

Ranad Shaheen

List of Publications by Year in descending order

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Version: 2024-02-01

151
papers

8,462
citations

29994

54
h-index

56606

83
g-index

159
all docs

159
docs citations

159
times ranked

13248
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in phospholipase C eta-1 (<i>PLCH1</i>) are associated with holoprosencephaly. <i>Journal of Medical Genetics</i> , 2022, 59, 358-365.	1.5	3
2	Mitochondrial "dysmorphology" in variant classification. <i>Human Genetics</i> , 2022, 141, 55-64.	1.8	0
3	Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. <i>Human Mutation</i> , 2022, 43, 403-419.	1.1	9
4	THUMPD1 bi-allelic variants cause loss of tRNA acetylation and a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2022, 109, 587-600.	2.6	19
5	Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. <i>JCI Insight</i> , 2022, 7, .	2.3	6
6	Survey of disorders of sex development in a large cohort of patients with diverse Mendelian phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2789-2800.	0.7	7
7	Further delineation of <i>MYO18B</i> -related autosomal recessive Klippel-Feil syndrome with myopathy and facial dysmorphism. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 370-376.	0.7	4
8	Bi-allelic UBE4A loss-of-function variants cause intellectual disability and global developmental delay. <i>Genetics in Medicine</i> , 2021, 23, 661-668.	1.1	2
9	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. <i>American Journal of Human Genetics</i> , 2021, 108, 115-133.	2.6	37
10	Further delineation of <i>SMG9</i> -related heart and brain malformation syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1624-1630.	0.7	3
11	Transcriptome of CD8+ tumor-infiltrating T cells: a link between diabetes and colorectal cancer. <i>Cancer Immunology, Immunotherapy</i> , 2021, 70, 2625-2638.	2.0	3
12	Clinical, neuroimaging, and molecular spectrum of <i>TECPR2</i> -associated hereditary sensory and autonomic neuropathy with intellectual disability. <i>Human Mutation</i> , 2021, 42, 762-776.	1.1	18
13	Genetic testing results of children suspected to have Stickler syndrome type collagenopathy after ocular examination. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1628.	0.6	3
14	Bi-allelic premature truncating variants in <i>LTBP1</i> cause cutis laxa syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 1095-1114.	2.6	19
15	<i>PLXNA2</i> as a candidate gene in patients with intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3859-3865.	0.7	6
16	Missense <i>NAA20</i> variants impairing the NatB protein N-terminal acetyltransferase cause autosomal recessive developmental delay, intellectual disability, and microcephaly. <i>Genetics in Medicine</i> , 2021, 23, 2213-2218.	1.1	11
17	Lethal variants in humans: lessons learned from a large molecular autopsy cohort. <i>Genome Medicine</i> , 2021, 13, 161.	3.6	13
18	Bi-allelic Mutations in Tetratricopeptide Repeat Domain 26 (Intraflagellar Transport 56) Cause Severe Biliary Ciliopathy in Humans. <i>Hepatology</i> , 2020, 71, 2067-2079.	3.6	28

