Mustafa A. Salih

List of Publications by Year
in descending order

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130 papers	5,593 citations	94433 37 h-index	88630 70 g-index
132 all docs	132 docs citations	132 times ranked	9110 citing authors

#	Article	IF	Citations
1	Genetic diagnosis in Sudanese and Tunisian families with syndromic intellectual disability through exome sequencing. Annals of Human Genetics, 2022, 86, 181-194.	0.8	7
2	Clinical, genetic, and functional characterization of the glycine receptor Î²-subunit A455P variant in a family affected by hyperekplexia syndrome. Journal of Biological Chemistry, 2022, , 102018.	3.4	0
3	The challenge of diagnosing and successfully treating anti-NMDA receptor encephalitis in a toddler. Sudanese Journal of Paediatrics, 2021, 21, 76-81.	0.6	1
4	A heterozygous mutation in the CCDC88C gene likely causes early-onset pure hereditary spastic paraplegia: a case report. BMC Neurology, 2021, 21, 78.	1.8	8
5	An identicalâ€byâ€descent novel spliceâ€donor variant in PRUNE1 causes a neurodevelopmental syndrome with prominent dystonia in two consanguineous Sudanese families. Annals of Human Genetics, 2021, 85, 186-195.	0.8	5
6	Pathogenic Variants in ABHD16A Cause a Novel Psychomotor Developmental Disorder With Spastic Paraplegia. Frontiers in Neurology, 2021, 12, 720201.	2.4	5
7	Exome Sequencing Reveals Novel TTN Variants in Saudi Patients with Congenital Titinopathies. Genetic Testing and Molecular Biomarkers, 2021, 25, 757-764.	0.7	4
8	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.	7.7	38
9	Acute Necrotizing Encephalopathy of Childhood: A Multicenter Experience in Saudi Arabia. Frontiers in Pediatrics, 2020, 8, 526.	1.9	20
10	The natural history of infantile neuroaxonal dystrophy. Orphanet Journal of Rare Diseases, 2020, 15, 109.	2.7	11
11	Ancient founder mutation in RUBCN: a second unrelated family confirms Salih ataxia (SCAR15). BMC Neurology, 2020, 20, 207.	1.8	7
12	The morbid genome of ciliopathies: an update. Genetics in Medicine, 2020, 22, 1051-1060.	2.4	68
13	Rett Syndrome, a Neurodevelopmental Disorder, Whole-Transcriptome, and Mitochondrial Genome Multiomics Analyses Identify Novel Variations and Disease Pathways. OMICS A Journal of Integrative Biology, 2020, 24, 160-171.	2.0	18
14	Broad beans () and the potential to protect from COVID-19 coronavirus infection. Sudanese Journal of Paediatrics, 2020, 20, 10-12.	0.6	6
15	The Genetic Landscape of Epilepsy of Infancy with Migrating Focal Seizures. Annals of Neurology, 2019, 86, 821-831.	5.3	96
16	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	6.2	184
17	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. Molecular Cytogenetics, 2019, 12, 21.	0.9	2
18	Overlap of polymicrogyria, hydrocephalus, and Joubert syndrome in a family with novel truncating mutations in ADGRG1/GPR56 and KIAA0556. Neurogenetics, 2019, 20, 91-98.	1.4	17

