Michela Barbaro

List of Publications by Year
in descending order

16	549	1040056 9	940533 16
papers	citations	h-index	g-index

16	16	16	1204
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Newborn Screening for Severe Primary Immunodeficiency Diseases in Swedenâ€"a 2-Year Pilot TREC and KREC Screening Study. Journal of Clinical Immunology, 2017, 37, 51-60.	3.8	123
2	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
3	Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€"dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	3.2	94
4	Characterization of deletions at 9p affecting the candidate regions for sex reversal and deletion 9p syndrome by MLPA. European Journal of Human Genetics, 2009, 17, 1439-1447.	2.8	73
5	Epigenetic Alterations Associated With Early Prenatal Dexamethasone Treatment. Journal of the Endocrine Society, 2019, 3, 250-263.	0.2	34
6	In vitro functional studies of rare <scp>CYP</scp>21A2 mutations and establishment of an activity gradient for nonclassic mutations improve phenotype predictions in congenital adrenal hyperplasia. Clinical Endocrinology, 2015, 82, 37-44.	2.4	22
7	Biotin and Thiamine Responsive Basal Ganglia Disease â€" A vital differential diagnosis in infants with severe encephalopathy. European Journal of Paediatric Neurology, 2016, 20, 457-461.	1.6	18
8	Multigeneration Inheritance through Fertile XX Carriers of anNR0B1(DAX1) Locus Duplication in a Kindred of Females with Isolated XY Gonadal Dysgenesis. International Journal of Endocrinology, 2012, 2012, 1-7.	1.5	17
9	Diagnostic pitfalls in vitamin B6â€dependent epilepsy caused by mutations in the PLPBP gene. JIMD Reports, 2019, 50, 1-8.	1.5	16
10	Newborn Screening for Primary Immune Deficiencies with a TREC/KREC/ACTB Triplex Assayâ€"A Three-Year Pilot Study in Sweden. International Journal of Neonatal Screening, 2017, 3, 11.	3.2	9
11	Functional and Structural Consequences of NineCYP21A2Mutations Ranging from Very Mild to Severe Effects. International Journal of Endocrinology, 2016, 2016, 1-10.	1.5	8
12	First Year of TREC-Based National SCID Screening in Sweden. International Journal of Neonatal Screening, 2021, 7, 59.	3.2	8
13	Partial protoporphyrinogen oxidase (PPOX) gene deletions, due to different Alu-mediated mechanisms, identified by MLPA analysis in patients with variegate porphyria. Orphanet Journal of Rare Diseases, 2013, 8, 13.	2.7	5
14	Very longâ€chain <scp>acylâ€CoA</scp> dehydrogenase deficiency in a Swedish cohort: Clinical symptoms, newborn screening, enzyme activity, and genetics. JIMD Reports, 2022, 63, 181-190.	1.5	3
15	Novel non-classic CYP21A2 variants, including combined alleles, identified in patients with congenital adrenal hyperplasia. Clinical Biochemistry, 2019, 73, 50-56.	1.9	2
16	Genome-wide investigation of DNA methylation in congenital adrenal hyperplasia. Journal of Steroid Biochemistry and Molecular Biology, 2020, 201, 105699.	2.5	1