Bassam R Ali

List of Publications by Year in descending order

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144 papers	3,916 citations	172457 29 h-index	161849 54 g-index
149 all docs	149 docs citations	149 times ranked	6525 citing authors

#	Article	IF	Citations
1	Spondylometaepiphyseal Dysplasia Short Limb-Abnormal Calcification Type in Turkish Patients Reveals a Novel Mutation and New Features. Molecular Syndromology, 2022, 13, 23-37.	0.8	1
2	Clinical implementation of drug metabolizing gene-based therapeutic interventions worldwide. Human Genetics, 2022, 141, 1137-1157.	3.8	6
3	<scp>Biâ€allelic</scp> null variant in matrix metalloproteinaseâ€15, causes congenital cardiac defect, cholestasis jaundice, and failure to thrive. Clinical Genetics, 2022, 101, 403-410.	2.0	4
4	Endoglin Wild Type and Variants Associated With Hereditary Hemorrhagic Telangiectasia Type 1 Undergo Distinct Cellular Degradation Pathways. Frontiers in Molecular Biosciences, 2022, 9, 828199.	3.5	4
5	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	2.5	8
6	HLA-DRB1 and â€“DQB1 Alleles, Haplotypes and Genotypes in Emirati Patients with Type 1 Diabetes Underscores the Benefits of Evaluating Understudied Populations. Frontiers in Genetics, 2022, 13, 841879.	2.3	4
7	Mendelian randomization in pharmacogenomics: The unforeseen potentials. Biomedicine and Pharmacotherapy, 2022, 150, 112952.	5.6	10
8	Congenital Teratocarcinosarcoma With CTNNB1 Gene Mutation Presenting as an Ocular Mass. Pediatric and Developmental Pathology, 2022, 25, 562-567.	1.0	4
9	Association of variants inPTPN22,CTLAâ€4,IL2â€RA, andINSgenes with type 1 diabetes in Emiratis. Annals of Human Genetics, 2021, 85, 48-57.	0.8	8
10	Computer-aided approaches reveal trihydroxychroman and pyrazolone derivatives as potential inhibitors of SARS-CoV-2 virus main protease. Acta Pharmaceutica, 2021, 71, 325-333.	2.0	4
11	ACE2 Nascence, trafficking, and SARS-CoV-2 pathogenesis: the saga continues. Human Genomics, 2021, 15, 8.	2.9	28
12	Prevalence of pharmacogenomic variants in 100 pharmacogenes among Southeast Asian populations under the collaboration of the Southeast Asian Pharmacogenomics Research Network (SEAPharm). Human Genome Variation, 2021, 8, 7.	0.7	19
13	Stakeholdersâ€™ Interest and Attitudes toward Genomic Medicine and Pharmacogenomics Implementation in the United Arab Emirates: A Qualitative Study. Public Health Genomics, 2021, 24, 99-109.	1.0	8
14	Mapping the Educational Environment of Genomics and Pharmacogenomics in the United Arab Emirates: A Mixed-Methods Triangulated Design. OMICS A Journal of Integrative Biology, 2021, 25, 285-293.	2.0	2
15	Pathological Crosstalk Between Oxidized LDL and ER Stress in Human Diseases: A Comprehensive Review. Frontiers in Cell and Developmental Biology, 2021, 9, 674103.	3.7	24
16	Current opinion on the pharmacogenomics of paclitaxel-induced toxicity. Expert Opinion on Drug Metabolism and Toxicology, 2021, 17, 785-801.	3.3	29
17	Electrical Detection of Innate Immune Cells. Sensors, 2021, 21, 5886.	3.8	1
18	Long-Term Effects of Pediatric Acute Lymphoblastic Leukemia Chemotherapy: Can Recent Findings Inform Old Strategies?. Frontiers in Oncology, 2021, 11, 710163.	2.8	21

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19	Development of the pharmacogenomics and genomics literacy framework for pharmacists. Human Genomics, 2021, 15, 62.	2.9	11
20	Role of Ceramides in the Molecular Pathogenesis and Potential Therapeutic Strategies of Cardiometabolic Diseases: What we Know so Far. Frontiers in Cell and Developmental Biology, 2021, 9, 816301.	3.7	6
21	A Novel Homozygous Missense Variant in the NAGA Gene with Extreme Intrafamilial Phenotypic Heterogeneity. Journal of Molecular Neuroscience, 2020, 70, 45-55.	2.3	2
22	Disorders of FZ-CRD; insights towards FZ-CRD folding and therapeutic landscape. Molecular Medicine, 2020, 26, 4.	4.4	3
