

Mustafa A. Salih

List of Publications by Year in descending order

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Version: 2024-02-01

130
papers

5,593
citations

94381

37
h-index

88593

70
g-index

132
all docs

132
docs citations

132
times ranked

9110
citing authors

#	ARTICLE	IF	CITATIONS
1	Charcot-Marie-Tooth type 4B is caused by mutations in the gene encoding myotubularin-related protein-2. <i>Nature Genetics</i> , 2000, 25, 17-19.	9.4	462
2	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
3	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. <i>Human Genetics</i> , 2017, 136, 921-939.	1.8	209
4	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2009, 85, 338-353.	2.6	208
5	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. <i>American Journal of Human Genetics</i> , 2019, 104, 1182-1201.	2.6	184
6	Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of Î±-Dystroglycan. <i>American Journal of Human Genetics</i> , 2013, 92, 354-365.	2.6	172
7	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. <i>American Journal of Human Genetics</i> , 2012, 91, 541-547.	2.6	167
8	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
9	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. <i>American Journal of Human Genetics</i> , 2013, 93, 482-495.	2.6	138
10	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. <i>Brain</i> , 2008, 131, 747-759.	3.7	134
11	Spinocerebellar ataxia with axonal neuropathy: consequence of a Tdp1 recessive neomorphic mutation?. <i>EMBO Journal</i> , 2007, 26, 4732-4743.	3.5	129
12	The tumour suppressor gene WWOX is mutated in autosomal recessive cerebellar ataxia with epilepsy and mental retardation. <i>Brain</i> , 2014, 137, 411-419.	3.7	127
13	Expanding the genetic heterogeneity of intellectual disability. <i>Human Genetics</i> , 2017, 136, 1419-1429.	1.8	122
14	Characterizing the morbid genome of ciliopathies. <i>Genome Biology</i> , 2016, 17, 242.	3.8	118
15	Congenital myasthenic syndromes due to mutations in <i>ALG2</i> and <i>ALG14</i> . <i>Brain</i> , 2013, 136, 944-956.	3.7	117
16	<i>SLC25A22</i> is a novel gene for migrating partial seizures in infancy. <i>Annals of Neurology</i> , 2013, 74, 873-882.	2.8	102
17	The clinical spectrum of homozygous <i>HOXA1</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1235-1240.	0.7	101
18	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. <i>Annals of Neurology</i> , 2019, 86, 821-831.	2.8	96

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19	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
20	Genomic and phenotypic delineation of congenital microcephaly. <i>Genetics in Medicine</i> , 2019, 21, 545-552.	1.1	85
21	Clinical characterization of the HOXA1 syndrome BSAS variant. <i>Neurology</i> , 2007, 69, 1245-1253.	1.5	83
22	Mutation in PHC1 implicates chromatin remodeling in primary microcephaly pathogenesis. <i>Human Molecular Genetics</i> , 2013, 22, 2200-2213.	1.4	81
23	Autozygome and high throughput confirmation of disease genes candidacy. <i>Genetics in Medicine</i> , 2019, 21, 736-742.	1.1	81
24	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. <i>Journal of Human Genetics</i> , 2017, 62, 243-252.	1.1	73
25	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. <i>Human Molecular Genetics</i> , 2014, 23, 5781-5792.	1.4	72
26	The morbid genome of ciliopathies: an update. <i>Genetics in Medicine</i> , 2020, 22, 1051-1060.	1.1	68
27	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. <i>European Journal of Human Genetics</i> , 2013, 21, 1074-1078.	1.4	64
28	Congenital Microcephaly with a Simplified Gyral Pattern: Associated Findings and Their Significance. <i>American Journal of Neuroradiology</i> , 2011, 32, 1123-1129.	1.2	62
29	CC2D1A 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
30	Molecular characterization of Joubert syndrome in Saudi Arabia. <i>Human Mutation</i> , 2012, 33, 1423-1428.	1.1	56
31	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
32	Megalencephalic leukoencephalopathy with cysts without <i>MLC1</i> defect. <i>Annals of Neurology</i> , 2010, 67, 834-837.	2.8	52
33	Ophthalmic Features of Joubert Syndrome. <i>Ophthalmology</i> , 2008, 115, 2286-2289.	2.5	48
34	Rundataxin, a novel protein with RUN and diacylglycerol binding domains, is mutant in a new recessive ataxia. <i>Brain</i> , 2010, 133, 2439-2447.	3.7	46
35	Expanding the phenome and variome of skeletal dysplasia. <i>Genetics in Medicine</i> , 2018, 20, 1609-1616.	1.1	46
36	Hemiconvulsion-hemiplegia-epilepsy syndrome. <i>Child's Nervous System</i> , 1997, 13, 257-263.	0.6	43