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19	Recessive, Deleterious Variants in SMG8 Expand the Role of Nonsense-Mediated Decay in Developmental Disorders in Humans. <i>American Journal of Human Genetics</i> , 2020, 107, 1178-1185.	2.6	20
20	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. <i>Genetics in Medicine</i> , 2020, 22, 2071-2080.	1.1	7
21	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. <i>Brain</i> , 2020, 143, 2911-2928.	3.7	13
22	The natural history of infantile neuroaxonal dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 109.	1.2	11
23	An exome-first approach to aid in the diagnosis of primary ciliary dyskinesia. <i>Human Genetics</i> , 2020, 139, 1273-1283.	1.8	16
24	Expanding the clinical and phenotypic heterogeneity associated with biallelic variants in ACO2. <i>Annals of Clinical and Translational Neurology</i> , 2020, 7, 1013-1028.	1.7	8
25	Analysis of transcript-deleterious variants in Mendelian disorders: implications for RNA-based diagnostics. <i>Genome Biology</i> , 2020, 21, 145.	3.8	59
26	A genomics approach to females with infertility and recurrent pregnancy loss. <i>Human Genetics</i> , 2020, 139, 605-613.	1.8	59
27	The morbid genome of ciliopathies: an update. <i>Genetics in Medicine</i> , 2020, 22, 1051-1060.	1.1	68
28	Phenome-based approach identifies RIC1-linked Mendelian syndrome through zebrafish models, biobank associations and clinical studies. <i>Nature Medicine</i> , 2020, 26, 98-109.	15.2	32
29	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. <i>Journal of Clinical Investigation</i> , 2020, 130, 4423-4439.	3.9	43
30	Exploiting the Autozygome to Support Previously Published Mendelian Gene-Disease Associations: An Update. <i>Frontiers in Genetics</i> , 2020, 11, 580484.	1.1	13
31	Biallelic variants in <i>CTU2</i> cause DREAM-PL syndrome and impair thiolation of tRNA wobble U34. <i>Human Mutation</i> , 2019, 40, 2108-2120.	1.1	25
32	MDH1 deficiency is a metabolic disorder of the malate-aspartate shuttle associated with early onset severe encephalopathy. <i>Human Genetics</i> , 2019, 138, 1247-1257.	1.8	31
33	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1543-1546.	0.7	5
34	Bi-allelic Mutations in FAM149B1 Cause Abnormal Primary Cilium and a Range of Ciliopathy Phenotypes in Humans. <i>American Journal of Human Genetics</i> , 2019, 104, 731-737.	2.6	23
35	Biallelic novel missense HHAT variant causes syndromic microcephaly and cerebellar-vermis hypoplasia. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1053-1057.	0.7	15
36	PUS7 mutations impair pseudouridylation in humans and cause intellectual disability and microcephaly. <i>Human Genetics</i> , 2019, 138, 231-239.	1.8	53

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37	Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , 2019, 21, 545-552.	1.1	85
38	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. <i>Clinical Genetics</i> , 2019, 95, 310-319.	1.0	12
39	Autozygome and high throughput confirmation of disease genes candidacy. <i>Genetics in Medicine</i> , 2019, 21, 736-742.	1.1	81
40	Congenital glaucoma and CYP1B1: an old story revisited. <i>Human Genetics</i> , 2019, 138, 1043-1049.	1.8	29
41	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , 2018, 20, 1609-1616.	1.1	46
42	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. <i>Clinical Genetics</i> , 2018, 93, 1210-1222.	1.0	38
43	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogyrosis. <i>American Journal of Human Genetics</i> , 2018, 102, 116-132.	2.6	46
44	GWAS signals revisited using human knockouts. <i>Genetics in Medicine</i> , 2018, 20, 64-68.	1.1	6
45	Molecular autopsy in maternal fetal medicine. <i>Genetics in Medicine</i> , 2018, 20, 420-427.	1.1	84
46	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2018, 128, 4313-4328.	3.9	89
47	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. <i>American Journal of Human Genetics</i> , 2018, 103, 612-620.	2.6	70
48	Warsaw breakage syndrome: Further clinical and genetic delineation. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2404-2418.	0.7	16
49	Mutations in known disease genes account for the majority of autosomal recessive retinal dystrophies. <i>Clinical Genetics</i> , 2018, 94, 554-563.	1.0	12
50	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. <i>Brain</i> , 2018, 141, 1934-1945.	3.7	70
51	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. <i>Nature Genetics</i> , 2017, 49, 537-549.	9.4	81
52	PLAA Mutations Cause a Lethal Infantile Epileptic Encephalopathy by Disrupting Ubiquitin-Mediated Endolysosomal Degradation of Synaptic Proteins. <i>American Journal of Human Genetics</i> , 2017, 100, 706-724.	2.6	37
53	GZF1 Mutations Expand the Genetic Heterogeneity of Larsen Syndrome. <i>American Journal of Human Genetics</i> , 2017, 100, 831-836.	2.6	14
54	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. <i>American Journal of Human Genetics</i> , 2017, 101, 23-36.	2.6	74