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19	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	2.4	85
20	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
21	The odyssey of diagnosing genetic disorders in evolving health services. Sudanese Journal of Paediatrics, 2019, 19, 2-5.	0.6	1
22	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
23	NEW OBSERVATIONS REGARDING THE RETINOPATHY OF GENETICALLY CONFIRMED KEARNSâ€“SAYRE SYNDROME. Retinal Cases and Brief Reports, 2018, 12, 349-358.	0.6	8
24	Intra-familial phenotypic heterogeneity in a Sudanese family with DARS2-related leukoencephalopathy, brainstem and spinal cord involvement and lactate elevation: a case report. BMC Neurology, 2018, 18, 175.	1.8	13
25	Impact of PYROXD1 deficiency on cellular respiration and correlations with genetic analyses of limb-girdle muscular dystrophy in Saudi Arabia and Sudan. Physiological Genomics, 2018, 50, 929-939.	2.3	15
26	Identification of novel genomic imbalances in Saudi patients with congenital heart disease. Molecular Cytogenetics, 2018, 11, 9.	0.9	4
27	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
28	Prevalence of epilepsy in 74,949 school children in Khartoum State, Sudan. Paediatrics and International Child Health, 2017, 37, 188-192.	1.0	10
29	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Human Genetics, 2017, 136, 921-939.	3.8	209
30	A recessive ataxia diagnosis algorithm for the next generation sequencing era. Annals of Neurology, 2017, 82, 892-899.	5.3	27
31	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	122
32	The sensitivity of exome sequencing in identifying pathogenic mutations for LGMD in the United States. Journal of Human Genetics, 2017, 62, 243-252.	2.3	73
33	Gonadal mosaicism for ACTA1 gene masquerading as autosomal recessive nemaline myopathy. American Journal of Medical Genetics, Part A, 2016, 170, 2219-2221.	1.2	8
34	Novel copy number variants and major limb reduction malformation: Report of three cases. American Journal of Medical Genetics, Part A, 2016, 170, 1245-1250.	1.2	8
35	Characterizing the morbid genome of ciliopathies. Genome Biology, 2016, 17, 242.	8.8	118
36	Hyperekplexia, microcephaly and simplified gyral pattern caused by novel ASNS mutations, case report. BMC Neurology, 2016, 16, 105.	1.8	32

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37	Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. American Journal of Human Genetics, 2016, 98, 1249-1255.	6.2	40
38	A null mutation in TNIK defines a novel locus for intellectual disability. Human Genetics, 2016, 135, 773-778.	3.8	23
39	Duane retraction syndrome in a patient with Duchenne muscular dystrophy. Ophthalmic Genetics, 2016, 37, 276-280.	1.2	4
40	Pathogenic variants inKCTD7perturb neuronal K⁺fluxes and glutamine transport. Brain, 2016, 139, 3109-3120.	7.6	31
41	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. Genetics in Medicine, 2016, 18, 686-695.	2.4	55
42	Acute poisoning in a child following topical treatment of head lice (pediculosis capitis) with an organophosphate pesticide. Sudanese Journal of Paediatrics, 2016, 16, 63-6.	0.6	6
43	Idiopathic intracranial hypertension in children: Diagnostic and management approach. Sudanese Journal of Paediatrics, 2016, 16, 67-76.	0.6	14
44	Cerebral Iron Accumulation Is Not a Major Feature of <scp>FA</scp>2H/<scp>SPG</scp>35. Movement Disorders Clinical Practice, 2015, 2, 56-60.	1.5	9
45	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Reports, 2015, 10, 148-161.	6.4	375
46	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725.	2.4	19
47	Mutation in GM2A Leads to a Progressive Chorea-dementia Syndrome. Tremor and Other Hyperkinetic Movements, 2015, 5, 306.	2.0	6
48	Sturge-Weber syndrome: Continued vigilance is needed. Sudanese Journal of Paediatrics, 2015, 15, 63-70.	0.6	5
49	Expanding the clinical spectrum and allelic heterogeneity in van den Endeâ€“Gupta syndrome. Clinical Genetics, 2014, 85, 492-494.	2.0	8
50	NPHP4 mutation is linked to cerebelloâ€oculoâ€renal syndrome and male infertility. Clinical Genetics, 2014, 85, 371-375.	2.0	18
51	Bilateral Congenital Entropion with Cutis Laxa. Pediatric Dermatology, 2014, 31, e82-e84.	0.9	3
52	POMK mutations disrupt muscle development leading to a spectrum of neuromuscular presentations. Human Molecular Genetics, 2014, 23, 5781-5792.	2.9	72
53	The tumour suppressor gene WWOX is mutated in autosomal recessive cerebellar ataxia with epilepsy and mental retardation. Brain, 2014, 137, 411-419.	7.6	127
54	CC2D1A Regulates Human Intellectual and Social Function as well as NF-ÎºB Signaling Homeostasis. Cell Reports, 2014, 8, 647-655.	6.4	60