23	Evaluating the Role of MAST1 as an Intellectual Disability Disease Gene: Identification of a Novel De Novo Variant in a Patient with Developmental Disabilities. Journal of Molecular Neuroscience, 2020, 70, 320-327.	2.3	10
24	Genomics and Pharmacogenomics Knowledge, Attitude and Practice of Pharmacists Working in United Arab Emirates: Findings from Focus Group Discussionsâ€"A Qualitative Study. Journal of Personalized Medicine, 2020, 10, 134.	2.5	17
25	Proteostasis Regulation in the Endoplasmic Reticulum: An Emerging Theme in the Molecular Pathology and Therapeutic Management of Familial Hypercholesterolemia. Frontiers in Genetics, 2020, 11, 570355.	2.3	6
26	Knowledge, Attitudes, and Perceived Barriers toward Genetic Testing and Pharmacogenomics among Healthcare Workers in the United Arab Emirates: A Cross-Sectional Study. Journal of Personalized Medicine, 2020, 10, 216.	2.5	17
27	Endoplasmic Reticulum Associated Protein Degradation (ERAD) in the Pathology of Diseases Related to TGFÎ² Signaling Pathway: Future Therapeutic Perspectives. Frontiers in Molecular Biosciences, 2020, 7, 575608.	3.5	10
28	Variation in 100 relevant pharmacogenes among emiratis with insights from understudied populations. Scientific Reports, 2020, 10, 21310.	3.3	14
29	Knowledge and Attitudes of Medical and Health Science Students in the United Arab Emirates toward Genomic Medicine and Pharmacogenomics: A Cross-Sectional Study. Journal of Personalized Medicine, 2020, 10, 191.	2.5	16
30	Multiomics Analysis Coupled with Text Mining Identify Novel Biomarker Candidates for Recurrent Cardiovascular Events. OMICS A Journal of Integrative Biology, 2020, 24, 205-215.	2.0	3
31	The pharmacological chaperone N-n-butyl-deoxygalactonojirimycin enhances Î²-galactosidase processing and activity in fibroblasts of a patient with infantile GM1-gangliosidosis. Human Genetics, 2020, 139, 657-673.	3.8	6
32	Toxicity and Pharmacogenomic Biomarkers in Breast Cancer Chemotherapy. Frontiers in Pharmacology, 2020, 11, 445.	3.5	30
33	A Novel SLC1A4 Mutation (p.Y191*) Causes Spastic Tetraplegia, Thin Corpus Callosum, and Progressive Microcephaly (SPATCCM) With Seizure Disorder. Child Neurology Open, 2019, 6, 2329048X1988064.	1.1	13
34	Endoplasmic reticulum quality control of LDLR variants associated with familial hypercholesterolemia. FEBS Open Bio, 2019, 9, 1994-2005.	2.3	20
35	Genomic variants in members of the KrÃ¼ppel-like factor gene family are associated with disease severity and hydroxyurea treatment efficacy in Î²-hemoglobinopathies patients. Pharmacogenomics, 2019, 20, 791-801.	1.3	3
36	VKORC1 variants as significant predictors of warfarin dose in Emiratis . Pharmacogenomics and Personalized Medicine, 2019, Volume 12, 47-57.	0.7	6

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37	Role of Genomic Biomarkers in Increasing Fetal Hemoglobin Levels Upon Hydroxyurea Therapy and in Î²-Thalassemia Intermedia: A Validation Cohort Study. Hemoglobin, 2019, 43, 27-33.	0.8	7
38	Further Delineation of the Microcephaly-Micromelia Syndrome Associated with Loss-of-Function Variants in DONSON. Molecular Syndromology, 2019, 10, 171-176.	0.8	6
39	Cost-effectiveness analysis of pharmacogenomics-guided clopidogrel treatment in Spanish patients undergoing percutaneous coronary intervention. Pharmacogenomics Journal, 2019, 19, 438-445.	2.0	18
40	A B3GALT6 variant in patient originally described as Alâ€Gazali syndrome and implicating the endoplasmic reticulum quality control in the mechanism of some Î²3GalT6â€pathy mutations. Clinical Genetics, 2018, 93, 1148-1158.	2.0	12
41	Degradation routes of trafficking-defective VLDLR mutants associated with Dysequilibrium syndrome. Scientific Reports, 2018, 8, 1583.	3.3	10
42	Compound heterozygous variants in the multiple PDZ domain protein (MPDZ) cause a case of mild non-progressive communicating hydrocephalus. BMC Medical Genetics, 2018, 19, 34.	2.1	12
43	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
44	Defect in phosphoinositide signalling through a homozygous variant inPLCB3causes a new form of spondylometaphyseal dysplasia with corneal dystrophy. Journal of Medical Genetics, 2018, 55, 122-130.	3.2	9