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55	The genetic landscape of familial congenital hydrocephalus. <i>Annals of Neurology</i> , 2017, 81, 890-897.	2.8	108
56	Autozygosity reveals recessive mutations and novel mechanisms in dominant genes: implications in variant interpretation. <i>Genetics in Medicine</i> , 2017, 19, 1144-1150.	1.1	77
57	Reply to "an extremely severe phenotype due to WDR81 nonsense mutations". <i>Annals of Neurology</i> , 2017, 82, 651-651.	2.8	1
58	A null mutation in MICU2 causes abnormal mitochondrial calcium homeostasis and a severe neurodevelopmental disorder. <i>Brain</i> , 2017, 140, 2806-2813.	3.7	38
59	Mutations of <i>KIF14</i> cause primary microcephaly by impairing cytokinesis. <i>Annals of Neurology</i> , 2017, 82, 562-577.	2.8	62
60	Molecular and clinical spectra of FBXL4 deficiency. <i>Human Mutation</i> , 2017, 38, 1649-1659.	1.1	41
61	Expanding the genetic heterogeneity of intellectual disability. <i>Human Genetics</i> , 2017, 136, 1419-1429.	1.8	122
62	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. <i>American Journal of Human Genetics</i> , 2017, 101, 206-217.	2.6	51
63	Expanding the allelic disorders linked to <i>TCTN1</i> to include Varadi syndrome (Orofaciodigital) Tj ETQq1 1 0.784314 rgBT /Overlo 0.7	0.7	4
64	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. <i>Nature Genetics</i> , 2017, 49, 36-45.	9.4	251
65	Increasing the sensitivity of clinical exome sequencing through improved filtration strategy. <i>Genetics in Medicine</i> , 2017, 19, 593-598.	1.1	59
66	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. <i>Molecular Psychiatry</i> , 2017, 22, 615-624.	4.1	187
67	Novel phenotypes and loci identified through clinical genomics approaches to pediatric cataract. <i>Human Genetics</i> , 2017, 136, 205-225.	1.8	73
68	A novel mechanism for variable phenotypic expressivity in Mendelian diseases uncovered by an AU-rich element (ARE)-creating mutation. <i>Genome Biology</i> , 2017, 18, 144.	3.8	19
69	Gonadal mosaicism for <i>ACTA1</i> gene masquerading as autosomal recessive nemaline myopathy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2219-2221.	0.7	8
70	<i>ADAT3</i> -related intellectual disability: Further delineation of the phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1142-1147.	0.7	32
71	Epileptic encephalopathy with continuous spike-and-wave during sleep maps to a homozygous truncating mutation in AMPA receptor component <i>FRRS1L</i> . <i>Clinical Genetics</i> , 2016, 90, 282-283.	1.0	16
72	Novel copy number variants and major limb reduction malformation: Report of three cases. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1245-1250.	0.7	8

