Lihadh Al-Gazali

List of Publications by Year
in descending order

153 papers	10,618 citations	47006 47 h-index	34986 98 g-index
162 all docs	162 docs citations	162 times ranked	14939 citing authors

#	Article	IF	Citations
1	An SCN9A channelopathy causes congenital inability to experience pain. Nature, 2006, 444, 894-898.	27.8	1,353
2	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	21.4	383
3	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	8.1	383
4	Mutations in the Pericentrin (PCNT) Gene Cause Primordial Dwarfism. Science, 2008, 319, 816-819.	12.6	370
5	Mutations in CEP290, which encodes a centrosomal protein, cause pleiotropic forms of Joubert syndrome. Nature Genetics, 2006, 38, 623-625.	21.4	368
6	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
7	Mutations in LRP2, which encodes the multiligand receptor megalin, cause Donnai-Barrow and facio-oculo-acoustico-renal syndromes. Nature Genetics, 2007, 39, 957-959.	21.4	284
8	Mutations in the AHI1 Gene, Encoding Jouberin, Cause Joubert Syndrome with Cortical Polymicrogyria. American Journal of Human Genetics, 2004, 75, 979-987.	6.2	275
9	Mutations in CCBE1 cause generalized lymph vessel dysplasia in humans. Nature Genetics, 2009, 41, 1272-1274.	21.4	269
10	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	28.9	253
11	Genetic disorders in the Arab world. BMJ: British Medical Journal, 2006, 333, 831-834.	2.3	239
12	Phenotypic and Genetic Heterogeneity in Congenital Generalized Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 4840-4847.	3.6	217
13	Mutations in PYCR1 cause cutis laxa with progeroid features. Nature Genetics, 2009, 41, 1016-1021.	21.4	211
14	Whole exome sequencing identifies a splicing mutation in NSUN2 as a cause of a Dubowitz-like syndrome. Journal of Medical Genetics, 2012, 49, 380-385.	3.2	198
15	Mutations in the human laminin Î²2 (LAMB2) gene and the associated phenotypic spectruma. Human Mutation, 2010, 31, 992-1002.	2.5	184
16	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
17	Mutations in the Embryonal Subunit of the Acetylcholine Receptor (CHRNG) Cause Lethal and Escobar Variants of Multiple Pterygium Syndrome. American Journal of Human Genetics, 2006, 79, 390-395.	6.2	145
18	Mutations in WNT7A Cause a Range of Limb Malformations, Including Fuhrmann Syndrome and Al-Awadi/Raas-Rothschild/Schinzel Phocomelia Syndrome. American Journal of Human Genetics, 2006, 79, 402-408.	6.2	144

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19	High-resolution mtDNA evidence for the late-glacial resettlement of Europe from an Iberian refugium. Genome Research, 2005, 15, 19-24.	5.5	137
20	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	6.2	137
21	Homozygosity Mapping in Families with Joubert Syndrome Identifies a Locus on Chromosome 9q34.3 and Evidence for Genetic Heterogeneity. American Journal of Human Genetics, 1999, 65, 1666-1671.	6.2	135
22	Defective membrane expression of the Na ⁺ -HCO ₃ ^{â^'} cotransporter NBCe1 is associated with familial migraine. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 15963-15968.	7.1	135
23	Diagnostic criteria, clinical characteristics, and natural history of Cohen syndrome. Journal of Medical Genetics, 2003, 40, 233-241.	3.2	134
24	Hennekam syndrome can be caused by FAT4 mutations and be allelic to Van Maldergem syndrome. Human Genetics, 2014, 133, 1161-1167.	3.8	122
25	Centriolar satellites assemble centrosomal microcephaly proteins to recruit CDK2 and promote centriole duplication. ELife, 2015, 4, .	6.0	118
26	In search of triallelism in Bardetâ€“Biedl syndrome. European Journal of Human Genetics, 2012, 20, 420-427.	2.8	111
27	Biallelic mutations in SNX14 cause a syndromic form of cerebellar atrophy and lysosome-autophagosome dysfunction. Nature Genetics, 2015, 47, 528-534.	21.4	111
28	Faulty Initiation of Proteoglycan Synthesis Causes Cardiac and Joint Defects. American Journal of Human Genetics, 2011, 89, 15-27.	6.2	108
29	Distinguishing the four genetic causes of jouberts syndrome-related disorders. Annals of Neurology, 2005, 57, 513-519.	5.3	104
30	Functional Analysis of NBC1 Mutants Associated with Proximal Renal Tubular Acidosis and Ocular Abnormalities. Journal of the American Society of Nephrology: JASN, 2005, 16, 2270-2278.	6.1	101
31	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
32	<scp>TECRL</scp>, a new lifeâ€threatening inherited arrhythmia gene associated with overlapping clinical features of both <scp>LQTS</scp> and <scp>CPVT</scp>. EMBO Molecular Medicine, 2016, 8, 1390-1408.	6.9	98
33	Linkage Analysis in Families with Joubert Syndrome Plus Oculo-Renal Involvement Identifies the CORS2 Locus on Chromosome 11p12-q13.3. American Journal of Human Genetics, 2003, 73, 656-662.	6.2	93
34	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
35	A novel cerebello-ocular syndrome with abnormal glycosylation due to abnormalities in dolichol metabolism. Brain, 2010, 133, 3210-3220.	7.6	87
36	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84

