Margherita Lerone

List of Publications by Year
in descending order

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95 papers	3,916 citations	172457 29 h-index	128289 60 g-index
97 all docs	97 docs citations	97 times ranked	3392 citing authors

#	Article	IF	Citations
1	Point mutations affecting the tyrosine kinase domain of the RET proto-oncogene in Hirschsprung's disease. Nature, 1994, 367, 377-378.	27.8	722
2	Different TBX5 interactions in heart and limb defined by Holt-Oram syndrome mutations. Proceedings of the National Academy of Sciences of the United States of America, 1999, 96, 2919-2924.	7.1	354
3	A gene for Hirschsprung disease maps to the proximal long arm of chromosome 10. Nature Genetics, 1993, 4, 346-350.	21.4	190
4	Common Mutations in Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy Patients of Different Origins. Molecular Endocrinology, 1998, 12, 1112-1119.	3.7	150
5	Frequency of RET mutations in long- and short-segment Hirschsprung disease. Human Mutation, 1997, 9, 243-249.	2.5	138
6	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
7	Genetic Mapping to 10q23.3-q24.2, in a Large Italian Pedigree, of a New Syndrome Showing Bilateral Cataracts, Gastroesophageal Reflux, and Spastic Paraparesis with Amyotrophy. American Journal of Human Genetics, 1999, 64, 586-593.	6.2	108
8	Special basic science review. Journal of Pediatric Surgery, 2000, 35, 1017-1025.	1.6	105
9	Currarino syndrome: Proposal of a diagnostic and therapeutic protocol. Journal of Pediatric Surgery, 2004, 39, 1305-1311.	1.6	105
10	Ectodermal dysplasias: not only â€˜skinâ€™ deep. Clinical Genetics, 2000, 58, 415-430.	2.0	85
11	Association of multiple endocrine neoplasia type 2 and Hirschsprung disease. Journal of Internal Medicine, 1998, 243, 515-520.	6.0	74
12	Total colonic aganglionosis associated with interstitial deletion of the long arm of chromosome 10. Pediatric Surgery International, 1992, 7, 308.	1.4	70
13	In vitro efficacy of ARQ 092, an allosteric AKT inhibitor, on primary fibroblast cells derived from patients with PIK3CA-related overgrowth spectrum (PROS). Neurogenetics, 2018, 19, 77-91.	1.4	65
14	Poland syndrome with bilateral features: Case description with review of the literature. American Journal of Medical Genetics, Part A, 2009, 149A, 1597-1602.	1.2	62
15	Heterogeneity and Low Detection Rate of RET Mutations in Hirschsprung Disease. European Journal of Human Genetics, 1994, 2, 272-280.	2.8	60
16	Severe end of Opitz trigonocephaly (C) syndrome or new syndrome?. , 1999, 85, 438-446.		56
17	Associated anomalies in intestinal neuronal dysplasia. Journal of Pediatric Surgery, 2002, 37, 219-223.	1.6	53
18	Single nucleotide polymorphic alleles in the 5' region of the RET proto-oncogene define a risk haplotype in Hirschsprung's disease. Journal of Medical Genetics, 2003, 40, 714-718.	3.2	50

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19	Identification of TBX5 mutations in a series of 94 patients with Tetralogy of Fallot. American Journal of Medical Genetics, Part A, 2014, 164, 3100-3107.	1.2	47
20	Dextrocardia in patients with Poland syndrome: Phenotypic characterization provides insight into the pathogenesis. Journal of Thoracic and Cardiovascular Surgery, 2010, 139, 1177-1182.	0.8	43
21	Variants of the ACTG2 gene correlate with degree of severity and presence of megacystis in chronic intestinal pseudo-obstruction. European Journal of Human Genetics, 2016, 24, 1211-1215.	2.8	43
22	Neuronal Intestinal Dysplasia: Clinical Experience in Italian Patients. European Journal of Pediatric Surgery, 1994, 4, 287-292.	1.3	42
23	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
24	Leiomyomatosis of oesophagus, congenital cataracts and hematuria. Pediatric Radiology, 1991, 21, 578-579.	2.0	41
25	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
26	Rett syndrome: exclusion mapping following the hypothesis of germinal mosaicism for new X-linked mutations. Human Genetics, 1991, 86, 604-6.	3.8	37
27	Evaluation of the HOX11L1 gene as a candidate for congenital disorders of intestinal innervation. Journal of Medical Genetics, 2000, 37, 9e-9.	3.2	36
28	ICF syndrome with variable expression in sibs.. Journal of Medical Genetics, 1993, 30, 429-432.	3.2	34
29	Previously undescribed nonsense mutation in SHH caused autosomal dominant holoprosencephaly with wide intrafamilial variability. , 2003, 117A, 112-115.		32
30	De novo deletion of chromosome 11q12.3 in monozygotic twins affected by Poland Syndrome. BMC Medical Genetics, 2014, 15, 63.	2.1	32
31	Genotype-Phenotype Correlation of 2q37 Deletions Including NPPC Gene Associated with Skeletal Malformations. PLoS ONE, 2013, 8, e66048.	2.5	32
32	Diagnostic and therapeutic approach to multiple endocrine neoplasia type 2B in pediatric patients. Pediatric Surgery International, 2002, 18, 378-383.	1.4	29
33	Hand and Upper Limb Anomalies in Poland Syndrome. Journal of Pediatric Orthopaedics, 2012, 32, 722-726.	1.2	29
34	Familial Poland anomaly revisited. American Journal of Medical Genetics, Part A, 2012, 158A, 140-149.	1.2	29
35	Ectodermal abnormalities in Kabuki syndrome. , 1997, 73, 263-266.		28
36	Inheritance of Niikawa-Kuroki (Kabuki makeup) syndrome. , 1996, 66, 368-368.		26

