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List of Publications by Year
in descending order

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27 papers	1,354 citations	516710 16 h-index	501196 28 g-index
29 all docs	29 docs citations	29 times ranked	2868 citing authors

#	Article	IF	Citations
1	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	1.2	447
2	Mutations in GTPBP3 Cause a Mitochondrial Translation Defect Associated with Hypertrophic Cardiomyopathy, Lactic Acidosis, and Encephalopathy. American Journal of Human Genetics, 2014, 95, 708-720.	6.2	123
3	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
4	The G93C Mutation in Superoxide Dismutase 1. Archives of Neurology, 2006, 63, 262.	4.5	81
5	Classic infantile Pompe patients approaching adulthood: a cohort study on consequences for the brain. Developmental Medicine and Child Neurology, 2018, 60, 579-586.	2.1	77
6	Mutations in PEX10 are a cause of autosomal recessive ataxia. Annals of Neurology, 2010, 68, 259-263.	5.3	74
7	PREPL deficiency with or without cystinuria causes a novel myasthenic syndrome. Neurology, 2014, 82, 1254-1260.	1.1	52
8	Isolated sulfite oxidase deficiency. Journal of Inherited Metabolic Disease, 2018, 41, 101-108.	3.6	51
9	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
10	NPC1 defect results in abnormal platelet formation and function: studies in Niemannâ€“Pick disease type C1 patients and zebrafish. Human Molecular Genetics, 2013, 22, 61-73.	2.9	39
11	COG5-CDG: expanding the clinical spectrum. Orphanet Journal of Rare Diseases, 2012, 7, 94.	2.7	36
12	Peroxisomal Disorders: A Review on Cerebellar Pathologies. Brain Pathology, 2015, 25, 663-678.	4.1	33
13	Cardiac outcome in classic infantile Pompe disease after 13â€¯years of treatment with recombinant human acid alpha-glucosidase. International Journal of Cardiology, 2018, 269, 104-110.	1.7	32
14	PREPL deficiency: delineation of the phenotype and development of a functional blood assay. Genetics in Medicine, 2018, 20, 109-118.	2.4	29
15	Neuromyelitis optica-IgG(+) optic neuritis associated with celiac disease and dysgammaglobulinemia: A role for tacrolimus?. European Journal of Paediatric Neurology, 2011, 15, 265-267.	1.6	22
16	PREPL, a Prolyl Endopeptidase-Like Enzyme by Name Only? â€“ Lessons from Patients. CNS and Neurological Disorders - Drug Targets, 2011, 10, 355-360.	1.4	16
17	Two novel deletions in hypotoniaâ€“cystinuria syndrome. Molecular Genetics and Metabolism, 2012, 107, 614-616.	1.1	16
18	Misdiagnosis as asphyxiating thoracic dystrophy and CMV-associated haemophagocytic lymphohistiocytosis in Shwachman-Diamond syndrome. European Journal of Pediatrics, 2013, 172, 613-622.	2.7	16

#	Article	IF	Citations
19	Cerebral Syphilitic Gumma in a Human Immunodeficiency Virusâ€“Positive Patient. Archives of Neurology, 2005, 62, 1310.	4.5	15
20	Recurrent NEDD4L Variant in Periventricular Nodular Heterotopia, Polymicrogyria and Syndactyly. Frontiers in Genetics, 2020, 11, 26.	2.3	12
21	Defining the phenotypical spectrum associated with variants in TUBB2A. Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11
22	Novel Infantile-Onset Leukoencephalopathy With High Lactate Level and Slow Improvement. Archives of Neurology, 2012, 69, 718-22.	4.5	9
23	Prolyl endopeptidase-like is a (thio)esterase involved in mitochondrial respiratory chain function. IScience, 2021, 24, 103460.	4.1	8
24	Necessity of Fractionated Urine Collection for Monitoring Patients with Cystinuria. Clinical Chemistry, 2011, 57, 780-781.	3.2	7
25	Parenteral hydroxocobalamin dose intensification in five patients with different types of early onset intracellular cobalamin defects: Clinical and biochemical responses. JIMD Reports, 2019, 49, 70-79.	1.5	7
26	Normal cognitive outcome in a PEX6 deficient girl despite neonatal multisystem presentation. American Journal of Medical Genetics, Part A, 2016, 170, 1642-1646.	1.2	4
27	Evaluation of the pediatric patient with hypotonia: donâ€™t forget the hypotoniaâ€cystinuria syndrome!. Developmental Medicine and Child Neurology, 2012, 54, 288-288.	2.1	2