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73	Characterizing the morbid genome of ciliopathies. <i>Genome Biology</i> , 2016, 17, 242.	3.8	118
74	Hyperekplexia, microcephaly and simplified gyral pattern caused by novel ASNS mutations, case report. <i>BMC Neurology</i> , 2016, 16, 105.	0.8	32
75	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. <i>Human Genetics</i> , 2016, 135, 525-540.	1.8	89
76	On the phenotypic spectrum of serine biosynthesis defects. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 373-381.	1.7	45
77	A homozygous truncating mutation in PUS3 expands the role of tRNA modification in normal cognition. <i>Human Genetics</i> , 2016, 135, 707-713.	1.8	112
78	Clinical genomics can facilitate countrywide estimation of autosomal recessive disease burden. <i>Genetics in Medicine</i> , 2016, 18, 1244-1249.	1.1	82
79	Cell-Intrinsic Adaptation Arising from Chronic Ablation of a Key Rho GTPase Regulator. <i>Developmental Cell</i> , 2016, 39, 28-43.	3.1	40
80	Mutations in CIT, encoding citron rho-interacting serine/threonine kinase, cause severe primary microcephaly in humans. <i>Human Genetics</i> , 2016, 135, 1191-1197.	1.8	30
81	A lethal phenotype associated with tissue plasminogen deficiency in humans. <i>Human Genetics</i> , 2016, 135, 1209-1211.	1.8	2
82	The syndrome dysmorphic facies, renal agenesis, ambiguous genitalia, microcephaly, polydactyly and lissencephaly (DREAMâ€PL): Report of two additional patients. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3222-3226.	0.7	17
83	Homozygous KCNMA1 mutation as a cause of cerebellar atrophy, developmental delay and seizures. <i>Human Genetics</i> , 2016, 135, 1295-1298.	1.8	65
84	Phenotype variability of infantile-onset multisystem neurologic, endocrine, and pancreatic disease IMNEPD. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 52.	1.2	13
85	FBXO32, encoding a member of the SCF complex, is mutated in dilated cardiomyopathy. <i>Genome Biology</i> , 2016, 17, 2.	3.8	35
86	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. <i>Genetics in Medicine</i> , 2016, 18, 686-695.	1.1	55
87	Mutations in SMC9, Encoding an Essential Component of Nonsense-Mediated Decay Machinery, Cause a Multiple Congenital Anomaly Syndrome in Humans and Mice. <i>American Journal of Human Genetics</i> , 2016, 98, 643-652.	2.6	51
88	Exome-based caseâ€control association study using extreme phenotype design reveals novel candidates with protective effect in diabetic retinopathy. <i>Human Genetics</i> , 2016, 135, 193-200.	1.8	45
89	Expanding the clinical, allelic, and locus heterogeneity of retinal dystrophies. <i>Genetics in Medicine</i> , 2016, 18, 554-562.	1.1	89
90	Matching Two Independent Cohorts Validates <i>DPH1</i> as a Gene Responsible for Autosomal Recessive Intellectual Disability with Short Stature, Craniofacial, and Ectodermal Anomalies. <i>Human Mutation</i> , 2015, 36, 1015-1019.	1.1	32

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91	Identification of embryonic lethal genes in humans by autozygosity mapping and exome sequencing in consanguineous families. <i>Genome Biology</i> , 2015, 16, 116.	3.8	91
92	A founder CEP120 mutation in Jeune asphyxiating thoracic dystrophy expands the role of centriolar proteins in skeletal ciliopathies. <i>Human Molecular Genetics</i> , 2015, 24, 1410-1419.	1.4	70
93	Mutation in WDR4 impairs tRNA m7G46 methylation and causes a distinct form of microcephalic primordial dwarfism. <i>Genome Biology</i> , 2015, 16, 210.	3.8	132
94	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. <i>Cell Reports</i> , 2015, 10, 148-161.	2.9	375
95	Revisiting disease genes based on whole-exome sequencing in consanguineous populations. <i>Human Genetics</i> , 2015, 134, 1029-1034.	1.8	17
96	Positional mapping of <i>PRKD1</i> , <i>NRP1</i> and <i>PRDM1</i> as novel candidate disease genes in truncus arteriosus. <i>Journal of Medical Genetics</i> , 2015, 52, 322-329.	1.5	30
97	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. <i>Genetics in Medicine</i> , 2015, 17, 719-725.	1.1	19
98	T (brachyury) is linked to a Mendelian form of neural tube defects in humans. <i>Human Genetics</i> , 2015, 134, 1139-1141.	1.8	13
99	Intrafamilial clinical heterogeneity of <i>CSPP1</i> -related ciliopathy. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2478-2480.	0.7	9
100	Identification of a novel MKS locus defined by <i>TMEM107</i> mutation. <i>Human Molecular Genetics</i> , 2015, 24, 5211-5218.	1.4	42
101	Variable phenotypic expression of COG6 mutations. <i>Journal of Medical Genetics</i> , 2014, 51, 425.2-426.	1.5	2
102	Mutation in <i>PLK4</i> , encoding a master regulator of centriole formation, defines a novel locus for primordial dwarfism. <i>Journal of Medical Genetics</i> , 2014, 51, 814-816.	1.5	35
103	Genomic analysis of primordial dwarfism reveals novel disease genes. <i>Genome Research</i> , 2014, 24, 291-299.	2.4	142
104	Mutations in <i>CSPP1</i> , Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. <i>American Journal of Human Genetics</i> , 2014, 94, 73-79.	2.6	77
105	Identification of three novel <i>ECEL1</i> mutations in three families with distal arthrogyriposis type 5D. <i>Clinical Genetics</i> , 2014, 85, 568-572.	1.0	29
106	The <i>ECEL1</i> -related strabismus phenotype is consistent with congenital cranial dysinnervation disorder. <i>Journal of AAPOS</i> , 2014, 18, 362-367.	0.2	22
107	<i>CC2D1A</i> Regulates Human Intellectual and Social Function as well as NF- κ B Signaling Homeostasis. <i>Cell Reports</i> , 2014, 8, 647-655.	2.9	60
108	Neu-Laxova Syndrome, an Inborn Error of Serine Metabolism, Is Caused by Mutations in <i>PHGDH</i> . <i>American Journal of Human Genetics</i> , 2014, 94, 898-904.	2.6	93

