Fahad A Bashiri

List of Publications by Year
in descending order

54 papers	1,942 citations	516215 16 h-index	276539 41 g-index
60 all docs	60 docs citations	60 times ranked	4023 citing authors

#	Article	IF	Citations
1	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Reports, 2015, 10, 148-161.	2.9	375
2	The landscape of genetic diseases in Saudi Arabia based on the first 1000 diagnostic panels and exomes. Human Genetics, 2017, 136, 921-939.	1.8	209
3	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. Molecular Psychiatry, 2017, 22, 615-624.	4.1	187
4	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	2.6	184
5	Biotin-responsive basal ganglia disease should be renamed biotin-thiamine-responsive basal ganglia disease: a retrospective review of the clinical, radiological and molecular findings of 18 new cases. Orphanet Journal of Rare Diseases, 2013, 8, 83.	1.2	123
6	Parental Attitudes and Hesitancy About COVID-19 vs. Routine Childhood Vaccinations: A National Survey. Frontiers in Public Health, 2021, 9, 752323.	1.3	106
7	SLC25A22 is a novel gene for migrating partial seizures in infancy. Annals of Neurology, 2013, 74, 873-882.	2.8	102
8	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582.	0.3	94
9	Genomic and phenotypic delineation of congenital microcephaly. Genetics in Medicine, 2019, 21, 545-552.	1.1	85
10	JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency. New England Journal of Medicine, 2020, 382, 256-265.	13.9	69
11	Mutations in NKX6-2 Cause Progressive Spastic Ataxia and Hypomyelination. American Journal of Human Genetics, 2017, 100, 969-977.	2.6	38
12	Confirming the recessive inheritance of <scp>SCN1B</scp> mutations in developmental epileptic encephalopathy. Clinical Genetics, 2017, 92, 327-331.	1.0	32
13	Satisfaction and perceived usefulness with newly-implemented Electronic Health Records System among pediatricians at a university hospital. Computer Methods and Programs in Biomedicine, 2019, 169, 51-57.	2.6	26
14	Phenotypic and molecular spectrum of pyridoxamineâ€5â€²â€phosphate oxidase deficiency: A scoping review of 87 cases of pyridoxamineâ€5â€²â€phosphate oxidase deficiency. Clinical Genetics, 2021, 99, 99-110.	1.0	25
15	Acute Necrotizing Encephalopathy of Childhood: A Multicenter Experience in Saudi Arabia. Frontiers in Pediatrics, 2020, 8, 526.	0.9	20
16	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725.	1.1	19
17	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. Frontiers in Pediatrics, 2021, 9, 633385.	0.9	15
18	Vitamin D supplementation to prevent vitamin D deficiency for children with epilepsy. Medicine (United) Tj ETQq0 0 0 rgBT /Overlock 10	0.4	14

#	Article	IF	Citations
19	Idiopathic intracranial hypertension in children: Diagnostic and management approach. Sudanese Journal of Paediatrics, 2016, 16, 67-76.	0.6	14
20	ARL6IP6, a susceptibility locus for ischemic stroke, is mutated in a patient with syndromic Cutis Marmorata Telangiectatica Congenita. Human Genetics, 2015, 134, 815-822.	1.8	13
21	Prevalence of autism spectrum disorder among Saudi children between 2 and 4 years old in Riyadh. Asian Journal of Psychiatry, 2022, 71, 103054.	0.9	13
22	Management of convulsive status epilepticus in children: an adapted clinical practice guideline for pediatricians in Saudi Arabia. Journal of King Abdulaziz University, Islamic Economics, 2017, 22, 146-155.	0.5	12
23	Expanding the phenotype and the genotype of Stromme syndrome: A novel variant of the CENPF gene and literature review. European Journal of Medical Genetics, 2020, 63, 103844.	0.7	12
24	Medical Facultyâ€™s and Studentsâ€™ Perceptions toward Pediatric Electronic OSCE during the COVID-19 Pandemic in Saudi Arabia. Healthcare (Switzerland), 2021, 9, 950.	1.0	11
25	Appraisal of clinical practice guidelines for the management of attention deficit hyperactivity disorder (ADHD) using the AGREE II Instrument: A systematic review. PLoS ONE, 2019, 14, e0219239.	1.1	9
26	Remote interviews for medical residency selection during the initial COVID-19 crisis: a national survey. BMC Medical Education, 2021, 21, 462.	1.0	9
27	Consensus Statement on the Management of Duchenne Muscular Dystrophy in Saudi Arabia During the Coronavirus Disease 2019 Pandemic. Frontiers in Pediatrics, 2021, 9, 629549.	0.9	7
28	Hypokalemic periodic paralysis due to CACNA1S gene mutation. Journal of King Abdulaziz University, Islamic Economics, 2019, 24, 225-230.	0.5	7
29	Auto-immune anti-N-methyl-D-aspartate receptor (anti-NMDAR) encephalitis: three case reports. Paediatrics and International Child Health, 2017, 37, 222-226.	0.3	6
30	GWAS signals revisited using human knockouts. Genetics in Medicine, 2018, 20, 64-68.	1.1	6
31	Adapting evidence-based clinical practice guidelines for people with attention deficit hyperactivity disorder in Saudi Arabia: process and outputs of a national initiative. Child and Adolescent Psychiatry and Mental Health, 2021, 15, 6.	1.2	6
32	Proxy-Reported Quality of Life and Access to Nusinersen Among Patients with Spinal Muscular Atrophy in Saudi Arabia. Patient Preference and Adherence, 2021, Volume 15, 729-739.	0.8	6
33	Childhood epilepsies: What should a pediatrician know?. Journal of King Abdulaziz University, Islamic Economics, 2017, 22, 14-19.	0.5	6
34	AGREEing on Clinical Practice Guidelines for Autism Spectrum Disorders in Children: A Systematic Review and Quality Assessment. Children, 2022, 9, 1050.	0.6	6
35	Assessment of physiciansâ€™ knowledge and attitudes in the management of febrile seizures. Journal of King Abdulaziz University, Islamic Economics, 2018, 23, 314-319.	0.5	5
36	Sturge-Weber syndrome: Continued vigilance is needed. Sudanese Journal of Paediatrics, 2015, 15, 63-70.	0.6	5