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55	C19orf12 mutation leads to a pallido-pyramidal syndrome. Gene, 2014, 537, 352-356.	2.2	28
56	Ophthalmic features ofPLA2G6-related paediatric neurodegeneration with brain iron accumulation. British Journal of Ophthalmology, 2014, 98, 889-893.	3.9	15
57	Neurologic Injury in Isolated Sulfite Oxidase Deficiency. Canadian Journal of Neurological Sciences, 2014, 41, 42-48.	0.5	13
58	Neural tube defects. Challenging, yet preventable. Journal of King Abdulaziz University, Islamic Economics, 2014, 35 Suppl 1, S3-4.	1.1	2
59	Message from the guest editor. Journal of King Abdulaziz University, Islamic Economics, 2014, 35 Suppl 1, S2.	1.1	0
60	Classification, clinical features, and genetics of neural tube defects. Journal of King Abdulaziz University, Islamic Economics, 2014, 35 Suppl 1, S5-S14.	1.1	15
61	Genetic, chromosomal, and syndromic causes of neural tube defects. Journal of King Abdulaziz University, Islamic Economics, 2014, 35 Suppl 1, S49-56.	1.1	8
62	Epidemiology, prenatal management, and prevention of neural tube defects. Journal of King Abdulaziz University, Islamic Economics, 2014, 35 Suppl 1, S15-28.	1.1	22
63	The Salih Ataxia Mutation Impairs Rubicon Endosomal Localization. Cerebellum, 2013, 12, 835-840.	2.5	18
64	Sleep-disordered breathing in children with craniosynostosis. Sleep and Breathing, 2013, 17, 389-393.	1.7	21
65	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
66	Ophthalmologic Observations in a Patient with Partial Mosaic Trisomy 8. Ophthalmic Genetics, 2013, 34, 249-253.	1.2	2
67	Mutations in B3GALNT2 Cause Congenital Muscular Dystrophy and Hypoglycosylation of Î±-Dystroglycan. American Journal of Human Genetics, 2013, 92, 354-365.	6.2	172
68	Mutation inADAT3, encoding adenosine deaminase acting on transfer RNA, causes intellectual disability and strabismus. Journal of Medical Genetics, 2013, 50, 425-430.	3.2	91
69	SLC25A22 is a novel gene for migrating partial seizures in infancy. Annals of Neurology, 2013, 74, 873-882.	5.3	102
70	Congenital myasthenic syndromes due to mutations inALG2andALG14. Brain, 2013, 136, 944-956.	7.6	117
71	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
72	Mutation in PHC1 implicates chromatin remodeling in primary microcephaly pathogenesis. Human Molecular Genetics, 2013, 22, 2200-2213.	2.9	81

