Majid Alfadhel

List of Publications by Year
in descending order

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171	4,947	136950	128289
papers	citations	32 h-index	60 g-index

178	178	178	8395
all docs	docs citations	times ranked	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.	6.4	375
2	Hypomorphic mutations in syndromic encephalocele genes are associated with Bardet-Biedl syndrome. Nature Genetics, 2008, 40, 443-448.	21.4	367
3	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	2.8	291
4	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254
5	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
6	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.	2.7	123
7	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	3.8	122
8	Whole-genome sequencing offers additional but limited clinical utility compared with reanalysis of whole-exome sequencing. Genetics in Medicine, 2018, 20, 1328-1333.	2.4	118
9	ISCA2mutation causes infantile neurodegenerative mitochondrial disorder. Journal of Medical Genetics, 2015, 52, 186-194.	3.2	90
10	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540.	3.8	89
11	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	7.8	86
12	Expanded Newborn Screening Program in Saudi Arabia: Incidence of screened disorders. Journal of Paediatrics and Child Health, 2017, 53, 585-591.	0.8	83
13	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	2.4	81
14	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
15	Carnitine Inborn Errors of Metabolism. Molecules, 2019, 24, 3251.	3.8	70
16	A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. Molecular Genetics and Metabolism, 2017, 121, 91-95.	1.1	68
17	Clinical Genetics of Polydactyly: An Updated Review. Frontiers in Genetics, 2018, 9, 447.	2.3	66
18	Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. European Journal of Human Genetics, 2021, 29, 141-153.	2.8	66

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19	Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Annals of Neurology, 2017, 82, 317-330.	5.3	65
20	Thirteen year retrospective review of the spectrum of inborn errors of metabolism presenting in a tertiary center in Saudi Arabia. Orphanet Journal of Rare Diseases, 2016, 11, 126.	2.7	61
21	KCNT1â€related epilepsy: An international multicenter cohort of 27 pediatric cases. Epilepsia, 2020, 61, 679-692.	5.1	50
22	Asparagine Synthetase Deficiency: New Inborn Errors of Metabolism. JIMD Reports, 2015, 22, 11-16.	1.5	46
23	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	2.4	46
24	MPV17-related mitochondrial DNA maintenance defect: New cases and review of clinical, biochemical, and molecular aspects. Human Mutation, 2018, 39, 461-470.	2.5	45
25	Clinical and Molecular Characteristics of Mitochondrial DNA Depletion Syndrome Associated with Neonatal Cholestasis and Liver Failure. Journal of Pediatrics, 2014, 164, 553-559.e2.	1.8	44
26	Treatment of biotin-responsive basal ganglia disease: Open comparative study between the combination of biotin plus thiamine versus thiamine alone. European Journal of Paediatric Neurology, 2015, 19, 547-552.	1.6	43
27	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
28	Phenotypic and Molecular Spectrum of Aicardi-GoutiÃ¨res Syndrome: A Study of 24 Patients. Pediatric Neurology, 2018, 78, 35-40.	2.1	40
29	GOLGA2, encoding a master regulator of golgi apparatus, is mutated in a patient with a neuromuscular disorder. Human Genetics, 2016, 135, 245-251.	3.8	38
30	Targeted SLC19A3 gene sequencing of 3000 Saudi newborn: a pilot study toward newborn screening. Annals of Clinical and Translational Neurology, 2019, 6, 2097-2103.	3.7	38
