Elena Andreucci

List of Publications by Year
in descending order

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25 papers	579 citations	687363 13 h-index	642732 23 g-index
26 all docs	26 docs citations	26 times ranked	1282 citing authors

#	Article	IF	Citations
1	Loss-of-Function Mutations in PTPN11 Cause Metachondromatosis, but Not Ollier Disease or Maffucci Syndrome. PLoS Genetics, 2011, 7, e1002050.	3.5	104
2	The genetic and clinical spectrum of a large cohort of patients with distal renal tubular acidosis. Kidney International, 2017, 91, 1243-1255.	5.2	79
3	TRPV4 related skeletal dysplasias: a phenotypic spectrum highlighted byclinical, radiographic, and molecular studies in 21 new families. Orphanet Journal of Rare Diseases, 2011, 6, 37.	2.7	43
4	Type A microsatellite instability in pediatric gliomas as an indicator of Turcot syndrome. European Journal of Human Genetics, 2009, 17, 919-927.	2.8	42
5	Rare variants in Toll-like receptor 7 results in functional impairment and downregulation of cytokine-mediated signaling in COVID-19 patients. Genes and Immunity, 2022, 23, 51-56.	4.1	41
6	Inner ear abnormalities in four patients with dRTA and SNHL: clinical and genetic heterogeneity. Pediatric Nephrology, 2009, 24, 2147-2153.	1.7	32
7	Medullary sponge kidney associated with primary distal renal tubular acidosis and mutations of the H+-ATPase genes. Nephrology Dialysis Transplantation, 2009, 24, 2734-2738.	0.7	29
8	Common, low-frequency, rare, and ultra-rare coding variants contribute to COVID-19 severity. Human Genetics, 2022, 141, 147-173.	3.8	22
9	Infrared fluorescent automated detection of thirteen short tandem repeat polymorphisms and one gender-determining system of the CODIS core system. Electrophoresis, 2000, 21, 3564-3570.	2.4	21
10	Spectrum ofPTCH mutations in Italian nevoid basal cell-carcinoma syndrome patients: Identification of thirteen novel alleles. Human Mutation, 2004, 24, 441-441.	2.5	20
11	CHARGEâ€like presentation, craniosynostosis and mild Mowatâ€“Wilson Syndrome diagnosed by recognition of the distinctive facial gestalt in a cohort of 28 new cases. American Journal of Medical Genetics, Part A, 2014, 164, 2557-2566.	1.2	20
12	Spectrum and distribution of MECP2 mutations in 64 Italian Rett syndrome girls: tentative genotype/phenotype correlation. Brain and Development, 2001, 23, S242-S245.	1.1	19
13	Therapeutic implications of novel mutations of the RFX6 gene associated with early-onset diabetes. Pharmacogenomics Journal, 2015, 15, 49-54.	2.0	18
14	Variants of SOS2 are a rare cause of Noonan syndrome with particular predisposition for lymphatic complications. European Journal of Human Genetics, 2021, 29, 51-60.	2.8	17
15	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. Genetics in Medicine, 2021, 23, 1116-1124.	2.4	17
16	Simpsonâ€“Golabiâ€“Behmel syndrome type 1 in a 27â€week macrosomic preterm newborn: The diagnostic value of rib malformations and index nail and finger hypoplasia. American Journal of Medical Genetics, Part A, 2012, 158A, 2245-2249.	1.2	12
17	Prenatal manifestation and management of a mother and child affected by spondyloperipheral dysplasia with a C-propeptide mutation in COL2A1: case report. Orphanet Journal of Rare Diseases, 2011, 6, 7.	2.7	9
18	Expanding the phenotype of Wiedemannâ€Steiner syndrome: Craniovertebral junction anomalies. American Journal of Medical Genetics, Part A, 2020, 182, 2877-2886.	1.2	9

#	Article	IF	Citations
19	Two novel splicing mutations in the SLC45A2 gene cause Oculocutaneous Albinism Type IV by unmasking cryptic splice sites. Journal of Human Genetics, 2015, 60, 467-471.	2.3	5
20	A novel OTX2 gene frameshift mutation in a child with microphthalmia, ectopic pituitary and growth hormone deficiency. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 603-5.	0.9	5
21	Mosaic Segmental and Whole-Chromosome Upd(11)mat in Silver-Russell Syndrome. Genes, 2021, 12, 581.	2.4	5
22	Multiorgan Infiltration by CD8+ T Cells and 1p;16p Translocation in a Patient with Hypogammaglobulinemia and a Reduced Number of B Cells. International Archives of Allergy and Immunology, 2012, 158, 206-210.	2.1	2
23	Prenatal diagnosis of Xâ€linked adrenoleukodystrophy associated with isolated pericardial effusion. Clinical Case Reports (discontinued), 2015, 3, 643-645.	0.5	2
24	Metatropic dysplasia in third trimester of pregnancy and a novel causative variant in the TRPV4 gene. European Journal of Medical Genetics, 2017, 60, 365-368.	1.3	2
25	Twin zygosity studies with the formula from DNA-View's Kinship Module after molecular analyses by polymorphic markers. International Congress Series, 2004, 1261, 452-453.	0.2	1