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109	A truncating mutation in B3GNT1 causes severe Walker-Warburg syndrome. <i>Neurogenetics</i> , 2013, 14, 243-245.	0.7	31
110	Mutations in EOGT Confirm the Genetic Heterogeneity of Autosomal-Recessive Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2013, 92, 598-604.	2.6	114
111	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 1108-1117.	2.6	147
112	Mutations in DDX59 Implicate RNA Helicase in the Pathogenesis of Orofaciodigital Syndrome. <i>American Journal of Human Genetics</i> , 2013, 93, 555-560.	2.6	45
113	Mutation in <i>ADAT3</i> , encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus. <i>Journal of Medical Genetics</i> , 2013, 50, 425-430.	1.5	91
114	Genomic analysis of Meckel-Gruber syndrome in Arabs reveals marked genetic heterogeneity and novel candidate genes. <i>European Journal of Human Genetics</i> , 2013, 21, 762-768.	1.4	56
115	Mutations in <i>TMEM231</i> cause Meckel-Gruber syndrome. <i>Journal of Medical Genetics</i> , 2013, 50, 160-162.	1.5	34
116	A novel syndrome of hypohidrosis and intellectual disability is linked to COG6 deficiency. <i>Journal of Medical Genetics</i> , 2013, 50, 431-436.	1.5	40
117	Autozygome-guided exome sequencing in retinal dystrophy patients reveals pathogenetic mutations and novel candidate disease genes. <i>Genome Research</i> , 2013, 23, 236-247.	2.4	226
118	WNT1 mutation with recessive osteogenesis imperfecta and profound neurological phenotype. <i>Journal of Medical Genetics</i> , 2013, 50, 491-492.	1.5	45
119	Autozygome maps dispensable DNA and reveals potential selective bias against nullizygoty. <i>Genetics in Medicine</i> , 2012, 14, 515-519.	1.1	10
120	Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes. <i>Journal of Medical Genetics</i> , 2012, 49, 234-241.	1.5	164
121	Study of autosomal recessive osteogenesis imperfecta in Arabia reveals a novel locus defined by <i>TMEM38B</i> mutation. <i>Journal of Medical Genetics</i> , 2012, 49, 630-635.	1.5	124
122	In search of triallelism in Bardet-Biedl syndrome. <i>European Journal of Human Genetics</i> , 2012, 20, 420-427.	1.4	111
123	Vanishing white matter disease caused by EIF2B2 mutation with the presentation of an adrenoleukodystrophy phenotype. <i>Gene</i> , 2012, 496, 141-143.	1.0	5
124	POC1A Truncation Mutation Causes a Ciliopathy in Humans Characterized by Primordial Dwarfism. <i>American Journal of Human Genetics</i> , 2012, 91, 330-336.	2.6	70
125	3M Syndrome: An Easily Recognizable yet Underdiagnosed Cause of Proportionate Short Stature. <i>Journal of Pediatrics</i> , 2012, 161, 139-145.e1.	0.9	27
126	Congenital disorder of glycosylation IIa: The trouble with diagnosing a dysmorphic inborn error of metabolism. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 245-246.	0.7	12