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37	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
38	Molecular characterization of a t(2;6) balanced translocation that is associated with a complex phenotype and leads to truncation of theTCBA1gene. Human Mutation, 2005, 26, 426-436.	2.5	25
39	Common PHOX2B poly-alanine contractions impair RET gene transcription, predisposing to Hirschsprung disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 1770-1777.	3.8	25
40	Exclusion of the Sonic Hedgehog gene as responsible for Currarino syndrome and anorectal malformations with sacral hypodevelopment. Human Genetics, 1999, 104, 108-110.	3.8	23
41	Sonographic and molecular diagnosis of thanatophoric dysplasia type I at 18 weeks of gestation. Prenatal Diagnosis, 2000, 20, 835-837.	2.3	23
42	Molecular characterisation of a supernumerary ring chromosome in a patient with VATER association. Journal of Medical Genetics, 2001, 38, 6e-6.	3.2	23
43	Question mark ears, temporo-mandibular joint malformation and hypotonia: auriculo-condylar syndrome or a distinct entity?. Clinical Dysmorphology, 2000, 9, 277-280.	0.3	22
44	Transcriptional regulation of TLX2 and impaired intestinal innervation: possible role of the PHOX2A and PHOX2B genes. European Journal of Human Genetics, 2007, 15, 848-855.	2.8	22
45	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
46	HOX11L1: a promoter study to evaluate possible expression defects in intestinal motility disorders. International Journal of Molecular Medicine, 2002, 10, 101-6.	4.0	22
47	Startle disease in an Italian family by mutation (K276E): The Î±-subunit of the inhibiting glycine receptor. Human Mutation, 1997, 9, 185-187.	2.5	21
48	Genotype-phenotype correlation and germline mosaicism in DMD/BMD patients with deletions of the dystrophin gene. Human Genetics, 1991, 87, 353-60.	3.8	20
49	Uncombable hair, retinal pigmentary distrophy, dental anomalies, and brachydactyly: Report of a new patient with additional findings. American Journal of Medical Genetics Part A, 1993, 47, 931-933.	2.4	20
50	Lower extremity counterpart of the Poland syndrome. Clinical Genetics, 1999, 55, 41-43.	2.0	20
51	HLXB9 homeobox gene and caudal regression syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 205-209.	1.6	20
52	Multiple endocrine neoplasias type 2B and RET proto-oncogene. Italian Journal of Pediatrics, 2012, 38, 9.	2.6	20
53	Assessment of copy number variations in 120 patients with Poland syndrome. BMC Medical Genetics, 2016, 17, 89.	2.1	20
54	EEC- and ADULT-AssociatedTP63Mutations Exhibit Functional Heterogeneity Toward P63 Responsive Sequences. Human Mutation, 2013, 34, 894-904.	2.5	19