#	Article	IF	Citations
37	Faculty Membersâ€™ Perspective on Virtual Interviews for Medical Residency Matching during the COVID-19 Crisis: A National Survey. Healthcare (Switzerland), 2022, 10, 16.	1.0	4
38	A Case of Neonatal Epileptic Encephalopathy due to SCN2A Mutation Responsive to a Ketogenic Diet. Journal of Pediatric Epilepsy, 2018, 07, 148-151.	0.1	3
39	Effect of new modalities of treatment on physiciansâ€™ management plan for patients with spinal muscular atrophy. Journal of King Abdulaziz University, Islamic Economics, 2019, 24, 16-21.	0.5	3
40	A consensus statement on spinal muscular atrophy management in Saudi Arabia in the context of COVID-19. Journal of King Abdulaziz University, Islamic Economics, 2020, 25, 230-237.	0.5	3
41	Childhood absence epilepsy. International Journal of Pediatrics and Adolescent Medicine, 2022, 9, 131-135.	0.5	3
42	Mycoplasma pneumoniae Infection. Global Pediatric Health, 2015, 2, 2333794X1559276.	0.3	2
43	Pyridox(am)ine 5â€²-Phosphate Oxidase Deficiency: Severe Prenatal Presentation with Hypoxic Ischemic Encephalopathy. Journal of Pediatric Epilepsy, 2019, 08, 049-055.	0.1	2
44	Effect of lumbar puncture educational video on parental knowledge and self-reported intended practice. International Journal of Pediatrics and Adolescent Medicine, 2021, 8, 112-116.	0.5	2
45	2020 Update to Spinal Muscular Atrophy Management in Saudi Arabia. Frontiers in Pediatrics, 2021, 9, 684134.	0.9	2
46	Respiratory support attitudes among pediatric intensive care staff for spinal muscular atrophy patients in Saudi Arabia. Journal of King Abdulaziz University, Islamic Economics, 2018, 23, 62-65.	0.5	2
47	Drowning in the desert: Family denial of brain death. Sudanese Journal of Paediatrics, 2018, 18, 48-52.	0.6	2
48	Clinical spectrum of neurofibromatosis type 1 among children in a tertiary care center. Journal of King Abdulaziz University, Islamic Economics, 2020, 25, 375-379.	0.5	2
49	Physicians' Understanding of Nutritional Factors Determining Brain Development and Cognition in the Middle East and Africa. Pediatric Gastroenterology, Hepatology and Nutrition, 2019, 22, 536.	0.4	1
50	Pediatric intracranial hypertension. Journal of King Abdulaziz University, Islamic Economics, 2019, 24, 257-263.	0.5	1
51	Syncopal attacks in children: Is it cardiac or epilepsy related?. Autonomic Neuroscience: Basic and Clinical, 2021, 231, 102771.	1.4	0
52	Predicting future handedness and hemispheric dominance during infancy by analyzing sleep spindles. Journal of King Abdulaziz University, Islamic Economics, 2020, 25, 188-192.	0.5	0
53	Nusinersen in spinal muscular atrophy: Respiratory outcomes at tertiary care centers. Journal of Nature and Science of Medicine, 2020, .	0.1	0
54	Clinical spectrum of neurofibromatosis type 1 among children in a tertiary care center. Neurosciences, 2020, 25, 375-379.	0.1	0