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127	Clinical, biochemical and molecular characterization of peroxisomal diseases in Arabs. <i>Clinical Genetics</i> , 2011, 79, 60-70.	1.0	18
128	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. <i>American Journal of Human Genetics</i> , 2011, 88, 536-547.	2.6	196
129	Human Mutations in NDE1 Cause Extreme Microcephaly with Lissencephaly. <i>American Journal of Human Genetics</i> , 2011, 88, 677.	2.6	3
130	Recessive Mutations in DOCK6, Encoding the Guanidine Nucleotide Exchange Factor DOCK6, Lead to Abnormal Actin Cytoskeleton Organization and Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2011, 89, 328-333.	2.6	115
131	Mutations in <i>FKBP10</i> cause both Bruck syndrome and isolated osteogenesis imperfecta in humans. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1448-1452.	0.7	63
132	A TCTN2 mutation defines a novel Meckel Gruber syndrome locus. <i>Human Mutation</i> , 2011, 32, 573-578.	1.1	72
133	Loss-of-function variant in DNASE1L3 causes a familial form of systemic lupus erythematosus. <i>Nature Genetics</i> , 2011, 43, 1186-1188.	9.4	366
134	Molecular Characterization of Newborn Glaucoma Including a Distinct Aniridic Phenotype. <i>Ophthalmic Genetics</i> , 2011, 32, 138-142.	0.5	40
135	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. <i>American Journal of Human Genetics</i> , 2010, 87, 306-307.	2.6	47
136	FKBP10 and Bruck Syndrome: Phenotypic Heterogeneity or Call for Reclassification?. <i>American Journal of Human Genetics</i> , 2010, 87, 571.	2.6	3
137	Persistence strategies of <i>Bacillus cereus</i> spores isolated from dairy silo tanks. <i>Food Microbiology</i> , 2010, 27, 347-355.	2.1	113
138	Clinical and molecular characterisation of Bardet-Biedl syndrome in consanguineous populations: the power of homozygosity mapping. <i>Journal of Medical Genetics</i> , 2010, 47, 236-241.	1.5	76
139	Novel CENPJ mutation causes Seckel syndrome. <i>Journal of Medical Genetics</i> , 2010, 47, 411-414.	1.5	149
140	Allelic heterogeneity in inbred populations: The Saudi experience with Alström syndrome as an illustrative example. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 662-665.	0.7	48
141	Zellweger syndrome caused by PEX13 deficiency: Report of two novel mutations. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1219-1223.	0.7	13
142	FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. <i>American Journal of Human Genetics</i> , 2009, 85, 414-418.	2.6	86
143	FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome. <i>American Journal of Human Genetics</i> , 2009, 85, 756.	2.6	1
144	Molecular characterization of retinitis pigmentosa in Saudi Arabia. <i>Molecular Vision</i> , 2009, 15, 2464-9.	1.1	61

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145	Environment driven cereulide production by emetic strains of <i>Bacillus cereus</i> . <i>International Journal of Food Microbiology</i> , 2008, 127, 60-67.	2.1	27
146	Occurrence of emetic toxin producing <i>Bacillus cereus</i> in the dairy production chain. <i>International Dairy Journal</i> , 2006, 16, 740-749.	1.5	69
147	Potential of selected infant food formulas for production of <i>Bacillus cereus</i> emetic toxin, cereulide. <i>International Journal of Food Microbiology</i> , 2006, 107, 287-294.	2.1	60
148	Emetic toxin-producing strains of <i>Bacillus cereus</i> show distinct characteristics within the <i>Bacillus cereus</i> group. <i>International Journal of Food Microbiology</i> , 2006, 109, 132-138.	2.1	141
149	Cereulide-producing strains of <i>Bacillus cereus</i> show diversity. <i>Archives of Microbiology</i> , 2005, 184, 141-151.	1.0	46
150	Identification and Partial Characterization of the Nonribosomal Peptide Synthetase Gene Responsible for Cereulide Production in Emetic <i>Bacillus cereus</i> . <i>Applied and Environmental Microbiology</i> , 2005, 71, 105-113.	1.4	249
151	Sperm bioassay for rapid detection of cereulide-producing <i>Bacillus cereus</i> in food and related environments. <i>International Journal of Food Microbiology</i> , 2004, 94, 175-183.	2.1	88