45	West syndrome, developmental and epileptic encephalopathy, and severe CNS disorder associated with WWOX mutations. Epileptic Disorders, 2018, 20, 401-412.	1.3	23
46	A null variant in PUS3 confirms its involvement in intellectual disability and further delineates the associated neurodevelopmental disease. Clinical Genetics, 2018, 94, 586-587.	2.0	23
47	A recessive truncating variant in thrombospondinâ€1 domain containing protein 1 gene THSD1 is the underlying cause of nonimmune hydrops fetalis, congenital cardiac defects, and haemangiomas in four patients from a consanguineous family. American Journal of Medical Genetics, Part A, 2018, 176, 1996-2003.	1.2	10
48	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
49	Can VEGFA and ICAM1 polymorphisms predict response to bevacizumab?. Journal of Clinical Oncology, 2018, 36, 2521-2521.	1.6	0
50	A de novo mutation in the X-linked PAK3 gene is the underlying cause of intellectual disability and macrocephaly in monozygotic twins. European Journal of Medical Genetics, 2017, 60, 212-216.	1.3	17
51	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. Human Genetics, 2017, 136, 377-386.	3.8	36
52	Pharmacogenomics in pediatric acute lymphoblastic leukemia: promises and limitations. Pharmacogenomics, 2017, 18, 687-699.	1.3	13
53	Key Pharmacogenomic Considerations for Sickle Cell Disease Patients. OMICS A Journal of Integrative Biology, 2017, 21, 314-322.	2.0	6
54	Endoplasmic reticulum retention of xylosyltransferase 1 (XYLT1) mutants underlying Desbuquois dysplasia type II. American Journal of Medical Genetics, Part A, 2017, 173, 1773-1781.	1.2	12

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55	A novel aberrant splice site mutation in COL27A1 is responsible for Steel syndrome and extension of the phenotype to include hearing loss. American Journal of Medical Genetics, Part A, 2017, 173, 1257-1263.	1.2	21
56	Genomic Medicine Without Borders: Which Strategies Should Developing Countries Employ to Invest in Precision Medicine? A New â€œFast-Second Winnerâ€•Strategy. OMICS A Journal of Integrative Biology, 2017, 21, 647-657.	2.0	29
57	Studies on Nâ€Acetyltransferase (NAT2) Genotype Relationships in Emiratis: Confirmation of the Existence of Phenotype Variation among Slow Acetylators. Annals of Human Genetics, 2017, 81, 190-196.	0.8	6
58	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
59	Genomic Medicine in Developing Countries and Resource-Limited Environments. , 2017, , 459-467.		0
60	Intratumoral FoxP3+Helios+ Regulatory T Cells Upregulating Immunosuppressive Molecules Are Expanded in Human Colorectal Cancer. Frontiers in Immunology, 2017, 8, 619.	4.8	69
61	Genetic polymorphisms of cytochrome P450-1A2 (CYP1A2) among Emiratis. PLoS ONE, 2017, 12, e0183424.	2.5	16
62	Genomic variants in the FTO gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients. Human Genomics, 2017, 11, 30.	2.9	21
63	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 Î²-type hemoglobinopathy patients. Human Genomics, 2017, 11, 24.	2.9	11
64	Preferential accumulation of regulatory T cells with highly immunosuppressive characteristics in breast tumor microenvironment. Oncotarget, 2017, 8, 33159-33171.	1.8	96
65	Clinical and molecular delineation of dysequilibrium syndrome type 2 and profound sensorineural hearing loss in an inbred Arab family. American Journal of Medical Genetics, Part A, 2016, 170, 540-543.	1.2	16
66	A recessive syndrome of intellectual disability, moderate overgrowth, and renal dysplasia predisposing to Wilms tumor is caused by a mutation in FIBP gene. American Journal of Medical Genetics, Part A, 2016, 170, 2111-2118.	1.2	11
67	Novel genetic risk variants for pediatric celiac disease. Human Genomics, 2016, 10, 34.	2.9	9
68	A novel de novo mutation in DYNC1H1 gene underlying malformation of cortical development and cataract. Meta Gene, 2016, 9, 124-127.	0.6	23