31	Deficiency of ADA2 mimicking autoimmune lymphoproliferative syndrome in the absence of livedo reticularis and vasculitis. Pediatric Blood and Cancer, 2018, 65, e26912.	1.5	37
32	Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. Human Molecular Genetics, 2018, 27, 3029-3045.	2.9	37
33	Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. Journal of Inherited Metabolic Disease, 2013, 36, 997-1004.	3.6	36
34	Mutation in SLC6A9 encoding a glycine transporter causes a novel form of non-ketotic hyperglycinemia in humans. Human Genetics, 2016, 135, 1263-1268.	3.8	35
35	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	6.2	35
36	The landscape of early infantile epileptic encephalopathy in a consanguineous population. Seizure: the Journal of the British Epilepsy Association, 2019, 69, 154-172.	2.0	34

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37	Guidelines for acute management of hyperammonemia in the Middle East region. Therapeutics and Clinical Risk Management, 2016, 12, 479.	2.0	32
38	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
39	Genetic Disorders Associated with Metal Metabolism. Cells, 2019, 8, 1598.	4.1	31
40	Congenital disorders of glycosylation: The Saudi experience. American Journal of Medical Genetics, Part A, 2017, 173, 2614-2621.	1.2	31
41	SLC19A3 Gene Defects Sorting the Phenotype and Acronyms: Review. Neuropediatrics, 2018, 49, 083-092.	0.6	30
42	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
43	Utilizing Whole-Exome Sequencing to Characterize the Phenotypic Variability of Sickle Cell Disease. Genetic Testing and Molecular Biomarkers, 2018, 22, 561-567.	0.7	29
44	Risk of Neuropsychiatric Adverse Effects of Lipid-Lowering Drugs: A Mendelian Randomization Study. International Journal of Neuropsychopharmacology, 2018, 21, 1067-1075.	2.1	29
45	Next Generation Sequencing Based Non-invasive Prenatal Testing (NIPT): First Report From Saudi Arabia. Frontiers in Genetics, 2021, 12, 630787.	2.3	29
46	Drug treatment of inborn errors of metabolism: a systematic review. Archives of Disease in Childhood, 2013, 98, 454-461.	1.9	28
47	Next-Generation Sequencing-Based Pre-Implantation Genetic Testing for Aneuploidy (PGT-A): First Report from Saudi Arabia. Genes, 2021, 12, 461.	2.4	28
48	Enzyme replacement therapy for Fabry disease: some answers but more questions. Therapeutics and Clinical Risk Management, 2011, 7, 69.	2.0	27
49	Clinical, biochemical, and molecular overview of transaldolase deficiency and evaluation of the endocrine function: Update of 34 patients. Journal of Inherited Metabolic Disease, 2019, 42, 147-158.	3.6	26
50	Biallelic variants in four genes underlying recessive osteogenesis imperfecta. European Journal of Medical Genetics, 2020, 63, 103954.	1.3	26
51	Worsening of Seizures After Asparagine Supplementation in a Child with Asparagine Synthetase Deficiency. Pediatric Neurology, 2016, 58, 98-100.	2.1	24
52	<scp>EMC10</scp> homozygous variant identified in a family with global developmental delay, mild intellectual disability, and speech delay. Clinical Genetics, 2020, 98, 555-561.	2.0	24
53	Broadening the phenotypic spectrum of pathogenic LARP7 variants: two cases with intellectual disability, variable growth retardation and distinct facial features. Journal of Human Genetics, 2016, 61, 229-233.	2.3	23
54	Secondary Hemophagocytic Syndrome Associated with COG6 Gene Defect: Report and Review. JIMD Reports, 2018, 42, 105-111.	1.5	23

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55	Novel homozygous mutation in the WWOX gene causes seizures and global developmental delay: Report and review. Translational Neuroscience, 2018, 9, 203-208.	1.4	23
