Allan Bayat

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/4082340/publications.pdf

Version: 2024-02-01

45 papers	895 citations	623734 14 h-index	526287 27 g-index
46 all docs	46 docs citations	46 times ranked	1902 citing authors

#	Article	IF	Citations
1	ITSN1: a novel candidate gene involved in autosomal dominant neurodevelopmental disorder spectrum. European Journal of Human Genetics, 2022, 30, 111-116.	2.8	4
2	Pyridoxine or pyridoxalâ€5â€phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study. Developmental Medicine and Child Neurology, 2022, 64, 789-798.	2.1	6
3	Phenotypic spectrum of the recurrent TRPM3 p.(<scp>Val837Met</scp>) substitution in seven individuals with global developmental delay and hypotonia. American Journal of Medical Genetics, Part A, 2022, 188, 1667-1675.	1.2	8
4	PIGNencephalopathy: Characterizing the epileptology. Epilepsia, 2022, 63, 974-991.	5.1	4
5	Spectrum of Phenotypic, Genetic, and Functional Characteristics in Patients With Epilepsy With KCNC2 Pathogenic Variants. Neurology, 2022, 98, .	1.1	11
6	Novel truncating variants in <scp>FGD1</scp> detected in two Danish families with <scp>Aarskogâ€“Scott</scp> syndrome and myopathic features. American Journal of Medical Genetics, Part A, 2022, 188, 2251-2257.	1.2	3
7	Impact of Genetic Testing on Therapeutic Decision-Making in Childhood-Onset Epilepsiesâ€”a Study in a Tertiary Epilepsy Center. Neurotherapeutics, 2022, 19, 1353-1367.	4.4	14
8	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	1.6	15
9	Phenotypic expansion of the <scp>BPTF</scp>â€related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies. American Journal of Medical Genetics, Part A, 2021, 185, 1366-1378.	1.2	8
10	PRICKLE2 revisitedâ€”further evidence implicating PRICKLE2 in neurodevelopmental disorders. European Journal of Human Genetics, 2021, 29, 1235-1244.	2.8	5
11	Expansion of the CCDC22 associated Ritscher-Schinzel/3C syndrome and review of the literature: Should the minimal diagnostic criteria be revised?. European Journal of Medical Genetics, 2021, 64, 104246.	1.3	11
12	Epilepsy Syndromes in the First Year of Life and Usefulness of Genetic Testing for Precision Therapy. Genes, 2021, 12, 1051.	2.4	36
13	5q11.2 deletion syndrome revisitedâ€”Further narrowing of the smallest region of overlap for the main clinical characteristics of the syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3844-3850.	1.2	0
14	PURA- Related Developmental and Epileptic Encephalopathy. Neurology: Genetics, 2021, 7, e613.	1.9	15
15	A clinical scoring system for congenital contractural arachnodactyly. Genetics in Medicine, 2020, 22, 124-131.	2.4	17
16	Expanding the phenotype of Wiedemannâ€Steiner syndrome: Craniovertebral junction anomalies. American Journal of Medical Genetics, Part A, 2020, 182, 2877-2886.	1.2	9
17	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	5.1	32
18	Mowat-Wilson syndrome: growth charts. Orphanet Journal of Rare Diseases, 2020, 15, 151.	2.7	12

#	Article	IF	Citations
19	Prevalence and causes of infantile nystagmus in a large populationâ€based Danish cohort. Acta Ophthalmologica, 2020, 98, 506-513.	1.1	17
20	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
21	Novel Clinical and Radiological Findings in a Family with Autosomal Recessive Omodysplasia. Molecular Syndromology, 2020, 11, 83-89.	0.8	4
22	Neurological manifestations of neurofibromatosis: a review. Neurological Sciences, 2020, 41, 2685-2690.	1.9	10
23	PEDIA: prioritization of exome data by image analysis. Genetics in Medicine, 2019, 21, 2807-2814.	2.4	58
24	PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. Genetics in Medicine, 2019, 21, 2216-2223.	2.4	21
25	Hereditary leukodystrophy with axonal spheroids (HDLS) presenting subacutely: a CNS-vasculitis mimic. Acta Neurologica Belgica, 2019, 119, 633-635.	1.1	2
26	Enhancement of cranial nerves, conus medullaris, and nerve roots in POLG mitochondrial disease. Neurology: Genetics, 2019, 5, e360.	1.9	2
27	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
28	Chronic intestinal pseudo-obstruction syndrome and gastrointestinal malrotation in an infantwith schaaf-yang syndrome - Expanding the phenotypic spectrum. European Journal of Medical Genetics, 2018, 61, 627-630.	1.3	9
29	Phenotype and genotype of 87 patients with Mowatâ€“Wilson syndrome and recommendations for care. Genetics in Medicine, 2018, 20, 965-975.	2.4	67
30	Letter to the editor: insular stroke presenting with acute onset of pain. Journal of Neurology, 2018, 265, 1472-1473.	3.6	2
31	Neonatal hyperinsulinemic hypoglycemia in a patient with 9p deletion syndrome. European Journal of Medical Genetics, 2018, 61, 473-477.	1.3	4
32	Case report: Hemorrhage in the wall of an abscess mimicking a hemorrhagic tumor. Neurologia I Neurochirurgia Polska, 2018, 52, 546-547.	1.2	0
33	Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Medicine, 2018, 10, 3.	8.2	67
34	Prominent and elongated coccyx, a new manifestation of KBG syndrome associated with novel mutation in ANKRD11. American Journal of Medical Genetics, Part A, 2018, 176, 1991-1995.	1.2	10
35	The evolving craniofacial phenotype of a patient with Sensenbrenner syndrome caused by IFT140 compound heterozygous mutations. Clinical Dysmorphology, 2017, 26, 247-251.	0.3	18
36	Neurodevelopmental Disorders Caused by De Novo Variants in KCNB1 Genotypes and Phenotypes. JAMA Neurology, 2017, 74, 1228.	9.0	79

#	Article	IF	Citations
37	Familial craniofacial abnormality and polymicrogyria associated with a microdeletion affecting the NFIA gene. Clinical Dysmorphology, 2017, 26, 148-153.	0.3	10
38	Neuroimaging findings in Mowatâ€“Wilson syndrome: a study of 54 patients. Genetics in Medicine, 2017, 19, 691-700.	2.4	45
39	Cover Image, Volume 170A, Number 6, June 2016. , 2016, 170, i-i.		0
40	Further delineation of facioaudiosymphalangism syndrome: Description of a family with a novel NOG mutation and without hearing loss. American Journal of Medical Genetics, Part A, 2016, 170, 1479-1484.	1.2	8
41	The incidence of <scp>SCN</scp>1Aâ€related Dravet syndrome in <scp>D</scp>enmark is 1:22,000: A populationâ€based study from 2004 to 2009. Epilepsia, 2015, 56, e36-9.	5.1	103
42	Mild Leschâ€“Nyhan Disease in a Boy with a Null Mutation in HPRT1: An Exception to the Known Genotypeâ€“Phenotype Correlation. JIMD Reports, 2014, 18, 135-137.	1.5	0
43	Characteristics and outcome of Goodpastureâ€™s disease in children. Clinical Rheumatology, 2012, 31, 1745-1751.	2.2	31
44	Small duct autoimmune sclerosing cholangitis and Crohn colitis in a 10-year-old child. A case report and review of the literature. Diagnostic Pathology, 2012, 7, 100.	2.0	6
45	Incidence of fetal akinesiaâ€hypokinesia deformation sequence: a populationâ€based study. Acta Paediatrica, International Journal of Paediatrics, 2009, 98, 3-4.	1.5	12