Majid Alfadhel

List of Publications by Year
in descending order

171	4,947	156536	145109
papers	citations	32 h-index	60 g-index

178	178	178	8928
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Common disease-associated gene variants in a Saudi Arabian population. Annals of Saudi Medicine, 2022, 42, 29-35.	0.5	9
2	Expanding the genotype and phenotype spectrum of SYT1-associated neurodevelopmental disorder. Genetics in Medicine, 2022, 24, 880-893.	1.1	14
3	Cut-off values in newborn screening for inborn errors of metabolism in Saudi Arabia. Annals of Saudi Medicine, 2022, 42, 107-118.	0.5	5
4	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	3.7	8
5	A disorder clinically resembling cystic fibrosis caused by biallelic variants in the AGR2 gene. Journal of Medical Genetics, 2022, 59, 993-1001.	1.5	5
6	The variant artificial intelligence easy scoring (VARIES) system. Computers in Biology and Medicine, 2022, 145, 105492.	3.9	1
7	HMG-CoA Lyase Deficiency: A Retrospective Study of 62 Saudi Patients. Frontiers in Genetics, 2022, 13, .	1.1	4
8	Novel homozygous pathogenic mitochondrial DNAJC19 variant in a patient with dilated cardiomyopathy and global developmental delay. Molecular Genetics & Genomic Medicine, 2022, 10, .	0.6	6
9	Beyond the caudate nucleus: Early atypical neuroimaging findings in biotin-thiamine- responsive basal ganglia disease. Brain and Development, 2022, 44, 618-622.	0.6	1
10	Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. Human Genetics, 2021, 140, 579-592.	1.8	14
11	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.	1.4	66
12	<scp>LRRK2</scp> Lossâ€ofâ€Function Variants in Patients with Rare Diseases: No Evidence for a Phenotypic Impact. Movement Disorders, 2021, 36, 1029-1031.	2.2	4
13	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.	0.3	5
14	Next Generation Sequencing Based Non-invasive Prenatal Testing (NIPT): First Report From Saudi Arabia. Frontiers in Genetics, 2021, 12, 630787.	1.1	29
15	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.	1.0	9
16	Biallelic variants in COPB1 cause a novel, severe intellectual disability syndrome with cataracts and variable microcephaly. Genome Medicine, 2021, 13, 34.	3.6	18
17	Next-Generation Sequencing-Based Pre-Implantation Genetic Testing for Aneuploidy (PGT-A): First Report from Saudi Arabia. Genes, 2021, 12, 461.	1.0	28
18	Inherited Metabolic Causes of Stroke in Children: Mechanisms, Types, and Management. Frontiers in Neurology, 2021, 12, 633119.	1.1	7

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19	Caring for patients with rare diseases during the COVID-19 pandemic. Journal of Infection in Developing Countries, 2021, 15, 450-462.	0.5	1
20	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.	1.1	30
21	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.	0.1	0
22	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. Frontiers in Pediatrics, 2021, 9, 633385.	0.9	15
23	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.	1.8	13
24	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	3.7	22
25	A homozygous nonsense mutation in DCBLD2 is a candidate cause of developmental delay, dysmorphic features and restrictive cardiomyopathy. Scientific Reports, 2021, 11, 12861.	1.6	8
26	Biallelic variant in DACH1, encoding Dachshund Homolog 1, defines a novel candidate locus for recessive postaxial polydactyly type A. Genomics, 2021, 113, 2495-2502.	1.3	16
27	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.	1.0	9
28	<i>FIG4</i>-Associated Yunis-Varon Syndrome: Identification of a Novel Missense Variant. Molecular Syndromology, 2021, 12, 386-392.	0.3	4
29	Molecular autopsy by proxy in preconception counseling. Clinical Genetics, 2021, 100, 678-691.	1.0	6
30	Editorial: Pediatric Neurometabolic Disorders. Frontiers in Neurology, 2021, 12, 737398.	1.1	2
31	The rate of secondary genomic findings in the Saudi population. American Journal of Medical Genetics, Part A, 2021, , .	0.7	5
32	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.	1.8	12
33	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.	1.1	5
34	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.	1.2	12
35	The phenotypic spectrum of dihydrolipoamide dehydrogenase deficiency in Saudi Arabia. Molecular Genetics and Metabolism Reports, 2021, 29, 100817.	0.4	1
36	Case Report: Homozygous Mutation Causes Monogenic Diabetes Mellitus Associated With Pancreatic Atrophy. Frontiers in Endocrinology, 2021, 12, 742278.	1.5	0