56	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
57	Hereditary Neurometabolic Causes of Infantile Spasms in 80 Children Presenting to a Tertiary Care Center. Pediatric Neurology, 2014, 51, 390-397.	2.1	21
58	Long-term Outcome of 4 Patients With Transcobalamin Deficiency Caused by 2 Novel TCN2 Mutations. Journal of Pediatric Hematology/Oncology, 2017, 39, e430-e436.	0.6	21
59	Further delineation of the phenotypic spectrum ofÂ ISCA2 defect: A report of ten new cases. European Journal of Paediatric Neurology, 2018, 22, 46-55.	1.6	21
60	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
61	Mutated RAP1GDS1 causes a new syndrome of dysmorphic feature, intellectual disability & speech delay. Annals of Clinical and Translational Neurology, 2020, 7, 956-964.	3.7	21
62	Further Delineation of the Clinical Phenotype of Cerebellar Ataxia, Mental Retardation, and Disequilibrium Syndrome Type 4. Journal of Central Nervous System Disease, 2018, 10, 117957351875968.	1.9	19
63	Assessment of methylcitrate and methylcitrate to citrate ratio in dried blood spots as biomarkers for inborn errors of propionate metabolism. Scientific Reports, 2019, 9, 12366.	3.3	19
64	SGCD Homozygous Nonsense Mutation (p.Arg97âˆ—) Causing Limb-Girdle Muscular Dystrophy Type 2F (LGMD2F) in a Consanguineous Family, a Case Report. Frontiers in Genetics, 2019, 9, 727.	2.3	19
65	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
66	Biallelic variants in COPB1 cause a novel, severe intellectual disability syndrome with cataracts and variable microcephaly. Genome Medicine, 2021, 13, 34.	8.2	18
67	Delta Like-1 Gene Mutation: A Novel Cause of Congenital Vertebral Malformation. Frontiers in Genetics, 2019, 10, 534.	2.3	17
68	Multiple Mitochondrial Dysfunctions Syndrome 4 Due to ISCA2 Gene Defects: A Review. Child Neurology Open, 2019, 6, 2329048X1984737.	1.1	16
69	Homozygous truncating NEK10 mutation, associated with primary ciliary dyskinesia: a case report. BMC Pulmonary Medicine, 2020, 20, 141.	2.0	16
70	Biallelic variant in DACH1, encoding Dachshund Homolog 1, defines a novel candidate locus for recessive postaxial polydactyly type A. Genomics, 2021, 113, 2495-2502.	2.9	16
71	Clinical, Biochemical, and Molecular Features in 37 Saudi Patients with Very Long Chain Acyl CoA Dehydrogenase Deficiency. JIMD Reports, 2017, 40, 47-53.	1.5	15
72	Biallelic Missense Mutation in the ECEL1 Underlies Distal Arthrogryposis Type 5 (DA5D). Frontiers in Pediatrics, 2019, 7, 343.	1.9	15

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73	Exome sequencing revealed a novel lossâ€ofâ€function variant in the GLI3 transcriptional activator 2 domain underlies nonsyndromic postaxial polydactyly. Molecular Genetics & Genomic Medicine, 2019, 7, e00627.	1.2	15
74	A Missense Mutation in the UGDH Gene Is Associated With Developmental Delay and Axial Hypotonia. Frontiers in Pediatrics, 2020, 8, 71.	1.9	15
75	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. Frontiers in Pediatrics, 2021, 9, 633385.	1.9	15
76	Tetrasomy 18p: case report and review of literature. The Application of Clinical Genetics, 2018, Volume 11, 9-14.	3.0	14
77	MYT1L mutation in a patient causes intellectual disability and early onset of obesity: a case report and review of the literature. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 409-413.	0.9	14
78	Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. Human Genetics, 2021, 140, 579-592.	3.8	14
79	Mitochondrial iron-sulfur cluster biogenesis from molecular understanding to clinical disease. Journal of King Abdulaziz University, Islamic Economics, 2017, 22, 4-13.	1.1	14