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37	New Findings in a Global Approach to Dissect the Whole Phenotype of PLA2G6 Gene Mutations. PLoS ONE, 2013, 8, e76831.	1.1	42
38	Features of a Large Epidemic of Group A Meningococcal Meningitis in Khartoum, Sudan in 1988. Scandinavian Journal of Infectious Diseases, 1990, 22, 161-170.	1.5	41
39	Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. American Journal of Human Genetics, 2016, 98, 1249-1255.	2.6	40
40	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Acta Neuropathologica, 2020, 139, 415-442.	3.9	38
41	The neurology of carbonic anhydrase type II deficiency syndrome. Brain, 2011, 134, 3502-3515.	3.7	37
42	Brain Stem and Cerebellar Findings in Joubert Syndrome. Journal of Computer Assisted Tomography, 2006, 30, 116-121.	0.5	35
43	Ophthalmic features of ataxia telangiectasia-like disorder. Journal of AAPOS, 2008, 12, 186-189.	0.2	35
44	A de novo marker chromosome derived from 9p in a patient with 9p partial duplication syndrome and autism features: genotype-phenotype correlation. BMC Medical Genetics, 2010, 11, 135.	2.1	34
45	A substance in broad beans (<i>Vicia faba</i>) is protective against experimentally induced convulsions in mice. Epilepsy and Behavior, 2008, 12, 25-29.	0.9	33
46	Hyperekplexia, microcephaly and simplified gyral pattern caused by novel ASNS mutations, case report. BMC Neurology, 2016, 16, 105.	0.8	32
47	Pathogenic variants in <i>KCTD7</i> perturb neuronal K ⁺ fluxes and glutamine transport. Brain, 2016, 139, 3109-3120.	3.7	31
48	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31
49	C19orf12 mutation leads to a pallido-pyramidal syndrome. Gene, 2014, 537, 352-356.	1.0	28
50	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	2.8	27
51	A Novel Form of Familial Congenital Muscular Dystrophy in Two Adolescents. Neuropediatrics, 1998, 29, 289-293.	0.3	25
52	A new complex homozygous large rearrangement of the PINK1 gene in a Sudanese family with early onset Parkinson's disease. Neurogenetics, 2009, 10, 265-270.	0.7	25
53	Case of partial trisomy 2q3 with clinical manifestations of Marshall-Smith syndrome. American Journal of Medical Genetics Part A, 1999, 85, 185-188.	2.4	24
54	Interleukin 10 Gene Polymorphisms and Development of Post Kala-Azar Dermal Leishmaniasis in a Selected Sudanese Population. Public Health Genomics, 2010, 13, 362-367.	0.6	24