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37	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.	1.2	13
38	Further delineation of <scp>HIDEA</scp> syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2999-3006.	0.7	7
39	What is the right sequencing approach? Solo VS extended family analysis in consanguineous populations. BMC Medical Genomics, 2020, 13, 103.	0.7	10
40	Diversity of Phenotype and Genetic Etiology of 23 Cystinuria Saudi Patients: A Retrospective Study. Frontiers in Pediatrics, 2020, 8, 569389.	0.9	9
41	Clinical, molecular, and biochemical delineation of asparagine synthetase deficiency in Saudi cohort. Genetics in Medicine, 2020, 22, 2071-2080.	1.1	7
42	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
43	<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.	1.0	24
44	Clinical presentation of seven patients with Methylenetetrahydrofolate reductase deficiency. Molecular Genetics and Metabolism Reports, 2020, 25, 100644.	0.4	7
45	Homozygous truncating NEK10 mutation, associated with primary ciliary dyskinesia: a case report. BMC Pulmonary Medicine, 2020, 20, 141.	0.8	16
46	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.	0.3	1
47	Biallelic variants in four genes underlying recessive osteogenesis imperfecta. European Journal of Medical Genetics, 2020, 63, 103954.	0.7	26
48	Genomic testing and counseling: The contribution of nextâ€generation sequencing to epilepsy genetics. Annals of Human Genetics, 2020, 84, 431-436.	0.3	5
49	Mutated RAP1GDS1 causes a new syndrome of dysmorphic feature, intellectual disability & speech delay. Annals of Clinical and Translational Neurology, 2020, 7, 956-964.	1.7	21
50	KAIMRCâ€™S Second Therapeutics Discovery Conference. Proceedings (mdpi), 2020, 43, 6.	0.2	0
51	A Missense Mutation in the UGDH Gene Is Associated With Developmental Delay and Axial Hypotonia. Frontiers in Pediatrics, 2020, 8, 71.	0.9	15
52	KCNT1â€related epilepsy: An international multicenter cohort of 27 pediatric cases. Epilepsia, 2020, 61, 679-692.	2.6	50
53	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.	1.0	18
54	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.	1.4	3

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55	Spinal Cord Involvement in Pediatric-Onset Metabolic Disorders With Mendelian and Mitochondrial Inheritance. Frontiers in Pediatrics, 2020, 8, 599861.	0.9	3
56	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.	1.7	1
57	Delta Like-1 Gene Mutation: A Novel Cause of Congenital Vertebral Malformation. Frontiers in Genetics, 2019, 10, 534.	1.1	17
58	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
59	Targeted SLC19A3 gene sequencing of 3000 Saudi newborn: a pilot study toward newborn screening. Annals of Clinical and Translational Neurology, 2019, 6, 2097-2103.	1.7	38
60	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.	1.6	19
61	Biallelic Missense Mutation in the ECEL1 Underlies Distal Arthrogryposis Type 5 (DA5D). Frontiers in Pediatrics, 2019, 7, 343.	0.9	15
62	Carnitine Inborn Errors of Metabolism. Molecules, 2019, 24, 3251.	1.7	70
63	SGCD Homozygous Nonsense Mutation (p.Arg97â^—) Causing Limb-Girdle Muscular Dystrophy Type 2F (LGMD2F) in a Consanguineous Family, a Case Report. Frontiers in Genetics, 2019, 9, 727.	1.1	19
64	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.	0.7	9
65	Biallelic SCN2A Gene Mutation Causing Early Infantile Epileptic Encephalopathy: Case Report and Review. Journal of Central Nervous System Disease, 2019, 11, 117957351984993.	0.7	10
66	Multiple Mitochondrial Dysfunctions Syndrome 4 Due to ISCA2 Gene Defects: A Review. Child Neurology Open, 2019, 6, 2329048X1984737.	0.5	16
67	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.	1.1	9
68	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.	0.6	15
69	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.	0.4	14
70	The landscape of early infantile epileptic encephalopathy in a consanguineous population. Seizure: the Journal of the British Epilepsy Association, 2019, 69, 154-172.	0.9	34
71	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
72	Glycine Transporter 1 Encephalopathy From Biochemical Pathway to Clinical Disease: Review. Child Neurology Open, 2019, 6, 2329048X1983148.	0.5	10