80	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 880-893.	2.4	14
81	Early Infantile Leigh-like SLC19A3 Gene Defects Have a Poor Prognosis: Report and Review. Journal of Central Nervous System Disease, 2017, 9, 117957351773752.	1.9	13
82	Pathogenic STX3 variants affecting the retinal and intestinal transcripts cause an early-onset severe retinal dystrophy in microvillus inclusion disease subjects. Human Genetics, 2021, 140, 1143-1156.	3.8	13
83	The Association between Obesity and Chronic Conditions: Results from a Large Electronic Health Records System in Saudi Arabia. International Journal of Environmental Research and Public Health, 2021, 18, 12361.	2.6	13
84	Precocious Puberty in Two Girls With PEHO Syndrome: A Clinical Feature Not Previously Described. Journal of Child Neurology, 2011, 26, 851-857.	1.4	12
85	Lossâ€ofâ€function mutation in RUSC2 causes intellectual disability and secondary microcephaly. Developmental Medicine and Child Neurology, 2016, 58, 1317-1322.	2.1	12
86	ALG13â€CDG in a male with seizures, normal cognitive development, and normal transferrin isoelectric focusing. American Journal of Medical Genetics, Part A, 2017, 173, 2772-2775.	1.2	12
87	Homozygous missense variant in the TTN gene causing autosomal recessive limb-girdle muscular dystrophy type 10. BMC Medical Genetics, 2019, 20, 166.	2.1	12
88	6-Pyruvoyltetrahydropterin Synthase Deficiency: Review and Report of 28 Arab Subjects. Pediatric Neurology, 2019, 96, 40-47.	2.1	12
89	Spectrum of mutations underlying Propionic acidemia and further insight into a genotype-phenotype correlation for the common mutation in Saudi Arabia. Molecular Genetics and Metabolism Reports, 2019, 18, 22-29.	1.1	12
90	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	2.0	12

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91	Adenosine Kinase Deficiency: Report and Review. Neuropediatrics, 2019, 50, 046-050.	0.6	12
92	Mutated VWA8 Is Associated With Developmental Delay, Microcephaly, and Scoliosis and Plays a Novel Role in Early Development and Skeletal Morphogenesis in Zebrafish. Frontiers in Cell and Developmental Biology, 2021, 9, 736960.	3.7	12
93	Long-term effectiveness of carglumic acid in patients with propionic acidemia (PA) and methylmalonic acidemia (MMA): a randomized clinical trial. Orphanet Journal of Rare Diseases, 2021, 16, 422.	2.7	12
94	Extreme intrafamilial variability of Saudi brothers with primary hyperoxaluria type 1. Therapeutics and Clinical Risk Management, 2012, 8, 373.	2.0	11
95	Early-Onset Parkinsonism: Case Report and Review of the Literature. Pediatric Neurology, 2017, 67, 102-106.e1.	2.1	10
96	Rare genetic variant in the CFB gene presenting as atypical hemolytic uremic syndrome and immune complex diffuse membranoproliferative glomerulonephritis, with crescents, successfully treated with eculizumab. Pediatric Nephrology, 2017, 32, 885-891.	1.7	10
97	PRUNE Syndrome Is a New Neurodevelopmental Disorder. Child Neurology Open, 2018, 5, 2329048X1775223.	1.1	10
98	Expanding the clinical and genetic spectra of NKX6â€2â€related disorder. Clinical Genetics, 2018, 93, 1087-1092.	2.0	10
99	Biallelic SCN2A Gene Mutation Causing Early Infantile Epileptic Encephalopathy: Case Report and Review. Journal of Central Nervous System Disease, 2019, 11, 117957351984993.	1.9	10
100	Glycine Transporter 1 Encephalopathy From Biochemical Pathway to Clinical Disease: Review. Child Neurology Open, 2019, 6, 2329048X1983148.	1.1	10
101	What is the right sequencing approach? Solo VS extended family analysis in consanguineous populations. BMC Medical Genomics, 2020, 13, 103.	1.5	10