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37	A novel NGF mutation clarifies the molecular mechanism and extends the phenotypic spectrum of the HSAN5 neuropathy. Journal of Medical Genetics, 2011, 48, 131-135.	3.2	83
38	Loss of the BMP Antagonist, SMOC-1, Causes Ophthalmo-Acromelic (Waardenburg Anophthalmia) Syndrome in Humans and Mice. PLoS Genetics, 2011, 7, e1002114.	3.5	81
39	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
40	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	2.5	75
41	A Novel Early Onset Lethal Form of Catecholaminergic Polymorphic Ventricular Tachycardia Maps to Chromosome 7p14-p22. Journal of Cardiovascular Electrophysiology, 2007, 18, 1060-1066.	1.7	74
42	Recurrent De Novo Mutations Affecting Residue Arg138 of Pyrroline-5-Carboxylate Synthase Cause a Progeroid Form of Autosomal-Dominant Cutis Laxa. American Journal of Human Genetics, 2015, 97, 483-492.	6.2	70
43	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
44	Mutation analysis of the MKKS gene in McKusick-Kaufman syndrome and selected Bardet-Biedl syndrome patients. Human Genetics, 2002, 110, 561-567.	3.8	65
45	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	6.2	62
46	A syndrome comprising childhood-onset glomerular kidney disease and ocular abnormalities with progressive loss of vision is caused by mutated LAMB2. Nephrology Dialysis Transplantation, 2006, 21, 3283-3286.	0.7	58
47	A novel disorder reveals clathrin heavy chain-22 is essential for human pain and touch development. Brain, 2015, 138, 2147-2160.	7.6	58
48	Genomic analysis of Meckelâ€“Gruber syndrome in Arabs reveals marked genetic heterogeneity and novel candidate genes. European Journal of Human Genetics, 2013, 21, 762-768.	2.8	56
49	Mutations inCEP120cause Joubert syndrome as well as complex ciliopathy phenotypes. Journal of Medical Genetics, 2016, 53, 608-615.	3.2	55
50	TCTEX1D2 mutations underlie Jeune asphyxiating thoracic dystrophy with impaired retrograde intraflagellar transport. Nature Communications, 2015, 6, 7074.	12.8	51
51	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
52	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
53	Consanguinity and Dysmorphology in Arabs. Human Heredity, 2014, 77, 93-107.	0.8	50
54	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	2.5	47