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73	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.	1.0	7
74	6-Pyruvoyltetrahydropterin Synthase Deficiency: Review and Report of 28 Arab Subjects. Pediatric Neurology, 2019, 96, 40-47.	1.0	12
75	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.	0.4	12
76	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.	1.7	26
77	Genetic Disorders Associated with Metal Metabolism. Cells, 2019, 8, 1598.	1.8	31
78	The many faces of peroxisomal disorders: Lessons from a large Arab cohort. Clinical Genetics, 2019, 95, 310-319.	1.0	12
79	Adenosine Kinase Deficiency: Report and Review. Neuropediatrics, 2019, 50, 046-050.	0.3	12
80	Autozygome and high throughput confirmation of disease genes candidacy. Genetics in Medicine, 2019, 21, 736-742.	1.1	81
81	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.	1.8	6
82	Report of a Case that Expands the Phenotype of Infantile Krabbe Disease. American Journal of Case Reports, 2019, 20, 643-646.	0.3	2
83	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	1.1	46
84	Secondary Hemophagocytic Syndrome Associated with COG6 Gene Defect: Report and Review. JIMD Reports, 2018, 42, 105-111.	0.7	23
85	PRUNE Syndrome Is a New Neurodevelopmental Disorder. Child Neurology Open, 2018, 5, 2329048X1775223.	0.5	10
86	Expanding the clinical and genetic spectra of NKX6â€2â€related disorder. Clinical Genetics, 2018, 93, 1087-1092.	1.0	10
87	Deficiency of ADA2 mimicking autoimmune lymphoproliferative syndrome in the absence of livedo reticularis and vasculitis. Pediatric Blood and Cancer, 2018, 65, e26912.	0.8	37
88	A new association between CDK5RAP2 microcephaly and congenital cataracts. Annals of Human Genetics, 2018, 82, 165-170.	0.3	9
89	MPV17-related mitochondrial DNA maintenance defect: New cases and review of clinical, biochemical, and molecular aspects. Human Mutation, 2018, 39, 461-470.	1.1	45
90	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.	0.7	19

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91	Whole-genome sequencing offers additional but limited clinical utility compared with reanalysis of whole-exome sequencing. Genetics in Medicine, 2018, 20, 1328-1333.	1.1	118
92	Further delineation of the phenotypic spectrum ofÂ ISCA2 defect: A report of ten new cases. European Journal of Paediatric Neurology, 2018, 22, 46-55.	0.7	21
93	SLC19A3 Gene Defects Sorting the Phenotype and Acronyms: Review. Neuropediatrics, 2018, 49, 083-092.	0.3	30
94	Phenotypic and Molecular Spectrum of Aicardi-GoutiÃ¨res Syndrome: A Study of 24 Patients. Pediatric Neurology, 2018, 78, 35-40.	1.0	40
95	Novel homozygous mutation in the WWOX gene causes seizures and global developmental delay: Report and review. Translational Neuroscience, 2018, 9, 203-208.	0.7	23
96	Loss of tubulin deglutamylase <scp>CCP</scp> 1 causes infantileâ€onset neurodegeneration. EMBO Journal, 2018, 37, .	3.5	86
97	Aortic calcification in Gaucher disease: a case report. The Application of Clinical Genetics, 2018, Volume 11, 107-110.	1.4	6
98	Clinical Genetics of Polydactyly: An Updated Review. Frontiers in Genetics, 2018, 9, 447.	1.1	66
99	Inborn errors of metabolism associated with hyperglycaemic ketoacidosis and diabetes mellitus: narrative review. Sudanese Journal of Paediatrics, 2018, 18, 10-23.	0.6	9
100	Tetrasomy 18p: case report and review of literature. The Application of Clinical Genetics, 2018, Volume 11, 9-14.	1.4	14
101	Utilizing Whole-Exome Sequencing to Characterize the Phenotypic Variability of Sickle Cell Disease. Genetic Testing and Molecular Biomarkers, 2018, 22, 561-567.	0.3	29
102	Methionine adenosyltransferase I/III deficiency: beyond the central nervous system manifestations. Therapeutics and Clinical Risk Management, 2018, Volume 14, 225-229.	0.9	9
103	A founder nonsense variant in NUDT2 causes a recessive neurodevelopmental disorder in Saudi Arab children. Clinical Genetics, 2018, 94, 393-395.	1.0	6
104	Extending the ophthalmological phenotype of Galloway-Mowat syndrome with distinct retinal dysfunction: a report and review of ocular findings. BMC Ophthalmology, 2018, 18, 147.	0.6	8
105	Risk of Neuropsychiatric Adverse Effects of Lipid-Lowering Drugs: A Mendelian Randomization Study. International Journal of Neuropsychopharmacology, 2018, 21, 1067-1075.	1.0	29
106	Epilepsy in Propionic Acidemia: Case Series of 14 Saudi Patients. Journal of Child Neurology, 2018, 33, 713-717.	0.7	3
107	KIF16B is a candidate gene for a novel autosomalâ€recessive intellectual disability syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1602-1609.	0.7	6
108	Multiple Sulfatase Deficiency: A Case Series With a Novel Mutation. Journal of Child Neurology, 2018, 33, 820-824.	0.7	8