69	Gonadal mosaicism in ARID1B gene causes intellectual disability and dysmorphic features in three siblings. American Journal of Medical Genetics, Part A, 2016, 170, 156-161.	1.2	16
70	Correlation of SIN3A genomic variants with Î²-hemoglobinopathies disease severity and hydroxyurea treatment efficacy. Pharmacogenomics, 2016, 17, 1785-1793.	1.3	12
71	Cover Image, Volume 170A, Number 8, August 2016. , 2016, 170, i-i.		0
72	A homozygous splicing mutation in ELAC2 suggests phenotypic variability including intellectual disability with minimal cardiac involvement. Orphanet Journal of Rare Diseases, 2016, 11, 139.	2.7	18

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73	FGF23â€“S129F mutant bypasses ER/Golgi to the circulation of hyperphosphatemic familial tumoral calcinosis patients. Bone, 2016, 93, 187-195.	2.9	11
74	The possible threat of Zika virus in the Middle East. Travel Medicine and Infectious Disease, 2016, 14, 277-278.	3.0	3
75	Genomic variants in the ASS1 gene, involved in the nitric oxide biosynthesis and signaling pathway, predict hydroxyurea treatment efficacy in compound sickle cell disease/Î²-thalassemia patients. Pharmacogenomics, 2016, 17, 393-403.	1.3	10
76	Secondary association of PDLIM5 with paranoid schizophrenia in Emirati patients. Meta Gene, 2015, 5, 135-139.	0.6	5
77	New Arab family with cerebral dysgenesis, neuropathy, ichthyosis and keratoderma syndrome suggests a possible founder effect for the c.223delG mutation. Journal of Dermatology, 2015, 42, 821-822.	1.2	8
78	Defective cellular trafficking of the bone morphogenetic protein receptor type II by mutations underlying familial pulmonary arterial hypertension. Gene, 2015, 561, 148-156.	2.2	22
79	Success stories in genomic medicine from resource-limited countries. Human Genomics, 2015, 9, 11.	2.9	41
80	A Novel Whole Exon Deletion in WWOX Gene Causes Early Epilepsy, Intellectual Disability and Optic Atrophy. Journal of Molecular Neuroscience, 2015, 56, 17-23.	2.3	40
81	Asparagine synthetase deficiency detected by whole exome sequencing causes congenital microcephaly, epileptic encephalopathy and psychomotor delay. Metabolic Brain Disease, 2015, 30, 687-694.	2.9	38
82	Improved plasma membrane expression of the trafficking defective P344R mutant of muscle, skeletal, receptor tyrosine kinase (MuSK) causing congenital myasthenic syndrome. International Journal of Biochemistry and Cell Biology, 2015, 60, 119-129.	2.8	6
83	A Novel Single-Nucleotide Deletion (c.1020delA) in NSUN2 Causes Intellectual Disability in an Emirati Child. Journal of Molecular Neuroscience, 2015, 57, 393-399.	2.3	39
84	A novel splice site deletion in the OFD1 gene is responsible for oralâ€“facialâ€“digital syndrome type 1 in an Emirati child. Hamdan Medical Journal, 2015, 8, 155.	0.1	0
85	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	2.9	47
86	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
87	Clinical and Molecular Analysis of a Novel COLQ Missense Mutation Causing Congenital Myasthenic Syndrome in a Syrian Family. Pediatric Neurology, 2014, 51, 165-169.	2.1	16
88	Analysis of two Arab families reveals additional support for a DFNB2 nonsyndromic phenotype of MYO7A. Molecular Biology Reports, 2014, 41, 193-200.	2.3	10
89	The mutational spectrum of the NF1 gene in neurofibromatosis type I patients from UAE. Child's Nervous System, 2014, 30, 1183-1189.	1.1	8
90	Impaired trafficking of the very low density lipoprotein receptor caused by missense mutations associated with dysequilibrium syndrome. Biochimica Et Biophysica Acta - Molecular Cell Research, 2014, 1843, 2871-2877.	4.1	11

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91	A novel mutation in DDR2 causing spondylo-meta-epiphyseal dysplasia with short limbs and abnormal calcifications (SMED-SL) results in defective intra-cellular trafficking. BMC Medical Genetics, 2014, 15, 42.	2.1	30
92	Mutation spectrum of Joubert syndrome and related disorders among Arabs. Human Genome Variation, 2014, 1, 14020.	0.7	31