102	Aromatic Amino Acid Decarboxylase Deficiency Not Responding to Pyridoxine and Bromocriptine Therapy: Case Report and Review of Response to Treatment. Journal of Central Nervous System Disease, 2014, 6, JCNSD.S12938.	1.9	9
103	Unravelling 5-oxoprolinuria (pyroglutamic aciduria) due to bi-allelic OPLAH mutations: 20 new mutations in 14 families. Molecular Genetics and Metabolism, 2016, 119, 44-49.	1.1	9
104	A new association between CDK5RAP2 microcephaly and congenital cataracts. Annals of Human Genetics, 2018, 82, 165-170.	0.8	9
105	Inborn errors of metabolism associated with hyperglycaemic ketoacidosis and diabetes mellitus: narrative review. Sudanese Journal of Paediatrics, 2018, 18, 10-23.	0.6	9
106	Methionine adenosyltransferase I/III deficiency: beyond the central nervous system manifestations. Therapeutics and Clinical Risk Management, 2018, Volume 14, 225-229.	2.0	9
107	Evaluation of long-term effectiveness of the use of carglumic acid in patients with propionic acidemia (PA) or methylmalonic acidemia (MMA): study protocol for a randomized controlled trial. BMC Pediatrics, 2019, 19, 195.	1.7	9
108	A Novel Homozygous Non-sense Mutation in the Catalytic Domain of MTHFR Causes Severe 5,10-Methylenetetrahydrofolate Reductase Deficiency. Frontiers in Neurology, 2019, 10, 411.	2.4	9

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109	Diversity of Phenotype and Genetic Etiology of 23 Cystinuria Saudi Patients: A Retrospective Study. Frontiers in Pediatrics, 2020, 8, 569389.	1.9	9
110	Pancytopenia, Recurrent Infection, Poor Wound Healing, Heterotopia of the Brain Probably Associated with A Candidate Novel de Novo CDC42 Gene Defect: Expanding the Molecular and Phenotypic Spectrum. Genes, 2021, 12, 294.	2.4	9
111	Short stature with low insulinâ€like growth factor 1 availability due to pregnancyâ€associated plasma protein <scp>A2</scp> deficiency in a Saudi family. Clinical Genetics, 2021, 100, 601-606.	2.0	9
112	Common disease-associated gene variants in a Saudi Arabian population. Annals of Saudi Medicine, 2022, 42, 29-35.	1.1	9
113	The SORCS3 gene is mutated in brothers with infantile spasms and intellectual disability. Discovery Medicine, 2018, 26, 147-153.	0.5	9
114	Variability of Phenotype in Two Sisters with Pyridoxine Dependent Epilepsy. Canadian Journal of Neurological Sciences, 2012, 39, 516-519.	0.5	8
115	Extending the ophthalmological phenotype of Galloway-Mowat syndrome with distinct retinal dysfunction: a report and review of ocular findings. BMC Ophthalmology, 2018, 18, 147.	1.4	8
116	Multiple Sulfatase Deficiency: A Case Series With a Novel Mutation. Journal of Child Neurology, 2018, 33, 820-824.	1.4	8
117	A homozygous nonsense mutation in DCBLD2 is a candidate cause of developmental delay, dysmorphic features and restrictive cardiomyopathy. Scientific Reports, 2021, 11, 12861.	3.3	8
118	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	7.6	8
119	Pulmonary hypertension and vasculopathy in incontinentia pigmenti: a case report. Therapeutics and Clinical Risk Management, 2017, Volume 13, 629-634.	2.0	7
120	Wholeâ€exome sequencing revealed a nonsense mutation in STKLD1 causing nonâ€syndromic preâ€axial polydactyly type A affecting only upper limb. Clinical Genetics, 2019, 96, 134-139.	2.0	7
121	Further delineation of <scp>HIDEA</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2999-3006.	1.2	7
122	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. Genetics in Medicine, 2020, 22, 2071-2080.	2.4	7
123	Clinical presentation of seven patients with Methylenetetrahydrofolate reductase deficiency. Molecular Genetics and Metabolism Reports, 2020, 25, 100644.	1.1	7
