Uros Hladnik

List of Publications by Year
in descending order

23	385	759233 12	794594 19
papers	citations	h-index	g-index

23	23	23	751
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Thyroid function in patients with Prader-Willi syndrome: an Italian multicenter study of 339 patients. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 159-165.	0.9	32
2	Growth rate and myofibroblast differentiation of desmoid fibroblast-like cells are modulated by TGF-Î² signaling. Histochemistry and Cell Biology, 2019, 151, 145-160.	1.7	8
3	A novel nonsense ATP7A pathogenic variant in a family exhibiting a variable occipital horn syndrome phenotype. Molecular Genetics and Metabolism Reports, 2017, 13, 14-17.	1.1	7
4	13 novel putative mutations in ATP7A found in a cohort of 25 Italian families. Metabolic Brain Disease, 2017, 32, 1173-1183.	2.9	7
5	Clinical and Molecular Characterization of Prader-Willi Syndrome. Indian Journal of Pediatrics, 2017, 84, 815-821.	0.8	12
6	The Wnt/Î²-catenin pathway in human fibrotic-like diseases and its eligibility as a therapeutic target. Molecular and Cellular Therapies, 2015, 3, 1.	0.2	54
7	Farber disease in infancy resembling juvenile idiopathic arthritis: identification of two new mutations and a good early response to allogeneic haematopoietic stem cell transplantation. Rheumatology, 2014, 53, 1533-1534.	1.9	27
8	An Italian Cohort Study Identifies Four New Pathologic Mutations in the ARSA Gene. Journal of Molecular Neuroscience, 2013, 50, 284-290.	2.3	8
9	HIV-1 TAT-mediated protein transduction of human HPRT into deficient cells. Biochemical and Biophysical Research Communications, 2013, 441, 114-119.	2.1	3
10	Nuclear <scp>GSK</scp>â€3Î² segregation in desmoidâ€type fibromatosis. Histopathology, 2013, 62, 1098-1108.	2.9	6
11	New Niemann-Pick Type C1 Gene Mutation Associated With Very Severe Disease Course and Marked Early Cerebellar Vermis Atrophy. Journal of Child Neurology, 2013, 28, 1694-1697.	1.4	8
12	Huntington's disease gene expansion associates with early onset nonprogressive chorea. Movement Disorders, 2013, 28, 684-684.	3.9	1
13	Quantitative Evaluation of the Clinical Effects ofS-Adenosylmethionine on Mood and Behavior in Lesch-Nyhan Patients. Nucleosides, Nucleotides and Nucleic Acids, 2013, 32, 174-188.	1.1	20
14	Two new large deletions of the AVPR2 gene causing nephrogenic diabetes insipidus and a review of previously published deletions. Nephrology Dialysis Transplantation, 2012, 27, 3705-3712.	0.7	15
15	Specific treatment of Praderâ€“Willi syndrome through cyclical rehabilitation programmes. Disability and Rehabilitation, 2011, 33, 1837-1847.	1.8	31
16	Two Novel Homozygous SACS Mutations in Unrelated Patients Including the First Reported Case of Paternal UPD as an Etiologic Cause of ARSACS. Journal of Molecular Neuroscience, 2011, 43, 346-349.	2.3	16
17	Metabonomics and population studies: age-related amino acids excretion and inferring networks through the study of urine samples in two Italian isolated populations. Amino Acids, 2010, 38, 65-73.	2.7	18
18	Analysis of the HPRT1 gene in 35 Italian Leschâ€“Nyhan families: 45 patients and 77 potential female carriers. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2010, 692, 1-5.	1.0	17

#	Article	IF	Citations
19	Human neural stem cells: a model system for the study of Leschâ€“Nyhan disease neurological aspects. Human Molecular Genetics, 2010, 19, 1939-1950.	2.9	28
20	A Case of Lymphedema-Distichiasis Syndrome Carrying a New de novo Frameshift FOXC2 Mutation. Ophthalmic Genetics, 2010, 31, 98-100.	1.2	8
21	Age-related hearing loss in four Italian genetic isolates: An epidemiological study. International Journal of Audiology, 2009, 48, 465-472.	1.7	17
22	Atypical arrhythmic complications in familial hypokalemic periodic paralysis. Journal of Cardiovascular Medicine, 2009, 10, 68-71.	1.5	12
23	Variable Expression of HPRT Deficiency in 5 Members of a Family With the Same Mutation. Archives of Neurology, 2008, 65, 1240-3.	4.5	30