Maria Teresa Divizia

List of Publications by Year
in descending order

40 papers	1,591 citations	394421 19 h-index	315739 38 g-index
41 all docs	41 docs citations	41 times ranked	2858 citing authors

#	Article	IF	Citations
1	Camurati-Engelmann disease: review of the clinical, radiological, and molecular data of 24 families and implications for diagnosis and treatment. Journal of Medical Genetics, 2005, 43, 1-11.	3.2	211
2	Disruption of Autoregulatory Feedback by a Mutation in a Remote, Ultraconserved PAX6 Enhancer Causes Aniridia. American Journal of Human Genetics, 2013, 93, 1126-1134.	6.2	172
3	Overexpression of the C-type natriuretic peptide (CNP) is associated with overgrowth and bone anomalies in an individual with balanced t(2;7) translocation. Human Mutation, 2007, 28, 724-731.	2.5	118
4	Recurrent microdeletion at 17q12 as a cause of Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome: two case reports. Orphanet Journal of Rare Diseases, 2009, 4, 25.	2.7	105
5	MKS3/TMEM67mutations are a major cause of COACH Syndrome, a Joubert Syndrome related disorder with liver involvement. Human Mutation, 2009, 30, E432-E442.	2.5	96
6	Clinical score of 62 Italian patients with Cornelia de Lange syndrome and correlations with the presence and type of NIPBL mutation. Clinical Genetics, 2007, 72, 98-108.	2.0	93
7	NovelTMEM67mutations and genotype-phenotype correlates in meckelin-related ciliopathies. Human Mutation, 2010, 31, n/a-n/a.	2.5	77
8	Phenotypic spectrum and prevalence of INPP5E mutations in Joubert Syndrome and related disorders. European Journal of Human Genetics, 2013, 21, 1074-1078.	2.8	64
9	Rubinstein-Taybi Syndrome: spectrum of CREBBP mutations in Italian patients. BMC Medical Genetics, 2006, 7, 77.	2.1	60
10	A multi-method approach to the molecular diagnosis of overt and borderline 11p15.5 defects underlying Silverâ€“Russell and Beckwithâ€“Wiedemann syndromes. Clinical Epigenetics, 2016, 8, 23.	4.1	54
11	Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. American Journal of Human Genetics, 2019, 105, 689-705.	6.2	48
12	Microarray based analysis of an inherited terminal 3p26.3 deletion, containing only the CHL1 gene, from a normal father to his two affected children. Orphanet Journal of Rare Diseases, 2011, 6, 12.	2.7	42
13	Malformations following methimazole exposure in utero: An open issue. Birth Defects Research Part A: Clinical and Molecular Teratology, 2003, 67, 989-992.	1.6	39
14	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
15	De novo deletion of chromosome 11q12.3 in monozygotic twins affected by Poland Syndrome. BMC Medical Genetics, 2014, 15, 63.	2.1	32
16	Genotype-Phenotype Correlation of 2q37 Deletions Including NPPC Gene Associated with Skeletal Malformations. PLoS ONE, 2013, 8, e66048.	2.5	32
17	Hand and Upper Limb Anomalies in Poland Syndrome. Journal of Pediatric Orthopaedics, 2012, 32, 722-726.	1.2	29
18	Familial Poland anomaly revisited. American Journal of Medical Genetics, Part A, 2012, 158A, 140-149.	1.2	29

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19	A spectrum of LMX1B mutations in Nail-Patella syndrome: New point mutations, deletion, and evidence of mosaicism in unaffected parents. Genetics in Medicine, 2010, 12, 431-439.	2.4	26
20	Intragenic duplication of KCNQ5 gene results in aberrant splicing leading to a premature termination codon in a patient with intellectual disability. European Journal of Medical Genetics, 2019, 62, 103555.	1.3	22
21	Assessment of copy number variations in 120 patients with Poland syndrome. BMC Medical Genetics, 2016, 17, 89.	2.1	20
22	EEC- and ADULT-AssociatedTP63Mutations Exhibit Functional Heterogeneity Toward P63 Responsive Sequences. Human Mutation, 2013, 34, 894-904.	2.5	19
23	The First Intron of the Human Osteopontin Gene Contains a C/EBP-Beta-Responsive Enhancer. Gene Expression, 2003, 11, 95-104.	1.2	18
24	Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet Journal of Rare Diseases, 2020, 15, 201.	2.7	17
25	Thrombocytopenia-absent radius (TAR) syndrome due to compound inheritance for a 1q21.1 microdeletion and a low-frequency noncoding RBM8A SNP: a new familial case. Molecular Cytogenetics, 2015, 8, 87.	0.9	16
26	Pituitary hypoplasia and growth hormone deficiency in Coffinâ€Siris syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 384-388.	1.2	15
27	A polymorphic variant inside the osteopontin gene shows association with disease course in oligoarticular juvenile idiopathic arthritis. Annals of the Rheumatic Diseases, 2006, 65, 662-665.	0.9	14
28	Craniosynostosis, hydrocephalus, Chiari I malformation and radioulnar synostosis: Probably a new syndrome. European Journal of Medical Genetics, 2009, 52, 17-22.	1.3	13
29	Neurodevelopmental Disorders in Patients With Complex Phenotypes and Potential Complex Genetic Basis Involving Non-Coding Genes, and Double CNVs. Frontiers in Genetics, 2021, 12, 732002.	2.3	12
30	Novel mutations in the L1CAM gene support the complexity of L1 syndrome. Journal of the Neurological Sciences, 2010, 294, 124-126.	0.6	10
31	How wide is the ocular spectrum of Delleman syndrome?. Clinical Dysmorphology, 2004, 13, 33-34.	0.3	8
32	Case Report: Whole Exome Sequencing Revealed Disease-Causing Variants in Two Genes in a Patient With Autism Spectrum Disorder, Intellectual Disability, Hyperactivity, Sleep and Gastrointestinal Disturbances. Frontiers in Genetics, 2021, 12, 625564.	2.3	8
33	Recurrence and Familial Inheritance of Intronic NIPBL Pathogenic Variant Associated With Mild CdLS. Frontiers in Neurology, 2018, 9, 967.	2.4	6
34	Auriculo-condylar syndrome or new syndrome?. Clinical Dysmorphology, 2002, 11, 143-144.	0.3	5
35	A novel Xp22.11 deletion causing a syndrome of craniosynostosis and periventricular nodular heterotopia. American Journal of Medical Genetics, Part A, 2011, 155, 3144-3147.	1.2	5
36	P63 modulates the expression of the WDFY2 gene which is implicated in cancer regulation and limb development. Bioscience Reports, 2019, 39, .	2.4	5

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37	Arrayâ€CGH analysis in a patient with WAGR syndrome and a reciprocal translocation t(2;11) inherited from the normal father with double translocation. American Journal of Medical Genetics, Part A, 2010, 152A, 2130-2133.	1.2	4
38	Renal involvement and StrÃ¸mme syndrome. CKJ: Clinical Kidney Journal, 2021, 14, 439-441.	2.9	3
39	Unilateral radio-ulnar synostosis associated with hypotonia, developmental delay, and facial dysmorphism. American Journal of Medical Genetics, Part A, 2005, 137A, 106-108.	1.2	1
40	Further case of metaphyseal acroscyphodysplasia with cone-shaped epiphyses (Bellini disease or) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 6	0.3	1