124	Inherited Metabolic Causes of Stroke in Children: Mechanisms, Types, and Management. Frontiers in Neurology, 2021, 12, 633119.	2.4	7
125	Mucolipidosis II: first report from Saudi Arabia. Annals of Saudi Medicine, 2013, 33, 382-386.	1.1	7
126	Spectrum of Mutations in 60 Saudi Patients with Mut Methylmalonic Acidemia. JIMD Reports, 2014, 29, 39-46.	1.5	6

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127	Early onset of Fazio-Londe syndrome: the first case report from the Arabian Peninsula. Human Genome Variation, 2017, 4, 17018.	0.7	6
128	Delineation of cystinuria in Saudi Arabia: A case series. BMC Nephrology, 2017, 18, 50.	1.8	6
129	Aortic calcification in Gaucher disease: a case report. The Application of Clinical Genetics, 2018, Volume 11, 107-110.	3.0	6
130	A founder nonsense variant in NUDT2 causes a recessive neurodevelopmental disorder in Saudi Arab children. Clinical Genetics, 2018, 94, 393-395.	2.0	6
131	KIF16B is a candidate gene for a novel autosomalâ€recessive intellectual disability syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1602-1609.	1.2	6
132	Proteomic and Molecular Assessment of the Common Saudi Variant in ACADVL Gene Through Mesenchymal Stem Cells. Frontiers in Cell and Developmental Biology, 2019, 7, 365.	3.7	6
133	Molecular autopsy by proxy in preconception counseling. Clinical Genetics, 2021, 100, 678-691.	2.0	6
134	Novel homozygous pathogenic mitochondrial DNAJC19 variant in a patient with dilated cardiomyopathy and global developmental delay. Molecular Genetics & Genomic Medicine, 2022, 10, .	1.2	6
135	Acute intermittent porphyria caused by novel mutation in HMBS gene, misdiagnosed as cholecystitis. Neuropsychiatric Disease and Treatment, 2014, 10, 2135.	2.2	5
136	Genomic testing and counseling: The contribution of nextâ€generation sequencing to epilepsy genetics. Annals of Human Genetics, 2020, 84, 431-436.	0.8	5
137	Identification of the TTC26 Splice Variant in a Novel Complex Ciliopathy Syndrome with Biliary, Renal, Neurological, and Skeletal Manifestations. Molecular Syndromology, 2021, 12, 133-140.	0.8	5
138	The rate of secondary genomic findings in the Saudi population. American Journal of Medical Genetics, Part A, 2021, , .	1.2	5
139	Neurometabolic disorders and congenital malformations of the central nervous system. Journal of King Abdulaziz University, Islamic Economics, 2018, 23, 97-103.	1.1	5
140	A Novel Homozygous Missense Mutation in the Zinc Finger DNA Binding Domain of GLI1 Causes Recessive Post-Axial Polydactyly. Frontiers in Genetics, 2021, 12, 746949.	2.3	5
141	Prevention genetics program is an efficient model for precision medicine. International Journal of Health Sciences, 2018, 12, 1-2.	0.4	5
142	Cut-off values in newborn screening for inborn errors of metabolism in Saudi Arabia. Annals of Saudi Medicine, 2022, 42, 107-118.	1.1	5
143	A disorder clinically resembling cystic fibrosis caused by biallelic variants in the AGR2 gene. Journal of Medical Genetics, 2022, 59, 993-1001.	3.2	5
144	Diabetic ketoacidosis in vanishing white matter. Clinical Case Reports (discontinued), 2016, 4, 717-720.	0.5	4

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145	Psychological Assessment of Patients With Biotin-Thiamine-Responsive Basal Ganglia Disease. Child Neurology Open, 2017, 4, 2329048X1773074.	1.1	4
146	<scp>LRRK2</scp> Lossâ€ofâ€Function Variants in Patients with Rare Diseases: No Evidence for a Phenotypic Impact. Movement Disorders, 2021, 36, 1029-1031.	3.9	4
147	<i>FIG4</i>-Associated Yunis-Varon Syndrome: Identification of a Novel Missense Variant. Molecular Syndromology, 2021, 12, 386-392.	0.8	4