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109	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	3.7	70
110	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.	1.4	37
111	Neurometabolic disorders and congenital malformations of the central nervous system. Journal of King Abdulaziz University, Islamic Economics, 2018, 23, 97-103.	0.5	5
112	Severe CrohnÊ¼s Disease Manifestations in a Child with Cystathionine Î²-Synthase Deficiency. ACG Case Reports Journal, 2018, 5, e931-4.	0.2	3
113	Prevention genetics program is an efficient model for precision medicine. International Journal of Health Sciences, 2018, 12, 1-2.	0.4	5
114	The SORCS3 gene is mutated in brothers with infantile spasms and intellectual disability. Discovery Medicine, 2018, 26, 147-153.	0.5	9
115	Early-Onset Parkinsonism: Case Report and Review of the Literature. Pediatric Neurology, 2017, 67, 102-106.e1.	1.0	10
116	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.	0.9	10
117	Long-term Outcome of 4 Patients With Transcobalamin Deficiency Caused by 2 Novel TCN2 Mutations. Journal of Pediatric Hematology/Oncology, 2017, 39, e430-e436.	0.3	21
118	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
119	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.	0.5	68
120	Expanded Newborn Screening Program in Saudi Arabia: Incidence of screened disorders. Journal of Paediatrics and Child Health, 2017, 53, 585-591.	0.4	83
121	Clinical, Biochemical, and Molecular Features in 37 Saudi Patients with Very Long Chain Acyl CoA Dehydrogenase Deficiency. JIMD Reports, 2017, 40, 47-53.	0.7	15
122	Psychological Assessment of Patients With Biotin-Thiamine-Responsive Basal Ganglia Disease. Child Neurology Open, 2017, 4, 2329048X1773074.	0.5	4
123	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	1.1	41
124	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	1.8	122
125	CDK10 Mutations in Humans and Mice Cause Severe Growth Retardation, Spine Malformations, and Developmental Delays. American Journal of Human Genetics, 2017, 101, 391-403.	2.6	35
126	Early onset of Fazio-Londe syndrome: the first case report from the Arabian Peninsula. Human Genome Variation, 2017, 4, 17018.	0.4	6

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127	Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Annals of Neurology, 2017, 82, 317-330.	2.8	65
128	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.	0.7	12
129	Delineation of cystinuria in Saudi Arabia: A case series. BMC Nephrology, 2017, 18, 50.	0.8	6
130	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	1.4	291
131	Early Infantile Leigh-like SLC19A3 Gene Defects Have a Poor Prognosis: Report and Review. Journal of Central Nervous System Disease, 2017, 9, 117957351773752.	0.7	13
132	Pulmonary hypertension and vasculopathy in incontinentia pigmenti: a case report. Therapeutics and Clinical Risk Management, 2017, Volume 13, 629-634.	0.9	7
133	Congenital disorders of glycosylation: The Saudi experience. American Journal of Medical Genetics, Part A, 2017, 173, 2614-2621.	0.7	31
134	Mitochondrial iron-sulfur cluster biogenesis from molecular understanding to clinical disease. Journal of King Abdulaziz University, Islamic Economics, 2017, 22, 4-13.	0.5	14
135	Guidelines for acute management of hyperammonemia in the Middle East region. Therapeutics and Clinical Risk Management, 2016, 12, 479.	0.9	32
136	Expanding the clinical and genetic heterogeneity of hereditary disorders of connective tissue. Human Genetics, 2016, 135, 525-540.	1.8	89
137	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	13.9	254
138	Lossâ€ofâ€function mutation in RUSC2 causes intellectual disability and secondary microcephaly. Developmental Medicine and Child Neurology, 2016, 58, 1317-1322.	1.1	12
139	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.	0.5	9
140	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.	1.2	61
141	Mutation in SLC6A9 encoding a glycine transporter causes a novel form of non-ketotic hyperglycinemia in humans. Human Genetics, 2016, 135, 1263-1268.	1.8	35
142	Diabetic ketoacidosis in vanishing white matter. Clinical Case Reports (discontinued), 2016, 4, 717-720.	0.2	4
143	Worsening of Seizures After Asparagine Supplementation in a Child with Asparagine Synthetase Deficiency. Pediatric Neurology, 2016, 58, 98-100.	1.0	24
144	GOLGA2, encoding a master regulator of golgi apparatus, is mutated in a patient with a neuromuscular disorder. Human Genetics, 2016, 135, 245-251.	1.8	38