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55	A null mutation in TNIK defines a novel locus for intellectual disability. <i>Human Genetics</i> , 2016, 135, 773-778.	1.8	23
56	Epidemiology, prenatal management, and prevention of neural tube defects. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2014, 35 Suppl 1, S15-28.	0.5	22
57	Lethal Congenital Muscular Dystrophy in Two Sibs with Arthrogryposis Multiplex: New Entity or Variant of Cobblestone Lissencephaly Syndrome?. <i>Neuropediatrics</i> , 1996, 27, 305-310.	0.3	21
58	Sleep-disordered breathing in children with craniosynostosis. <i>Sleep and Breathing</i> , 2013, 17, 389-393.	0.9	21
59	Acute Necrotizing Encephalopathy of Childhood: A Multicenter Experience in Saudi Arabia. <i>Frontiers in Pediatrics</i> , 2020, 8, 526.	0.9	20
60	Embryonal rhabdomyosarcoma and chromosomal breakage in a newborn infant with possible Dubowitz syndrome. , 2000, 92, 107-110.		19
61	Molecular and neurological characterizations of three Saudi families with lipid proteinosis. <i>BMC Medical Genetics</i> , 2011, 12, 31.	2.1	19
62	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
63	Genetic Refinement and Physical Mapping of the CMT4B Gene on Chromosome 11q22. <i>Genomics</i> , 2000, 63, 271-278.	1.3	18
64	The Salih Ataxia Mutation Impairs Rubicon Endosomal Localization. <i>Cerebellum</i> , 2013, 12, 835-840.	1.4	18
65	<i>NPHP4</i> mutation is linked to cerebelloâ€œculoâ€œrenal syndrome and male infertility. <i>Clinical Genetics</i> , 2014, 85, 371-375.	1.0	18
66	Rett Syndrome, a Neurodevelopmental Disorder, Whole-Transcriptome, and Mitochondrial Genome Multiomics Analyses Identify Novel Variations and Disease Pathways. <i>OMICS A Journal of Integrative Biology</i> , 2020, 24, 160-171.	1.0	18
67	Muscular dystrophy associated with ?-dystroglycan deficiency. <i>Annals of Neurology</i> , 1996, 40, 925-928.	2.8	17
68	Sotos syndrome (cerebral gigantism): a clinical and radiological study of 14 cases from Saudi Arabia. <i>Annals of Tropical Paediatrics</i> , 1999, 19, 197-203.	1.0	17
69	Ophthalmologic abnormalities in a de novo terminal 6q deletion. <i>Ophthalmic Genetics</i> , 2010, 31, 1-11.	0.5	17
70	Overlap of polymicrogyria, hydrocephalus, and Joubert syndrome in a family with novel truncating mutations in ADGRG1/GPR56 and KIAA0556. <i>Neurogenetics</i> , 2019, 20, 91-98.	0.7	17
71	Autosomal recessive hereditary neuropathy with focally folded myelin sheaths and linked to chromosome 11q23: a distinct and homogeneous entity. <i>Neuromuscular Disorders</i> , 2000, 10, 10-15.	0.3	16
72	Moyamoya syndrome with unusual angiographic findings and protein C deficiency: Review of the literature. <i>Journal of the Neurological Sciences</i> , 1998, 159, 11-16.	0.3	15