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55	METTL23, a transcriptional partner of GABPA, is essential for human cognition. Human Molecular Genetics, 2014, 23, 3456-3466.	2.9	47
56	Specific combinations of biallelic POLR3A variants cause Wiedemann-Rautenstrauch syndrome. Journal of Medical Genetics, 2018, 55, 837-846.	3.2	44
57	Distinguishing 3 classes of corpus callosal abnormalities in consanguineous families. Neurology, 2011, 76, 373-382.	1.1	43
58	A new locus for non-syndromal, autosomal recessive, sensorineural hearing loss (DFNB16) maps to human chromosome 15q21-q22.. Journal of Medical Genetics, 1997, 34, 1015-1017.	3.2	42
59	A new autosomal recessive syndrome of ocular colobomas, ichthyosis, brain malformations and endocrine abnormalities in an inbred Emirati family. American Journal of Medical Genetics, Part A, 2008, 146A, 813-819.	1.2	41
60	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
61	Centromeric inactivation in a dicentric human Y;21 translocation chromosome. Chromosoma, 1997, 106, 199-206.	2.2	40
62	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
63	Germline mutations in ABL1 cause an autosomal dominant syndrome characterized by congenital heart defects and skeletal malformations. Nature Genetics, 2017, 49, 613-617.	21.4	40
64	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
65	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
66	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
67	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
68	The spectrum of beta thalassaemia mutations in the UAE national population.. Journal of Medical Genetics, 1994, 31, 59-61.	3.2	35
69	Microlissencephaly. Pediatric Neurology, 1998, 18, 362-365.	2.1	32
70	Endoplasmic Reticulum Quality Control Is Involved in the Mechanism of Endoglin-Mediated Hereditary Haemorrhagic Telangiectasia. PLoS ONE, 2011, 6, e26206.	2.5	32
71	A missense founder mutation in VLDLR is associated with Dysequilibrium Syndrome without quadrupedal locomotion. BMC Medical Genetics, 2012, 13, 80.	2.1	31
72	Mutation spectrum of Joubert syndrome and related disorders among Arabs. Human Genome Variation, 2014, 1, 14020.	0.7	31

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73	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
74	Combining exome/genome sequencing with data repository analysis reveals novel geneâ€“disease associations for a wide range of genetic disorders. Genetics in Medicine, 2021, 23, 1551-1568.	2.4	30
75	Autosomal Recessive Micrencephaly with Simplified Gyral Pattern, Abnormal Myelination and Arthrogryposis. Neuropediatrics, 1999, 30, 141-145.	0.6	26
76	Community genetics. Its definition 2010. Journal of Community Genetics, 2010, 1, 19-22.	1.2	26
77	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
78	Scientists of the world speak up for equality. Nature, 2013, 495, 35-38.	27.8	23
79	A novel de novo mutation in DYNC1H1 gene underlying malformation of cortical development and cataract. Meta Gene, 2016, 9, 124-127.	0.6	23
80	PSMD12 haploinsufficiency in a neurodevelopmental disorder with autistic features. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 736-745.	1.7	23
81	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
82	A genetic aetiological survey of severe childhood deafness in the United Arab Emirates. Journal of Tropical Pediatrics, 1998, 44, 157-160.	1.5	22
83	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
84	LINS, a modulator of the WNT signaling pathway, is involved in human cognition. Orphanet Journal of Rare Diseases, 2013, 8, 87.	2.7	21
85	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
86	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
87	StÃ¼veâ€“Wiedemann syndrome and related bent bone dysplasias. Clinical Genetics, 2012, 82, 12-21.	2.0	20
88	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
89	Endoplasmic reticulum quality control of LDLR variants associated with familial hypercholesterolemia. FEBS Open Bio, 2019, 9, 1994-2005.	2.3	20
90	Clathrin heavy chain 22 contributes to the control of neuropeptide degradation and secretion during neuronal development. Scientific Reports, 2018, 8, 2340.	3.3	19

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91	DYRK1A-related intellectual disability: a syndrome associated with congenital anomalies of the kidney and urinary tract. Genetics in Medicine, 2019, 21, 2755-2764.	2.4	19
92	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
93	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
94	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
95	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
96	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
97	Life-threatening arrhythmias with autosomal recessive TECRL variants. Europace, 2021, 23, 781-788.	1.7	17
98	A Novel Aberrant Splice Site Mutation in <i>RAB23</i> Leads to an Eight Nucleotide Deletion in the mRNA and Is Responsible for Carpenter Syndrome in a Consanguineous Emirati Family. Molecular Syndromology, 2012, 3, 255-261.	0.8	16
99	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
100	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
101	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
102	Molecular and clinical analysis of Ellis-van Creveld syndrome in the United Arab Emirates. BMC Medical Genetics, 2010, 11, 33.	2.1	15
103	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
104	Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases. Clinical Genetics, 2017, 91, 868-880.	2.0	14
105	Phosphoglucomutase-1 deficiency: Early presentation, metabolic management and detection in neonatal blood spots. Molecular Genetics and Metabolism, 2020, 131, 135-146.	1.1	14
106	PDZD8 Disruption Causes Cognitive Impairment in Humans, Mice, and Fruit Flies. Biological Psychiatry, 2022, 92, 323-334.	1.3	14
107	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
108	Regulation of human cerebral cortical development by EXOC7 and EXOC8, components of the exocyst complex, and roles in neural progenitor cell proliferation and survival. Genetics in Medicine, 2020, 22, 1040-1050.	2.4	13