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145	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.	1.1	23
146	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
147	ISCA2mutation causes infantile neurodegenerative mitochondrial disorder. Journal of Medical Genetics, 2015, 52, 186-194.	1.5	90
148	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.	0.7	43
149	Asparagine Synthetase Deficiency: New Inborn Errors of Metabolism. JIMD Reports, 2015, 22, 11-16.	0.7	46
150	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.	0.7	9
151	Acute intermittent porphyria caused by novel mutation in HMBS gene, misdiagnosed as cholecystitis. Neuropsychiatric Disease and Treatment, 2014, 10, 2135.	1.0	5
152	Spectrum of Mutations in 60 Saudi Patients with Mut Methylmalonic Acidemia. JIMD Reports, 2014, 29, 39-46.	0.7	6
153	Clinical and Molecular Characteristics of Mitochondrial DNA Depletion Syndrome Associated with Neonatal Cholestasis and Liver Failure. Journal of Pediatrics, 2014, 164, 553-559.e2.	0.9	44
154	Hereditary Neurometabolic Causes of Infantile Spasms in 80 Children Presenting to a Tertiary Care Center. Pediatric Neurology, 2014, 51, 390-397.	1.0	21
155	A novel MPV17 gene mutation in a Saudi infant causing fatal progressive liver failure. Annals of Saudi Medicine, 2014, 34, 175-178.	0.5	2
156	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
157	Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. Journal of Inherited Metabolic Disease, 2013, 36, 997-1004.	1.7	36
158	Drug treatment of inborn errors of metabolism: a systematic review. Archives of Disease in Childhood, 2013, 98, 454-461.	1.0	28
159	Mucolipidosis II: first report from Saudi Arabia. Annals of Saudi Medicine, 2013, 33, 382-386.	0.5	7
160	Variability of Phenotype in Two Sisters with Pyridoxine Dependent Epilepsy. Canadian Journal of Neurological Sciences, 2012, 39, 516-519.	0.3	8
161	Extreme intrafamilial variability of Saudi brothers with primary hyperoxaluria type 1. Therapeutics and Clinical Risk Management, 2012, 8, 373.	0.9	11
162	Enzyme replacement therapy for Fabry disease: some answers but more questions. Therapeutics and Clinical Risk Management, 2011, 7, 69.	0.9	27

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163	Precocious Puberty in Two Girls With PEHO Syndrome: A Clinical Feature Not Previously Described. Journal of Child Neurology, 2011, 26, 851-857.	0.7	12
164	Hypomorphic mutations in syndromic encephalocele genes are associated with Bardet-Biedl syndrome. Nature Genetics, 2008, 40, 443-448.	9.4	367
165	Senior-Loken syndrome in a Saudi child. Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia, 2008, 19, 443-5.	0.4	0
166	The association between IVF and chromosomal abnormalities compared to spontaneous conception. Journal of Biochemical and Clinical Genetics, 0, , 42-47.	0.1	1
167	Variants of uncertain significance is a clinical dilemma. Journal of Biochemical and Clinical Genetics, 0, , 68-69.	0.1	0
168	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
169	Epileptic encephalopathy caused by biallelic mutation in PPM1D: a case report. Journal of Biochemical and Clinical Genetics, 0, , 47-50.	0.1	0
170	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
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