148	HMG-CoA Lyase Deficiency: A Retrospective Study of 62 Saudi Patients. Frontiers in Genetics, 2022, 13, .	2.3	4
149	Epilepsy in Propionic Acidemia: Case Series of 14 Saudi Patients. Journal of Child Neurology, 2018, 33, 713-717.	1.4	3
150	A homozygous missense variant in the homeobox domain of the NKX6â€2 results in progressive spastic ataxia type 8 associated with lower limb weakness and neurological manifestations. Journal of Gene Medicine, 2020, 22, e3196.	2.8	3
151	Spinal Cord Involvement in Pediatric-Onset Metabolic Disorders With Mendelian and Mitochondrial Inheritance. Frontiers in Pediatrics, 2020, 8, 599861.	1.9	3
152	Genetic carrier screening for disorders included in newborn screening in the Saudi population. Journal of Biochemical and Clinical Genetics, 0, , 70-75.	0.1	3
153	Severe CrohnÊ¼s Disease Manifestations in a Child with Cystathionine Î²-Synthase Deficiency. ACG Case Reports Journal, 2018, 5, e931-4.	0.4	3
154	Editorial: Pediatric Neurometabolic Disorders. Frontiers in Neurology, 2021, 12, 737398.	2.4	2
155	Report of a Case that Expands the Phenotype of Infantile Krabbe Disease. American Journal of Case Reports, 2019, 20, 643-646.	0.8	2
156	A novel MPV17 gene mutation in a Saudi infant causing fatal progressive liver failure. Annals of Saudi Medicine, 2014, 34, 175-178.	1.1	2
157	A new case of BainbridgeÂ–Ropers syndrome (BRPS): delineating the phenotype and review of literature. Journal of Biochemical and Clinical Genetics, 0, , 65-69.	0.1	2
158	Prevention of hemoglobinopathies in Saudi Arabia: efficacy of national premarital screening and the feasibility of preimplantation genetic diagnosis. Journal of Biochemical and Clinical Genetics, 0, , 94-99.	0.1	2
159	Clinical, biochemical, and molecular overview of transaldolase deficiency and evaluation of the endocrine function: update of 34 patients. Journal of Inherited Metabolic Disease, 2019, 42, 147.	3.6	1
160	MEFV c.2230G>T p.(Ala744Ser) rs61732874 previously misclassified as pathogenic variant due to lack of a population specific database. Annals of Human Genetics, 2020, 84, 370-379.	0.8	1
161	The association between IVF and chromosomal abnormalities compared to spontaneous conception. Journal of Biochemical and Clinical Genetics, 0, , 42-47.	0.1	1
162	Caring for patients with rare diseases during the COVID-19 pandemic. Journal of Infection in Developing Countries, 2021, 15, 450-462.	1.2	1

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163	The phenotypic spectrum of dihydrolipoamide dehydrogenase deficiency in Saudi Arabia. Molecular Genetics and Metabolism Reports, 2021, 29, 100817.	1.1	1
164	The variant artificial intelligence easy scoring (VARIES) system. Computers in Biology and Medicine, 2022, 145, 105492.	7.0	1
165	Beyond the caudate nucleus: Early atypical neuroimaging findings in biotin-thiamine- responsive basal ganglia disease. Brain and Development, 2022, 44, 618-622.	1.1	1
166	KAIMRCâ€™S Second Therapeutics Discovery Conference. Proceedings (mdpi), 2020, 43, 6.	0.2	0
167	Variants of uncertain significance is a clinical dilemma. Journal of Biochemical and Clinical Genetics, 0, , 68-69.	0.1	0
168	Case Report and Literature Review: Homozygous DNAJC3 Mutation in a Saudi Family Causing Maturity Onset Diabetes of the Young (MODY), Hypothyroidism, Short Stature, Neurodegeneration, and Hearing Loss. Journal of the Endocrine Society, 2021, 5, A696-A697.	0.2	0
169	Epileptic encephalopathy caused by biallelic mutation in PPM1D: a case report. Journal of Biochemical and Clinical Genetics, 0, , 47-50.	0.1	0
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