Margherita Lerone

List of Publications by Year
in descending order

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Version: 2024-02-01

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	Renal involvement and StrÃ¸mme syndrome. CKJ: Clinical Kidney Journal, 2021, 14, 439-441.	2.9	3
2	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
3	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
4	Molecular Genetics in Neuroblastoma Prognosis. Children, 2021, 8, 456.	1.5	10
5	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
6	Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet Journal of Rare Diseases, 2020, 15, 201.	2.7	17
7	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
8	P63 modulates the expression of the WDFY2 gene which is implicated in cancer regulation and limb development. Bioscience Reports, 2019, 39, .	2.4	5
9	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
10	Diagnostic Criteria of Pediatric Intestinal Myopathies. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, 383-386.	1.8	0
11	Constitutional 3p26.3 terminal microdeletion in an adolescent with neuroblastoma. Cancer Biology and Therapy, 2017, 18, 285-289.	3.4	10
12	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
13	Assessment of copy number variations in 120 patients with Poland syndrome. BMC Medical Genetics, 2016, 17, 89.	2.1	20
14	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
15	Clinical and molecular characterization of a patient with interstitial 6q21q22.1 deletion. Molecular Cytogenetics, 2015, 8, 31.	0.9	17
16	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
17	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
18	De novo deletion of chromosome 11q12.3 in monozygotic twins affected by Poland Syndrome. BMC Medical Genetics, 2014, 15, 63.	2.1	32

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19	EEC- and ADULT-AssociatedTP63Mutations Exhibit Functional Heterogeneity Toward P63 Responsive Sequences. Human Mutation, 2013, 34, 894-904.	2.5	19
20	Genotype-Phenotype Correlation of 2q37 Deletions Including NPPC Gene Associated with Skeletal Malformations. PLoS ONE, 2013, 8, e66048.	2.5	32
21	Hand and Upper Limb Anomalies in Poland Syndrome. Journal of Pediatric Orthopaedics, 2012, 32, 722-726.	1.2	29
22	Multiple endocrine neoplasias type 2B and RET proto-oncogene. Italian Journal of Pediatrics, 2012, 38, 9.	2.6	20
23	Familial Poland anomaly revisited. American Journal of Medical Genetics, Part A, 2012, 158A, 140-149.	1.2	29
24	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
25	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
26	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
27	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
28	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
29	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
30	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
31	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
32	Search for pathogenetic variants of the SPRY2 gene in intestinal innervation defects. Internal Medicine Journal, 2009, 39, 335-337.	0.8	14
33	Pituitary hypoplasia and growth hormone deficiency in Coffinâ€Siris syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 384-388.	1.2	15
34	Further case of metaphyseal acroscyphodysplasia with cone-shaped epiphyses (Bellini disease or) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 1	0.3	1
35	The Italian XLMR bank: a clinical and molecular database. Human Mutation, 2007, 28, 13-18.	2.5	2
36	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

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37	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
38	HLXB9 homeobox gene and caudal regression syndrome. Birth Defects Research Part A: Clinical and Molecular Teratology, 2006, 76, 205-209.	1.6	20
39	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
40	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
41	Currarino syndrome: Proposal of a diagnostic and therapeutic protocol. Journal of Pediatric Surgery, 2004, 39, 1305-1311.	1.6	105
42	How wide is the ocular spectrum of Delleman syndrome?. Clinical Dysmorphology, 2004, 13, 33-34.	0.3	8
43	Previously undescribed nonsense mutation in SHH caused autosomal dominant holoprosencephaly with wide intrafamilial variability. , 2003, 117A, 112-115.		32
44	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
45	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
46	HOX11L1: a promoter study to evaluate possible expression defects in intestinal motility disorders. International Journal of Molecular Medicine, 2002, 10, 101.	4.0	1
47	Auriculo-condylar syndrome or new syndrome?. Clinical Dysmorphology, 2002, 11, 143-144.	0.3	5
48	Associated anomalies in intestinal neuronal dysplasia. Journal of Pediatric Surgery, 2002, 37, 219-223.	1.6	53
49	Mutational analysis of theRNX gene in congenital central hypoventilation syndrome. American Journal of Medical Genetics Part A, 2002, 113, 178-182.	2.4	17
50	Diagnostic and therapeutic approach to multiple endocrine neoplasia type 2B in pediatric patients. Pediatric Surgery International, 2002, 18, 378-383.	1.4	29
51	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
52	Fontaine-farriaux craniosynostosis: Second report in the literature. American Journal of Medical Genetics Part A, 2001, 100, 214-218.	2.4	10
53	Molecular characterisation of a supernumerary ring chromosome in a patient with VATER association. Journal of Medical Genetics, 2001, 38, 6e-6.	3.2	23
54	Fontaineâ€farriaux craniosynostosis: Second report in the literature. American Journal of Medical Genetics Part A, 2001, 100, 214-218.	2.4	0

