Luc RÃ©gal

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	Defining the phenotypical spectrum associated with variants in TUBB2A. Journal of Medical Genetics, 2021, 58, 33-40.	3.2	11
2	Prolyl endopeptidase-like is a (thio)esterase involved in mitochondrial respiratory chain function. IScience, 2021, 24, 103460.	4.1	8
3	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
4	Recurrent NEDD4L Variant in Periventricular Nodular Heterotopia, Polymicrogyria and Syndactyly. Frontiers in Genetics, 2020, 11, 26.	2.3	12
5	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
6	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
7	PREPL deficiency: delineation of the phenotype and development of a functional blood assay. Genetics in Medicine, 2018, 20, 109-118.	2.4	29
8	Isolated sulfite oxidase deficiency. Journal of Inherited Metabolic Disease, 2018, 41, 101-108.	3.6	51
9	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
10	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
11	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
12	Peroxisomal Disorders: A Review on Cerebellar Pathologies. Brain Pathology, 2015, 25, 663-678.	4.1	33
13	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
14	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
15	PREPL deficiency with or without cystinuria causes a novel myasthenic syndrome. Neurology, 2014, 82, 1254-1260.	1.1	52
16	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
17	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
18	Novel Infantile-Onset Leukoencephalopathy With High Lactate Level and Slow Improvement. Archives of Neurology, 2012, 69, 718-22.	4.5	9

#	Article	IF	Citations
19	Two novel deletions in hypotoniaâ€“cystinuria syndrome. Molecular Genetics and Metabolism, 2012, 107, 614-616.	1.1	16
20	COG5-CDG: expanding the clinical spectrum. Orphanet Journal of Rare Diseases, 2012, 7, 94.	2.7	36
21	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
22	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
23	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
24	Necessity of Fractionated Urine Collection for Monitoring Patients with Cystinuria. Clinical Chemistry, 2011, 57, 780-781.	3.2	7
25	Mutations in PEX10 are a cause of autosomal recessive ataxia. Annals of Neurology, 2010, 68, 259-263.	5.3	74
26	The G93C Mutation in Superoxide Dismutase 1. Archives of Neurology, 2006, 63, 262.	4.5	81
27	Cerebral Syphilitic Gumma in a Human Immunodeficiency Virusâ€“Positive Patient. Archives of Neurology, 2005, 62, 1310.	4.5	15