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73	A newly recognized autosomal recessive syndrome affecting neurologic function and vision. American Journal of Medical Genetics, Part A, 2013, 161, 1207-1213.	1.2	9
74	Preimplantation Genetic Diagnosis in Isolated Sulfite Oxidase Deficiency. Canadian Journal of Neurological Sciences, 2013, 40, 109-112.	0.5	10
75	New Findings in a Global Approach to Dissect the Whole Phenotype of PLA2G6 Gene Mutations. PLoS ONE, 2013, 8, e76831.	2.5	42
76	Genomic analysis of mitochondrial diseases in a consanguineous population reveals novel candidate disease genes. Journal of Medical Genetics, 2012, 49, 234-241.	3.2	164
77	A novel syndrome of lethal familial hyperekplexia associated with brain malformation. BMC Neurology, 2012, 12, 125.	1.8	9
78	Exome Sequencing and Functional Validation in Zebrafish Identify GTDC2 Mutations as a Cause of Walker-Warburg Syndrome. American Journal of Human Genetics, 2012, 91, 541-547.	6.2	167
79	Molecular characterization of Joubert syndrome in Saudi Arabia. Human Mutation, 2012, 33, 1423-1428.	2.5	56
80	Congenital disorder of glycosylation IIa: The trouble with diagnosing a dysmorphic inborn error of metabolism. American Journal of Medical Genetics, Part A, 2012, 158A, 245-246.	1.2	12
81	When straight eyes won't move: phenotypic overlap of genetically distinct ocular motility disturbances. Canadian Journal of Ophthalmology, 2011, 46, 477-480.	0.7	3
82	Molecular and neurological characterizations of three Saudi families with lipoid proteinosis. BMC Medical Genetics, 2011, 12, 31.	2.1	19
83	Ritscherâ€“Schinzel (cranioâ€cerebelloâ€cardiac, 3C) syndrome: Report of four new cases with renal involvement. American Journal of Medical Genetics, Part A, 2011, 155, 1393-1397.	1.2	12
84	Congenital Microcephaly with a Simplified Gyral Pattern: Associated Findings and Their Significance. American Journal of Neuroradiology, 2011, 32, 1123-1129.	2.4	62
85	Congenital Myasthenic Syndrome Due to Homozygous CHRNE Mutations: Report of Patients in Arabia. Journal of Neuro-Ophthalmology, 2011, 31, 42-47.	0.8	12
86	The neurology of carbonic anhydrase type II deficiency syndrome. Brain, 2011, 134, 3502-3515.	7.6	37
87	Efficient identification of novel mutations in patients with limb girdle muscular dystrophy. Neurogenetics, 2010, 11, 449-455.	1.4	15
88	Megalencephalic leukoencephalopathy with cysts without MLC1 defect. Annals of Neurology, 2010, 67, 834-837.	5.3	52
89	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
90	Interleukin 10 Gene Polymorphisms and Development of Post Kala-Azar Dermal Leishmaniasis in a Selected Sudanese Population. Public Health Genomics, 2010, 13, 362-367.	1.0	24

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91	Rundataxin, a novel protein with RUN and diacylglycerol binding domains, is mutant in a new recessive ataxia. Brain, 2010, 133, 2439-2447.	7.6	46
92	Optic disk and white matter abnormalities in a patient with ade novo18p partial monosomy. Ophthalmic Genetics, 2010, 31, 147-154.	1.2	4
93	Ophthalmologic abnormalities in a de novo terminal 6q deletion. Ophthalmic Genetics, 2010, 31, 1-11.	1.2	17
94	A new complex homozygous large rearrangement of the PINK1 gene in a Sudanese family with early onset Parkinsonâ€™s disease. Neurogenetics, 2009, 10, 265-270.	1.4	25
95	Mutations of the FHL1 Gene Cause Emery-Dreifuss Muscular Dystrophy. American Journal of Human Genetics, 2009, 85, 338-353.	6.2	208
96	The clinical spectrum of homozygous HOXA1 mutations. American Journal of Medical Genetics, Part A, 2008, 146A, 1235-1240.	1.2	101
97	Ophthalmic features of ataxia telangiectasiaâ€“like disorder. Journal of AAPOS, 2008, 12, 186-189.	0.3	35
98	Ophthalmic Features of Joubert Syndrome. Ophthalmology, 2008, 115, 2286-2289.	5.2	48
99	A substance in broad beans (Vicia faba) is protective against experimentally induced convulsions in mice. Epilepsy and Behavior, 2008, 12, 25-29.	1.7	33
100	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. Brain, 2008, 131, 747-759.	7.6	134
101	Clinical characterization of the HOXA1 syndrome BSAS variant. Neurology, 2007, 69, 1245-1253.	1.1	83
102	Spinocerebellar ataxia with axonal neuropathy: consequence of a Tdp1 recessive neomorphic mutation?. EMBO Journal, 2007, 26, 4732-4743.	7.8	129
103	Brain Stem and Cerebellar Findings in Joubert Syndrome. Journal of Computer Assisted Tomography, 2006, 30, 116-121.	0.9	35
104	Stroke from cervicocephalic arterial dissection in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S103-7.	1.1	2
105	Study project on stroke in Saudi children. Conclusions, recommendations and acknowledgements. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S108-10.	1.1	0
106	Stroke in Saudi children. Epidemiology, clinical features and risk factors. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S12-20.	1.1	11
107	Hematologic risk factors for stroke in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S21-34.	1.1	0
108	Perinatal stroke in Saudi children. Clinical features and risk factors. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S35-40.	1.1	5

