterior segment anomalies of the eye, growth retardation associated with hypoplastic pituitary gland and endocrine abnormalities: Jung syndrome or a new syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 251-256.	1.2	5
121	Novel KCNQ2 Mutation in a Large Emirati Family With Benign Familial Neonatal Seizures. <i>Pediatric Neurology</i> , 2013, 48, 63-66.	2.1	5
122	Secondary association of PDLIM5 with paranoid schizophrenia in Emirati patients. <i>Meta Gene</i> , 2015, 5, 135-139.	0.6	5
123	The induction of chickpea glutathione S-transferase by oxadiazon. <i>Phytochemistry</i> , 1991, 30, 2131-2134.	2.9	4
124	Computer-aided approaches reveal trihydroxychroman and pyrazolone derivatives as potential inhibitors of SARS-CoV-2 virus main protease. <i>Acta Pharmaceutica</i> , 2021, 71, 325-333.	2.0	4
125	<i>Bi</i> allelic null variant in matrix metalloproteinase-15, causes congenital cardiac defect, cholestasis jaundice, and failure to thrive. <i>Clinical Genetics</i> , 2022, 101, 403-410.	2.0	4
126	Endoglin Wild Type and Variants Associated With Hereditary Hemorrhagic Telangiectasia Type 1 Undergo Distinct Cellular Degradation Pathways. <i>Frontiers in Molecular Biosciences</i> , 2022, 9, 828199.	3.5	4

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127	HLA-DRB1 and â€œDQB1 Alleles, Haplotypes and Genotypes in Emirati Patients with Type 1 Diabetes Underscores the Benefits of Evaluating Understudied Populations. <i>Frontiers in Genetics</i> , 2022, 13, 841879.	2.3	4
128	Congenital Teratocarcinoma With <i>CTNNB1</i> Gene Mutation Presenting as an Ocular Mass. <i>Pediatric and Developmental Pathology</i> , 2022, 25, 562-567.	1.0	4
129	The possible threat of Zika virus in the Middle East. <i>Travel Medicine and Infectious Disease</i> , 2016, 14, 277-278.	3.0	3
130	Genomic variants in members of the KrÃ¼ppel-like factor gene family are associated with disease severity and hydroxyurea treatment efficacy in Î²-hemoglobinopathies patients. <i>Pharmacogenomics</i> , 2019, 20, 791-801.	1.3	3
131	Disorders of FZ-CRD; insights towards FZ-CRD folding and therapeutic landscape. <i>Molecular Medicine</i> , 2020, 26, 4.	4.4	3
132	Multomics Analysis Coupled with Text Mining Identify Novel Biomarker Candidates for Recurrent Cardiovascular Events. <i>OMICS A Journal of Integrative Biology</i> , 2020, 24, 205-215.	2.0	3
133	A Novel Homozygous Missense Variant in the NAGA Gene with Extreme Intrafamilial Phenotypic Heterogeneity. <i>Journal of Molecular Neuroscience</i> , 2020, 70, 45-55.	2.3	2
134	Mapping the Educational Environment of Genomics and Pharmacogenomics in the United Arab Emirates: A Mixed-Methods Triangulated Design. <i>OMICS A Journal of Integrative Biology</i> , 2021, 25, 285-293.	2.0	2
135	A response to Dr. Alzahrani's letter to the editor regarding the mechanism underlying fibrochondrogenesis. <i>Gene</i> , 2013, 528, 367-368.	2.2	1
136	Electrical Detection of Innate Immune Cells. <i>Sensors</i> , 2021, 21, 5886.	3.8	1
137	Spondylometaepiphyseal Dysplasia Short Limb-Abnormal Calcification Type in Turkish Patients Reveals a Novel Mutation and New Features. <i>Molecular Syndromology</i> , 2022, 13, 23-37.	0.8	1
138	Genetic Disorders in the United Arab Emirates. , 2010, , 639-676.		1
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