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55	New clinical findings in oculo-ectodermal syndrome. Clinical Dysmorphology, 2000, 9, 39-41.	0.3	10
56	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
57	Sonographic and molecular diagnosis of thanatophoric dysplasia type I at 18 weeks of gestation. Prenatal Diagnosis, 2000, 20, 835-837.	2.3	23
58	Pfeiffer syndrome type 2 associated with a single amino acid deletion in the PGFR2 gene. Clinical Genetics, 2000, 58, 81-83.	2.0	12
59	Ectodermal dysplasias: not only â€˜skinâ€™ deep. Clinical Genetics, 2000, 58, 415-430.	2.0	85
60	Total Anonychia congenita in a Woman with Normal Intelligence: Report of a Further Case. Dermatology, 2000, 200, 84-85.	2.1	9
61	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
62	Special basic science review. Journal of Pediatric Surgery, 2000, 35, 1017-1025.	1.6	105
63	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
64	Lower extremity counterpart of the Poland syndrome. Clinical Genetics, 1999, 55, 41-43.	2.0	20
65	Correspondence. Clinical Genetics, 1999, 56, 176-176.	2.0	1
66	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
67	Severe end of Opitz trigonocephaly (C) syndrome or new syndrome?. , 1999, 85, 438-446.		56
68	Congenital hypoplastic anaemia in a patient with a new multiple congenital anomalies-mental retardation syndrome. , 1999, 87, 36-39.		0
69	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
70	Association of multiple endocrine neoplasia type 2 and Hirschsprung disease. Journal of Internal Medicine, 1998, 243, 515-520.	6.0	74
71	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
72	Common Mutations in Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy Patients of Different Origins. Molecular Endocrinology, 1998, 12, 1112-1119.	3.7	150

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73	HB Siirt [Î²27(Î²9)ALAâ†'GLY]: A New, Electrophoretically Silent, Hemoglobin Variant. Hemoglobin, 1997, 21, 495-497.	0.8	4
74	Ectodermal abnormalities in Kabuki syndrome. , 1997, 73, 263-266.		28
75	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
76	Frequency of RET mutations in long- and short-segment Hirschsprung disease. Human Mutation, 1997, 9, 243-249.	2.5	138
77	Inheritance of Niikawa-Kuroki (Kabuki makeup) syndrome. , 1996, 66, 368-368.		26
78	Neuronal Intestinal Dysplasia: Clinical Experience in Italian Patients. European Journal of Pediatric Surgery, 1994, 4, 287-292.	1.3	42
79	Point mutations affecting the tyrosine kinase domain of the RET proto-oncogene in Hirschsprung's disease. Nature, 1994, 367, 377-378.	27.8	722
80	Heterogeneity and Low Detection Rate of RET Mutations in Hirschsprung Disease. European Journal of Human Genetics, 1994, 2, 272-280.	2.8	60
81	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
82	A gene for Hirschsprung disease maps to the proximal long arm of chromosome 10. Nature Genetics, 1993, 4, 346-350.	21.4	190
83	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
84	ICF syndrome with variable expression in sibs.. Journal of Medical Genetics, 1993, 30, 429-432.	3.2	34
85	Total colonic aganglionosis associated with interstitial deletion of the long arm of chromosome 10. Pediatric Surgery International, 1992, 7, 308.	1.4	70
86	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
87	Autosomal recessive microcephaly with early onset seizures and spasticity. Clinical Genetics, 1992, 42, 152-155.	2.0	11
88	Oculocerebral syndrome with hypopigmentation (Cross syndrome): report of a new case. Clinical Genetics, 1992, 41, 87-89.	2.0	17
89	Leiomyomatosis of oesophagus, congenital cataracts and hematuria. Pediatric Radiology, 1991, 21, 578-579.	2.0	41
90	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

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91	Rett syndrome: exclusion mapping following the hypothesis of germinal mosaicism for new X-linked mutations. Human Genetics, 1991, 86, 604-6.	3.8	37
92	Multiple sutural synostosis and congenital cataracts. Human Genetics, 1991, 87, 758.	3.8	1
93	A new syndrome with cerebro-oculo-skeletal-renal involvement. Pediatric Radiology, 1990, 20, 612-614.	2.0	9
94	Inherited diseases of the connective tissue. Current Opinion in Pediatrics, 1989, 1, 465-468.	2.0	0
95	PREVENTION OF THALASSAEMIA MAJOR IN LATIUM (ITALY). Lancet, The, 1985, 326, 888-889.	13.7	12