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73	Efficient identification of novel mutations in patients with limb girdle muscular dystrophy. <i>Neurogenetics</i> , 2010, 11, 449-455.	0.7	15
74	Ophthalmic features of <i>PLA2G6</i> -related paediatric neurodegeneration with brain iron accumulation. <i>British Journal of Ophthalmology</i> , 2014, 98, 889-893.	2.1	15
75	Impact of PYROXD1 deficiency on cellular respiration and correlations with genetic analyses of limb-girdle muscular dystrophy in Saudi Arabia and Sudan. <i>Physiological Genomics</i> , 2018, 50, 929-939.	1.0	15
76	Classification, clinical features, and genetics of neural tube defects. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2014, 35 Suppl 1, S5-S14.	0.5	15
77	Idiopathic intracranial hypertension in children: Diagnostic and management approach. <i>Sudanese Journal of Paediatrics</i> , 2016, 16, 67-76.	0.6	14
78	Childhood muscular dystrophy: an African review. <i>Annals of Tropical Paediatrics</i> , 1985, 5, 167-173.	1.0	13
79	Neurologic Injury in Isolated Sulfite Oxidase Deficiency. <i>Canadian Journal of Neurological Sciences</i> , 2014, 41, 42-48.	0.3	13
80	Intra-familial phenotypic heterogeneity in a Sudanese family with DARS2-related leukoencephalopathy, brainstem and spinal cord involvement and lactate elevation: a case report. <i>BMC Neurology</i> , 2018, 18, 175.	0.8	13
81	Ritscher-Schinzel (cranio-cerebello-cardiac, 3C) syndrome: Report of four new cases with renal involvement. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1393-1397.	0.7	12
82	Congenital Myasthenic Syndrome Due to Homozygous CHRNE Mutations: Report of Patients in Arabia. <i>Journal of Neuro-Ophthalmology</i> , 2011, 31, 42-47.	0.4	12
83	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
84	The natural history of infantile neuroaxonal dystrophy. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 109.	1.2	11
85	Stroke in Saudi children. Epidemiology, clinical features and risk factors. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S12-20.	0.5	11
86	Preimplantation Genetic Diagnosis in Isolated Sulfite Oxidase Deficiency. <i>Canadian Journal of Neurological Sciences</i> , 2013, 40, 109-112.	0.3	10
87	Prevalence of epilepsy in 74,949 school children in Khartoum State, Sudan. <i>Paediatrics and International Child Health</i> , 2017, 37, 188-192.	0.3	10
88	A novel syndrome of lethal familial hyperekplexia associated with brain malformation. <i>BMC Neurology</i> , 2012, 12, 125.	0.8	9
89	A newly recognized autosomal recessive syndrome affecting neurologic function and vision. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 1207-1213.	0.7	9
90	Cerebral Iron Accumulation Is Not a Major Feature of <i>FA2H</i> / <i>SPG35</i> Movement Disorders <i>Clinical Practice</i> , 2015, 2, 56-60.	0.8	9

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91	Expanding the clinical spectrum and allelic heterogeneity in van den Endeâ€“Gupta syndrome. <i>Clinical Genetics</i> , 2014, 85, 492-494.	1.0	8
92	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
93	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
94	NEW OBSERVATIONS REGARDING THE RETINOPATHY OF GENETICALLY CONFIRMED KEARNSâ€“SAYRE SYNDROME. <i>Retinal Cases and Brief Reports</i> , 2018, 12, 349-358.	0.3	8
95	A heterozygous mutation in the <i>CCDC88C</i> gene likely causes early-onset pure hereditary spastic paraplegia: a case report. <i>BMC Neurology</i> , 2021, 21, 78.	0.8	8
96	Genetic, chromosomal, and syndromic causes of neural tube defects. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2014, 35 Suppl 1, S49-56.	0.5	8
97	Infectious and inflammatory disorders of the circulatory system as risk factors for stroke in Saudi children. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S41-52.	0.5	8
98	Ancient founder mutation in <i>RUBCN</i> : a second unrelated family confirms Salih ataxia (<i>SCAR15</i>). <i>BMC Neurology</i> , 2020, 20, 207.	0.8	7
99	Genetic diagnosis in Sudanese and Tunisian families with syndromic intellectual disability through exome sequencing. <i>Annals of Human Genetics</i> , 2022, 86, 181-194.	0.3	7
100	Mutation in <i>GM2A</i> Leads to a Progressive Chorea-dementia Syndrome. <i>Tremor and Other Hyperkinetic Movements</i> , 2015, 5, 306.	1.1	6
101	Acute poisoning in a child following topical treatment of head lice (<i>pediculosis capitis</i>) with an organophosphate pesticide. <i>Sudanese Journal of Paediatrics</i> , 2016, 16, 63-6.	0.6	6
102	Broad beans () and the potential to protect from COVID-19 coronavirus infection. <i>Sudanese Journal of Paediatrics</i> , 2020, 20, 10-12.	0.6	6
103	Congenital and genetic cerebrovascular anomalies as risk factors for stroke in Saudi children. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S53-60.	0.5	6
104	Respiratory insufficiency in a severe autosomal recessive form of muscular dystrophy. <i>Annals of Tropical Paediatrics</i> , 1984, 4, 45-48.	1.0	5
105	An identicalâ€“byâ€“descent novel spliceâ€“donor variant in <i>PRUNE1</i> causes a neurodevelopmental syndrome with prominent dystonia in two consanguineous Sudanese families. <i>Annals of Human Genetics</i> , 2021, 85, 186-195.	0.3	5
106	Pathogenic Variants in <i>ABHD16A</i> Cause a Novel Psychomotor Developmental Disorder With Spastic Paraplegia. <i>Frontiers in Neurology</i> , 2021, 12, 720201.	1.1	5
107	Sturge-Weber syndrome: Continued vigilance is needed. <i>Sudanese Journal of Paediatrics</i> , 2015, 15, 63-70.	0.6	5
108	Perinatal stroke in Saudi children. Clinical features and risk factors. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S35-40.	0.5	5