93	A progeroid syndrome with neonatal presentation and long survival maps to 19p13.3p13.2. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 456-462.	1.6	9
94	LINS, a modulator of the WNT signaling pathway, is involved in human cognition. Orphanet Journal of Rare Diseases, 2013, 8, 87.	2.7	21
95	Determination of the CCR5âˆ†32 frequency in Emiratis and Tunisians and the screening of the CCR5 gene for novel alleles in Emiratis. Gene, 2013, 529, 113-118.	2.2	7
96	Delineation of the Clinical, Molecular and Cellular Aspects of Novel JAM 3 Mutations Underlying the Autosomal Recessive Hemorrhagic Destruction of the Brain, Subependymal Calcification, and Congenital Cataracts. Human Mutation, 2013, 34, 498-505.	2.5	30
97	Retention in the endoplasmic reticulum is the underlying mechanism of some hereditary haemorrhagic telangiectasia type 2 ALK1 missense mutations. Molecular and Cellular Biochemistry, 2013, 373, 247-257.	3.1	18
98	Novel KCNQ2 Mutation in a Large Emirati Family With Benign Familial Neonatal Seizures. Pediatric Neurology, 2013, 48, 63-66.	2.1	5
99	A response to Dr. Alzahrani's letter to the editor regarding the mechanism underlying fibrochondrogenesis. Gene, 2013, 528, 367-368.	2.2	1
100	Novel mutations in ADAMTSL2 gene underlying geleophysic dysplasia in families from United Arab Emirates. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 764-769.	1.6	8
101	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
102	Clinical and molecular analysis of isovaleric acidemia patients in the United Arab Emirates reveals remarkable phenotypes and four novel mutations in the IVD gene. European Journal of Medical Genetics, 2012, 55, 671-676.	1.3	17
103	Prevalence and Novel Mutations of Lysosomal Storage Disorders in United Arab Emirates. JIMD Reports, 2012, 10, 1-9.	1.5	44
104	A missense founder mutation in VLDLR is associated with Dysequilibrium Syndrome without quadrupedal locomotion. BMC Medical Genetics, 2012, 13, 80.	2.1	31
105	A mutation in KIF7 is responsible for the autosomal recessive syndrome of macrocephaly, multiple epiphyseal dysplasia and distinctive facial appearance. Orphanet Journal of Rare Diseases, 2012, 7, 27.	2.7	29
106	Identification of Mutations Underlying 20 Inborn Errors of Metabolism in the United Arab Emirates Population. Genetic Testing and Molecular Biomarkers, 2012, 16, 366-371.	0.7	7
107	A novel mutation in PRG4 gene underlying camptodactylyâ€arthropathyâ€coxa varaâ€pericarditis syndrome with the possible expansion of the phenotype to include congenital cataract. Birth Defects Research Part A: Clinical and Molecular Teratology, 2012, 94, 553-556.	1.6	17
108	Clinical and molecular analysis of UAE fibrochondrogenesis patients expands the phenotype and reveals two COL11A1 homozygous null mutations. Clinical Genetics, 2012, 82, 147-156.	2.0	20

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109	StÃ¼veâ€“Wiedemann syndrome and related bent bone dysplasias. Clinical Genetics, 2012, 82, 12-21.	2.0	20
110	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	6.2	108
111	Is autosomal recessive Silverâ€“Russel syndrome a separate entity or is it part of the 3â€M syndrome spectrum?. American Journal of Medical Genetics, Part A, 2011, 155, 1236-1245.	1.2	25
112	Endoplasmic Reticulum Quality Control Is Involved in the Mechanism of Endoglin-Mediated Hereditary Haemorrhagic Telangiectasia. PLoS ONE, 2011, 6, e26206.	2.5	32
113	Identification of New Alleles and the Determination of Alleles and Genotypes Frequencies at the CYP2D6 Gene in Emiratis. PLoS ONE, 2011, 6, e28943.	2.5	27
114	New and known mutations associated with inborn errors of metabolism in a heterogeneous Middle Eastern population. Journal of King Abdulaziz University, Islamic Economics, 2011, 32, 353-9.	1.1	8
115	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
116	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
117	Molecular and clinical analysis of Ellis-van Creveld syndrome in the United Arab Emirates. BMC Medical Genetics, 2010, 11, 33.	2.1	15
