Andrea Bordugo

List of Publications by Year
in descending order

28 papers	739 citations	686830 13 h-index	525886 27 g-index
29 all docs	29 docs citations	29 times ranked	953 citing authors

#	Article	IF	Citations
1	A Gain-of-Function Mutation on BCKDK Gene and Its Possible Pathogenic Role in Branched-Chain Amino Acid Metabolism. Genes, 2022, 13, 233.	1.0	3
2	First paediatric COVID â€19 associated death in Italy. Journal of Paediatrics and Child Health, 2021, 57, 736-737.	0.4	7
3	Epilepsy and movement disorders in CDG : Report on the oldestâ€known MOGSâ€CDG patient. American Journal of Medical Genetics, Part A, 2021, 185, 219-222.	0.7	10
4	HILIC-UPLC-MS for high throughput and isomeric N-glycan separation and characterization in Congenital Disorders Glycosylation and human diseases. Glycoconjugate Journal, 2021, 38, 201-211.	1.4	20
5	Genotype and residual enzyme activity in mediumâ€chain <scp>acylâ€CoA</scp> dehydrogenase (<scp>MCAD</scp>) deficiency: Are predictions possible?. Journal of Inherited Metabolic Disease, 2021, 44, 916-925.	1.7	7
6	Newborn Screening for Biotinidase Deficiency. The Experience of a Regional Center in Italy. Frontiers in Pediatrics, 2021, 9, 661416.	0.9	9
7	Assessing Gut Microbiota in an Infant with Congenital Propionic Acidemia before and after Probiotic Supplementation. Microorganisms, 2021, 9, 2599.	1.6	5
8	The impact of COVID-19 pandemic on the diagnosis and management of inborn errors of metabolism: A global perspective. Molecular Genetics and Metabolism, 2020, 131, 285-288.	0.5	31
9	Molecular Genetics of Niemannâ€“Pick Type C Disease in Italy: An Update on 105 Patients and Description of 18 NPC1 Novel Variants. Journal of Clinical Medicine, 2020, 9, 679.	1.0	21
10	Dietary lipids in glycogen storage disease type III: A systematic literature study, case studies, and future recommendations. Journal of Inherited Metabolic Disease, 2020, 43, 770-777.	1.7	23
11	Multiple acyl-COA dehydrogenase deficiency in elderly carriers. Journal of Neurology, 2020, 267, 1414-1419.	1.8	23
12	Bone marrow features in Pearson syndrome with neonatal onset: A case report and review of the literature. Pediatric Blood and Cancer, 2018, 65, e26939.	0.8	15
13	A neonate with abdominal distension and failure to thrive. Archives of Disease in Childhood: Education and Practice Edition, 2017, 102, 166-166.	0.3	0
14	Newborn with rhizomelia and ï»¿difficulty breathing. Skeletal Radiology, 2017, 46, 231-231.	1.2	0
15	Newborn with rhizomelia and difficulty breathing. Skeletal Radiology, 2017, 46, 291-292.	1.2	1
16	High-protein goatâ€™s milk diet identified through newborn screening: clinical warning of a potentially dangerous dietetic practice. Public Health Nutrition, 2017, 20, 2806-2809.	1.1	4
17	Images from 18F-DOPA Scan in Congenital Hyperinsulinism: Not Always a Clue for Diagnosis. Nuclear Medicine and Molecular Imaging, 2017, 51, 362-363.	0.6	1
18	Clinical relevance of short-chain acyl-CoA dehydrogenase (SCAD) deficiency: Exploring the role of new variants including the first SCAD-disease-causing allele carrying a synonymous mutation. BBA Clinical, 2016, 5, 114-119.	4.1	27

#	Article	IF	Citations
19	Comprehensive Evaluation of Plasma 7-Ketocholesterol and Cholestan-3Î²,5Î±,6Î²-Triol in an Italian Cohort of Patients Affected by Niemann-Pick Disease due to NPC1 and SMPD1 Mutations. Clinica Chimica Acta, 2016, 455, 39-45.	0.5	42
20	Successful Use of Longâ€Acting Octreotide for Intractable Chronic Gastrointestinal Bleeding in Children. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, 48-53.	0.9	12
21	A neonate with a â€˜milkyâ€™ blood. What can it be?. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2014, 99, F514-F514.	1.4	3
22	Metabolic stroke in a late-onset form of isolated sulfite oxidase deficiency. Molecular Genetics and Metabolism, 2013, 108, 263-266.	0.5	30
23	Puberty is associated with increased deterioration of renal function in patients with CKD: data from the ItalKid Project. Archives of Disease in Childhood, 2012, 97, 885-888.	1.0	57
24	Application of the WHOQOL-100 for the assessment of quality of life of adult patients with inherited metabolic diseases. Molecular Genetics and Metabolism, 2012, 106, 25-30.	0.5	20
25	Longâ€term followâ€up results in enzyme replacement therapy for Pompe disease: a case report. Journal of Inherited Metabolic Disease, 2010, 33, 389-393.	1.7	10
26	10p12.1 deletion: HDR phenotype without DGS2 features. Experimental and Molecular Pathology, 2009, 86, 74-76.	0.9	15
27	Is Antibiotic Prophylaxis in Children With Vesicoureteral Reflux Effective in Preventing Pyelonephritis and Renal Scars? A Randomized, Controlled Trial. Pediatrics, 2008, 121, e1489-e1494.	1.0	318
28	Behavioral Therapy for Primary Nocturnal Enuresis. Journal of Urology, 2004, 171, 408-410.	0.2	23