Mafalda Mucciolo

List of Publications by Year
in descending order

27 papers	1,226 citations	759233 12 h-index	552781 26 g-index
28 all docs	28 docs citations	28 times ranked	3230 citing authors

#	Article	IF	Citations
1	Sulfonylurea-Insensitive Permanent Neonatal Diabetes Caused by a Severe Gain-of-Function Tyr330His Substitution in Kir6.2. Hormone Research in Paediatrics, 2022, 95, 215-223.	1.8	2
2	Unusual Presentation of Denys-Drash Syndrome in a Girl with Undisclosed Consumption of Biotin. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, 13, 347-352.	0.9	2
3	Providing more evidence on LZTR1 variants in Noonan syndrome patients. American Journal of Medical Genetics, Part A, 2020, 182, 409-414.	1.2	16
4	SOS1 mutations in Noonan syndrome: Cardiomyopathies and not only congenital heart defects! Report of six patients including two novel variants and literature review. American Journal of Medical Genetics, Part A, 2019, 179, 2083-2090.	1.2	10
5	Renal Tubular Dysfunction Fully Accounts for Plasma Biochemical Abnormalities in Type 1A Pseudohypoparathyroidism. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 823-826.	3.6	0
6	Uniparental isodisomy of chromosome 1 results in glycogen storage disease type III with profound growth retardation. Molecular Genetics & Genomic Medicine, 2019, 7, e634.	1.2	7
7	Persistent Hypoglycemia in Children: Targeted Gene Panel Improves the Diagnosis of Hypoglycemia Due to Inborn Errors of Metabolism. Journal of Pediatrics, 2018, 202, 272-278.e4.	1.8	11
8	Next-Generation Sequencing Identifies Different Genetic Defects in 2 Patients with Primary Adrenal Insufficiency and Gonadotropin-Independent Precocious Puberty. Hormone Research in Paediatrics, 2018, 90, 203-211.	1.8	11
9	Complete Scrotal Agenesis: New Surgical Approach Using Self-inflating Tissue Expander. Urology, 2018, 112, 169-171.	1.0	2
10	Lipoid congenital adrenal hyperplasia by steroidogenic acute regulatory protein (STAR) gene mutation in an Italian infant: an uncommon cause of adrenal insufficiency. Italian Journal of Pediatrics, 2017, 43, 57.	2.6	9
11	A Genome Wide Copy Number Variations Analysis in Autism Spectrum Disorder (Asd) and Intellectual Disability (Id) in Italian Families. Journal of Genetic Syndromes & Gene Therapy, 2016, 7, .	0.2	3
12	Next Generation Sequencing Approach in a Prenatal Case of Cardio-Facio-Cutaneus Syndrome. International Journal of Molecular Sciences, 2016, 17, 952.	4.1	6
13	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	6.2	53
14	Bone marrow failure and developmental delay caused by mutations in poly(A)-specific ribonuclease (PARN). Journal of Medical Genetics, 2015, 52, 738-748.	3.2	71
15	Bone Marrow Failure and Developmental Delay Caused By Mutations in Poly(A)-Specific Ribonuclease. Blood, 2015, 126, 2404-2404.	1.4	11
16	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	2.9	61
17	9q31.1q31.3 deletion in two patients with similar clinical features: A newly recognized microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2014, 164, 685-690.	1.2	9
18	Overlapping microdeletions involving 15q22.2 narrow the critical region for intellectual disability to NARG2 and RORA. European Journal of Medical Genetics, 2014, 57, 163-168.	1.3	11

#	Article	IF	Citations
19	Interstitial 22q13 deletions not involving SHANK3 gene: A new contiguous gene syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1666-1676.	1.2	49
20	A 600â€…kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. Journal of Medical Genetics, 2012, 49, 660-668.	3.2	251
21	Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. Clinical Genetics, 2012, 82, 248-255.	2.0	126
22	Xq28 duplications including MECP2 in five females: Expanding the phenotype toÂsevere mental retardation. European Journal of Medical Genetics, 2012, 55, 404-413.	1.3	42
23	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
24	Alport syndrome and leiomyomatosis: the first deletion extending beyond COL4A6 intron 2. Pediatric Nephrology, 2011, 26, 717-724.	1.7	27
25	Investigation of modifier genes within copy number variations in Rett syndrome. Journal of Human Genetics, 2011, 56, 508-515.	2.3	25
26	3.2 Mb microdeletion in chromosome 7 bands q22.2â€“q22.3 associated with overgrowth and delayed bone age. European Journal of Medical Genetics, 2010, 53, 168-170.	1.3	16
27	Is HSD17B1 a new sex reversal gene in human?. Molecular and Cellular Endocrinology, 2009, 313, 70-70.	3.2	0