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109	Optic disk and white matter abnormalities in a patient with a <i>de novo</i> 18p partial monosomy. <i>Ophthalmic Genetics</i> , 2010, 31, 147-154.	0.5	4
110	Duane retraction syndrome in a patient with Duchenne muscular dystrophy. <i>Ophthalmic Genetics</i> , 2016, 37, 276-280.	0.5	4
111	Identification of novel genomic imbalances in Saudi patients with congenital heart disease. <i>Molecular Cytogenetics</i> , 2018, 11, 9.	0.4	4
112	Exome Sequencing Reveals Novel <i>TTN</i> Variants in Saudi Patients with Congenital Titinopathies. <i>Genetic Testing and Molecular Biomarkers</i> , 2021, 25, 757-764.	0.3	4
113	When straight eyes won't move: phenotypic overlap of genetically distinct ocular motility disturbances. <i>Canadian Journal of Ophthalmology</i> , 2011, 46, 477-480.	0.4	3
114	Bilateral Congenital Entropion with Cutis Laxa. <i>Pediatric Dermatology</i> , 2014, 31, e82-e84.	0.5	3
115	Outcome of stroke in Saudi children. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S91-6.	0.5	3
116	Ophthalmologic Observations in a Patient with Partial Mosaic Trisomy 8. <i>Ophthalmic Genetics</i> , 2013, 34, 249-253.	0.5	2
117	First report of two successive deletions on chromosome 15q13 cytogenetic bands in a boy and girl: additional data to 15q13.3 syndrome with a report of high IQ patient. <i>Molecular Cytogenetics</i> , 2019, 12, 21.	0.4	2
118	Neural tube defects. Challenging, yet preventable. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2014, 35 Suppl 1, S3-4.	0.5	2
119	Stroke from cervicocephalic arterial dissection in Saudi children. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S103-7.	0.5	2
120	Cardiac diseases as a risk factor for stroke in Saudi children. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S61-8.	0.5	2
121	Moyamoya syndrome as a risk factor for stroke in Saudi children. Novel and usual associations. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S69-80.	0.5	2
122	Stroke from systemic vascular disorders in Saudi children. The devastating role of hypernatremic dehydration. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S97-102.	0.5	2
123	The challenge of diagnosing and successfully treating anti-NMDA receptor encephalitis in a toddler. <i>Sudanese Journal of Paediatrics</i> , 2021, 21, 76-81.	0.6	1
124	The odyssey of diagnosing genetic disorders in evolving health services. <i>Sudanese Journal of Paediatrics</i> , 2019, 19, 2-5.	0.6	1
125	Stroke due to mitochondrial disorders in Saudi children. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2006, 27 Suppl 1, S81-90.	0.5	1
126	Message from the guest editor. <i>Journal of King Abdulaziz University, Islamic Economics</i> , 2014, 35 Suppl 1, S2.	0.5	0

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127	Study project on stroke in Saudi children. Conclusions, recommendations and acknowledgements. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S108-10.	0.5	0
128	Hematologic risk factors for stroke in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S21-34.	0.5	0
129	Diagnostic approach and management strategy of childhood stroke. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S4-11.	0.5	0
130	Clinical, genetic, and functional characterization of the glycine receptor $\hat{1}^2$ -subunit A455P variant in a family affected by hyperekplexia syndrome. Journal of Biological Chemistry, 2022, , 102018.	1.6	0