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55	Deleted and normal chromosome 10 homologs from a patient with Hirschsprung disease isolated in two cell hybrids through enrichment by immunomagnetic selection. Cytogenetic and Genome Research, 1993, 63, 102-106.	1.1	18
56	Mutational analysis of theRNX gene in congenital central hypoventilation syndrome. American Journal of Medical Genetics Part A, 2002, 113, 178-182.	2.4	17
57	Oculocerebral syndrome with hypopigmentation (Cross syndrome): report of a new case. Clinical Genetics, 1992, 41, 87-89.	2.0	17
58	Clinical and molecular characterization of a patient with interstitial 6q21q22.1 deletion. Molecular Cytogenetics, 2015, 8, 31.	0.9	17
59	Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet Journal of Rare Diseases, 2020, 15, 201.	2.7	17
60	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
61	Pituitary hypoplasia and growth hormone deficiency in Coffinâ€Siris syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 384-388.	1.2	15
62	Search for pathogenetic variants of the SPRY2 gene in intestinal innervation defects. Internal Medicine Journal, 2009, 39, 335-337.	0.8	14
63	Congenital diaphragmatic hernia associated with ipsilateral upper limb reduction defects: Report of a case with thumb hypoplasia. American Journal of Medical Genetics Part A, 1992, 44, 827-829.	2.4	13
64	PREVENTION OF THALASSAEMIA MAJOR IN LATIUM (ITALY). Lancet, The, 1985, 326, 888-889.	13.7	12
65	Pfeiffer syndrome type 2 associated with a single amino acid deletion in the PGFR2 gene. Clinical Genetics, 2000, 58, 81-83.	2.0	12
66	Novel ACTG2 variants disclose allelic heterogeneity and biâ€allelic inheritance in pediatric chronic intestinal pseudoâ€obstruction. Clinical Genetics, 2021, 99, 430-436.	2.0	12
67	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
68	Autosomal recessive microcephaly with early onset seizures and spasticity. Clinical Genetics, 1992, 42, 152-155.	2.0	11
69	New clinical findings in oculo-ectodermal syndrome. Clinical Dysmorphology, 2000, 9, 39-41.	0.3	10
70	Fontaine-farriaux craniosynostosis: Second report in the literature. American Journal of Medical Genetics Part A, 2001, 100, 214-218.	2.4	10
71	Constitutional 3p26.3 terminal microdeletion in an adolescent with neuroblastoma. Cancer Biology and Therapy, 2017, 18, 285-289.	3.4	10
72	Molecular Genetics in Neuroblastoma Prognosis. Children, 2021, 8, 456.	1.5	10

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73	A new syndrome with cerebro-oculo-skeletal-renal involvement. Pediatric Radiology, 1990, 20, 612-614.	2.0	9
74	Total Anonychia congenita in a Woman with Normal Intelligence: Report of a Further Case. Dermatology, 2000, 200, 84-85.	2.1	9
75	How wide is the ocular spectrum of Delleman syndrome?. Clinical Dysmorphology, 2004, 13, 33-34.	0.3	8
76	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
77	Auriculo-condylar syndrome or new syndrome?. Clinical Dysmorphology, 2002, 11, 143-144.	0.3	5
78	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
79	P63 modulates the expression of the WDFY2 gene which is implicated in cancer regulation and limb development. Bioscience Reports, 2019, 39, .	2.4	5
80	HB Siirt [Î²27(Î²9)ALAâ†'GLY]: A New, Electrophoretically Silent, Hemoglobin Variant. Hemoglobin, 1997, 21, 495-497.	0.8	4
81	Ectodermal dysplasia, primary hypothyroidism, and agenesis of the corpus callosum: variable expression of a single syndrome?. Journal of Medical Genetics, 1998, 35, 157-158.	3.2	4
82	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
83	Renal involvement and StrÃ¸mme syndrome. CKJ: Clinical Kidney Journal, 2021, 14, 439-441.	2.9	3
84	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	2.5	2
85	Response to Klinger and Merlob re: Case description with review of the literature. Am J Med Genet Part A 149A:1597â€"1602, 2009. American Journal of Medical Genetics, Part A, 2009, 149A, 2899-2899.	1.2	2
86	Multiple sutural synostosis and congenital cataracts. Human Genetics, 1991, 87, 758.	3.8	1
87	Correspondence. Clinical Genetics, 1999, 56, 176-176.	2.0	1
88	HOX11L1: a promoter study to evaluate possible expression defects in intestinal motility disorders. International Journal of Molecular Medicine, 2002, 10, 101.	4.0	1
89	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
90	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

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91	A t(7;12) balanced translocation with breakpoints overlapping those of the Williamsâ€“Beuren and 12q14 microdeletion syndromes. American Journal of Medical Genetics, Part A, 2010, 152A, 1285-1294.	1.2	1
92	Inherited diseases of the connective tissue. Current Opinion in Pediatrics, 1989, 1, 465-468.	2.0	0
93	Congenital hypoplastic anaemia in a patient with a new multiple congenital anomalies-mental retardation syndrome. , 1999, 87, 36-39.		0
94	Diagnostic Criteria of Pediatric Intestinal Myopathies. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, 383-386.	1.8	0
95	Fontaineâ€farriaux craniosynostosis: Second report in the literature. American Journal of Medical Genetics Part A, 2001, 100, 214-218.	2.4	0