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109	Diagnostic approach and management strategy of childhood stroke. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S4-11.	1.1	0
110	Infectious and inflammatory disorders of the circulatory system as risk factors for stroke in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S41-52.	1.1	8
111	Congenital and genetic cerebrovascular anomalies as risk factors for stroke in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S53-60.	1.1	6
112	Cardiac diseases as a risk factor for stroke in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S61-8.	1.1	2
113	Moyamoya syndrome as a risk factor for stroke in Saudi children. Novel and usual associations. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S69-80.	1.1	2
114	Stroke due to mitochondrial disorders in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S81-90.	1.1	1
115	Outcome of stroke in Saudi children. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S91-6.	1.1	3
116	Stroke from systemic vascular disorders in Saudi children. The devastating role of hypernatremic dehydration. Journal of King Abdulaziz University, Islamic Economics, 2006, 27 Suppl 1, S97-102.	1.1	2
117	Embryonal rhabdomyosarcoma and chromosomal breakage in a newborn infant with possible Dubowitz syndrome. , 2000, 92, 107-110.		19
118	Charcot-Marie-Tooth type 4B is caused by mutations in the gene encoding myotubularin-related protein-2. Nature Genetics, 2000, 25, 17-19.	21.4	462
119	Genetic Refinement and Physical Mapping of the CMT4B Gene on Chromosome 11q22. Genomics, 2000, 63, 271-278.	2.9	18
120	Autosomal recessive hereditary neuropathy with focally folded myelin sheaths and linked to chromosome 11q23: a distinct and homogeneous entity. Neuromuscular Disorders, 2000, 10, 10-15.	0.6	16
121	Sotos syndrome (cerebral gigantism): a clinical and radiological study of 14 cases from Saudi Arabia. Annals of Tropical Paediatrics, 1999, 19, 197-203.	1.0	17
122	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
123	Moyamoya syndrome with unusual angiographic findings and protein C deficiency: Review of the literature. Journal of the Neurological Sciences, 1998, 159, 11-16.	0.6	15
124	A Novel Form of Familial Congenital Muscular Dystrophy in Two Adolescents. Neuropediatrics, 1998, 29, 289-293.	0.6	25
125	Hemiconvulsion-hemiplegia-epilepsy syndrome. Child's Nervous System, 1997, 13, 257-263.	1.1	43
126	Muscular dystrophy associated with ?-dystroglycan deficiency. Annals of Neurology, 1996, 40, 925-928.	5.3	17

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127	Lethal Congenital Muscular Dystrophy in Two Sibs with Arthrogryposis Multiplex: New Entity or Variant of Cobblestone Lissencephaly Syndrome?. Neuropediatrics, 1996, 27, 305-310.	0.6	21
128	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
129	Childhood muscular dystrophy: an African review. Annals of Tropical Paediatrics, 1985, 5, 167-173.	1.0	13
130	Respiratory insufficiency in a severe autosomal recessive form of musclar dystrophy. Annals of Tropical Paediatrics, 1984, 4, 45-48.	1.0	5