118	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
119	A novel statin-mediated â€œprenylation block-and-releaseâ€•assay provides insight into the membrane targeting mechanisms of small GTPases. Biochemical and Biophysical Research Communications, 2010, 397, 34-41.	2.1	29
120	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	28.9	253
121	Genetic Disorders in the United Arab Emirates. , 2010, , 639-676.		1
122	Anterior segment anomalies of the eye, growth retardation associated with hypoplastic pituitary gland and endocrine abnormalities: Jung syndrome or a new syndrome?. American Journal of Medical Genetics, Part A, 2009, 149A, 251-256.	1.2	5
123	R58fs Mutation in the HGD Gene in a Family with Alkaptonuria in the UAE. Annals of Human Genetics, 2009, 73, 125-130.	0.8	15
124	Is cystic fibrosis-related diabetes an apoptotic consequence of ER stress in pancreatic cells?. Medical Hypotheses, 2009, 72, 55-57.	1.5	27
125	A novel mutation in ARG1 gene is responsible for arginase deficiency in an Asian family. Journal of King Abdulaziz University, Islamic Economics, 2009, 30, 1601-3.	1.1	9
126	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

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127	Defective cellular trafficking of missense NPR-B mutants is the major mechanism underlying acromesomelic dysplasia-type Maroteaux. Human Molecular Genetics, 2008, 18, 267-277.	2.9	36
128	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
129	Novel Robinow syndrome causing mutations in the proximal region of the frizzled-like domain of ROR2 are retained in the endoplasmic reticulum. Human Genetics, 2007, 122, 389-395.	3.8	38
130	Targeting of Rab GTPases to cellular membranes. Biochemical Society Transactions, 2005, 33, 652-656.	3.4	74
131	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
132	Reconstitution of glycopeptide export in mixed detergent-solubilised and resealed microsomes depleted of lumenal components. Journal of Proteomics, 2005, 62, 1-12.	2.4	0
133	Multiple regions contribute to membrane targeting of Rab GTPases. Journal of Cell Science, 2004, 117, 6401-6412.	2.0	100
134	Membrane Targeting of Rab GTPases Is Influenced by the Prenylation Motif. Molecular Biology of the Cell, 2003, 14, 1882-1899.	2.1	137
135	A Microsomal GTPase Is Required for Glycopeptide Export from the Mammalian Endoplasmic Reticulum. Journal of Biological Chemistry, 2000, 275, 33222-33230.	3.4	7
136	Export of a misprocessed GPI-anchored protein from the endoplasmic reticulum in vitro in an ATP- and cytosol-dependent manner. FEBS Letters, 2000, 483, 32-36.	2.8	8
137	The farnesyltransferase inhibitor manumycin A is a novel trypanocide with a complex mode of action including major effects on mitochondria. Molecular and Biochemical Parasitology, 1999, 104, 67-80.	1.1	27
138	Synergistic interaction of the cellulosome integrating protein (CipA) fromClostridium thermocellumwith a cellulosomal endoglucanase. FEBS Letters, 1998, 422, 221-224.	2.8	29
139	Similar processes mediate glycopeptide export from the endoplasmic reticulum in mammalian cells and Saccharomyces cerevisiae. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 6730-6734.	7.1	42
140	Characterization of the subunits in an apparently homogeneous subpopulation of Clostridium thermocellum cellulosomes. Enzyme and Microbial Technology, 1995, 17, 705-711.	3.2	40
141	Synthesis of 3-arsonoalanine and its action on aspartate aminotransferase and aspartate ammonia-lyase. Comparison with arsenical analogues of malate and fumarate. FEBS Journal, 1993, 215, 161-166.	0.2	6
142	The induction of chickpea glutathione S-transferase by oxadiazon. Phytochemistry, 1991, 30, 2131-2134.	2.9	4
143	Glutathione S-transferase from oxadiazon treated chickpea. Phytochemistry, 1990, 29, 2431-2435.	2.9	17
144	Pontocerebellar Hypoplasia Type 9: A New Case with a Novel Mutation and Review of Literature. Journal of Pediatric Genetics, 0, , .	0.7	0