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109	A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. Genetics in Medicine, 2021, 23, 1158-1162.	2.4	13
110	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
111	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
112	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
113	Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene. Journal of Medical Genetics, 2002, 39, 634-638.	3.2	11
114	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
115	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
116	Hypotonia, developmental delay and features of scalpâ€“earâ€“nipple syndrome in an inbred Arab family. Clinical Dysmorphology, 2007, 16, 105-107.	0.3	10
117	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
118	Degradation routes of trafficking-defective VLDLR mutants associated with Dysequilibrium syndrome. Scientific Reports, 2018, 8, 1583.	3.3	10
119	Expanding the clinical and genetic spectra of NKX6â€2â€related disorder. Clinical Genetics, 2018, 93, 1087-1092.	2.0	10
120	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
121	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
122	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
123	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
124	Stuveâ€“Wiedemann syndrome: a skeletal dysplasia characterized by bowed long bones. Ultrasound in Obstetrics and Gynecology, 2011, 38, 553-558.	1.7	8
125	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
126	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

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127	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
128	Homozygous missense variant in BMPR1A resulting in BMPR signaling disruption and syndromic features. Molecular Genetics & Genomic Medicine, 2019, 7, e969.	1.2	8
129	Breast cancer, consanguinity, and lethal tumor genes: simulation of BRCA1/2 prevalence over 40 generations. International Journal of Molecular Medicine, 2002, 10, 713-9.	4.0	8
130	Normal glycosylation screening does not rule out SRD5A3-CDG. European Journal of Human Genetics, 2011, 19, 1019-1019.	2.8	7
131	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
132	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
133	An Integrative Computational Approach for Prioritization of Genomic Variants. PLoS ONE, 2014, 9, e114903.	2.5	7
134	Improved diagnosis of a common mutation (R248C) in the human growth factor receptor 3 (FGFR3) gene that causes type I Thanatophoric dysplasia. Clinical Biochemistry, 2003, 36, 151-153.	1.9	6
135	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
136	VKORC1 variants as significant predictors of warfarin dose in Emiratis . Pharmacogenomics and Personalized Medicine, 2019, Volume 12, 47-57.	0.7	6
137	Further Delineation of the Microcephaly-Micromelia Syndrome Associated with Loss-of-Function Variants in DONSON. Molecular Syndromology, 2019, 10, 171-176.	0.8	6
138	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
139	Spectrum of neuroâ€genetic disorders in the United Arab Emirates national population. Clinical Genetics, 2021, 100, 573-600.	2.0	6
140	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
141	Novel KCNQ2 Mutation in a Large Emirati Family With Benign Familial Neonatal Seizures. Pediatric Neurology, 2013, 48, 63-66.	2.1	5
142	Fibrochondrogenesis: Prenatal diagnosis and outcome. Journal of Obstetrics and Gynaecology, 2013, 33, 663-668.	0.9	5
143	<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
144	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

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145	A syndrome of immune complex glomerulonephritis and ophthalmic abnormalities. Journal of Medical Genetics, 1999, 36, 641-4.	3.2	2
146	Human Genome Sequencing: Celebrating 10 Years. Science, 2011, 331, 873-873.	12.6	1
147	A response to Dr. Alzahrani's letter to the editor regarding the mechanism underlying fibrochondrogenesis. Gene, 2013, 528, 367-368.	2.2	1
148	Genetics and environmental risk factors associated with febrile seizures. Journal of Pediatric Neurology, 2015, 04, 239-243.	0.2	1
149	Genetic Disorders in the United Arab Emirates. , 2010, , 639-676.		1
150	412 Combination of Genomic Technologies and Consanguinity in Order to Identify Pathogenic Variants in Recessive Disorders. Archives of Disease in Childhood, 2012, 97, A121-A121.	1.9	0
151	Cover Image, Volume 170A, Number 8, August 2016. , 2016, 170, i-i.		0
152	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
153	Pontocerebellar Hypoplasia Type 9: A New Case with a Novel Mutation and Review of Literature. Journal of Pediatric Genetics, 0, , .	0.7	0