Mara Giordano

List of Publications by Year
in descending order

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78	2,030	257450	265206
papers	citations	24	42
		h-index	g-index

80	80	80	2831
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Copy number variations residing outside the SHOX enhancer region are involved in Short Stature and LÃ©riâ€Weill dyschondrosteosis. Molecular Genetics & Genomic Medicine, 2022, 10, e1793.	1.2	4
2	Variants in the 5â€²UTR reduce SHOX expression and contribute to SHOX haploinsufficiency. European Journal of Human Genetics, 2021, 29, 110-121.	2.8	12
3	Screening for haemoglobin disorders: The experience of the piedmont northâ€eastern quadrant. International Journal of Laboratory Hematology, 2021, 43, e61-e63.	1.3	0
4	The Prevalence of Thyroid Autoimmunity in Children with Developmental Dyslexia. BioMed Research International, 2021, 2021, 1-5.	1.9	0
5	Retrospective Diagnosis of a Novel ACAN Pathogenic Variant in a Family With Short Stature: A Case Report and Review of the Literature. Frontiers in Genetics, 2021, 12, 708864.	2.3	5
6	Consumption of complement in a 26-year-old woman with severe thrombotic thrombocytopenia after ChAdOx1 nCov-19 vaccination. Journal of Autoimmunity, 2021, 124, 102728.	6.5	5
7	Circulating Platelet-Derived Extracellular Vesicles Are a Hallmark of Sars-Cov-2 Infection. Cells, 2021, 10, 85.	4.1	87
8	A Long Contiguous Stretch of Homozygosity Disclosed a Novel STAG3 Biallelic Pathogenic Variant Causing Primary Ovarian Insufficiency: A Case Report and Review of the Literature. Genes, 2021, 12, 1709.	2.4	7
9	Long-term sequelae are highly prevalent one year after hospitalization for severe COVID-19. Scientific Reports, 2021, 11, 22666.	3.3	84
10	Metabolomics Diagnosis of COVID-19 from Exhaled Breath Condensate. Metabolites, 2021, 11, 847.	2.9	22
11	The first case of the TARDBP p.G294V mutation in a homozygous state: is a single pathogenic allele sufficient to cause ALS?. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 273-279.	1.7	10
12	Haptoglobin Phenotypes Are Associated with the Postload Glucose and Insulin Levels in Pediatric Obesity. International Journal of Endocrinology, 2020, 2020, 1-8.	1.5	3
13	Hereditary Deficiency of the Second Component of Complement: Early Diagnosis and 21-Year Follow-Up of a Family. Medicina (Lithuania), 2020, 56, 120.	2.0	0
14	Identification and functional characterization of a novel splicing variant in the F8 coagulation gene causing severe hemophilia A. Journal of Thrombosis and Haemostasis, 2020, 18, 1050-1064.	3.8	2
15	Identification of Haptoglobin as a Readout of rhGH Therapy in GH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 5263-5273.	3.6	3
16	Ovotesticular Disorder of Sex Development: A Rare Case of Lateral Subtype 45X/46XY kariotype Diagnosed in Adulthood. Urology, 2019, 129, 68-70.	1.0	3
17	Co-occurrence of genomic imbalances on Xp22.1 in the SHOX region and 15q25.2 in a girl with short stature, precocious puberty, urogenital malformations and bone anomalies. BMC Medical Genomics, 2019, 12, 5.	1.5	11
18	Improving clinical diagnosis in SHOX deficiency: the importance of growth velocity. Pediatric Research, 2018, 83, 438-444.	2.3	11

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19	Novel GLI 2 mutations identified in patients with Combined Pituitary Hormone Deficiency (CPHD): evidence for a pathogenic effect by functional characterization. Clinical Endocrinology, 2018, 90, 449-456.	2.4	17
20	An intragenic deletion within CTNNA2 intron 7 in a boy with short stature and speech delay: A case report. SAGE Open Medical Case Reports, 2017, 5, 2050313X1769396.	0.3	0
21	Genetic variations at the human growth hormone receptor (GHR) gene locus are associated with idiopathic short stature. Journal of Cellular and Molecular Medicine, 2017, 21, 2985-2999.	3.6	19
22	Copy number variants analysis in a cohort of isolated and syndromic developmental delay/intellectual disability reveals novel genomic disorders, position effects and candidate disease genes. Clinical Genetics, 2017, 92, 415-422.	2.0	43
23	Invasive meningococcal disease in three siblings with hereditary deficiency of the 8th component of complement: evidence for the importance of an early diagnosis. Orphanet Journal of Rare Diseases, 2016, 11, 64.	2.7	9
24	A 18p11.23-p11.31 microduplication in a boy with psychomotor delay, cerebellar vermis hypoplasia, chorioretinal coloboma, deafness and GH deficiency. Molecular Cytogenetics, 2016, 9, 89.	0.9	12
25	Genetic causes of isolated and combined pituitary hormone deficiency. Best Practice and Research in Clinical Endocrinology and Metabolism, 2016, 30, 679-691.	4.7	53
26	Testing for the cytosine insertion in the VNTR of the MUC1 gene in a cohort of Italian patients with autosomal dominant tubulointerstitial kidney disease. Journal of Nephrology, 2016, 29, 451-455.	2.0	10
27	Variations in the high-mobility group-A2 gene (HMGA2) are associated with idiopathic short stature. Pediatric Research, 2016, 79, 258-261.	2.3	4
28	A 5.8ÂMb interstitial deletion on chromosome Xq21.1 in a boy with intellectual disability, cleft palate, hearing impairment and combined growth hormone deficiency. BMC Medical Genetics, 2015, 16, 74.	2.1	13
29	Frequency of genetic defects in combined pituitary hormone deficiency: a systematic review and analysis of a multicentre Italian cohort. Clinical Endocrinology, 2015, 83, 849-860.	2.4	57
30	Hypomagnesemia and progressive chronic kidney disease: thinking of HNF1B and other genetic nephropathies. Kidney International, 2015, 88, 641.	5.2	2
31	Effects of Growth Hormone (GH) Therapy Withdrawal on Glucose Metabolism in Not Confirmed GH Deficient Adolescents at Final Height. PLoS ONE, 2014, 9, e87157.	2.5	16
32	Chronic renal failure of unknown origin is caused by <scp>HNF1B</scp> mutations in 9% of adult patients: A single centre cohort analysis. Nephrology, 2014, 19, 202-209.	1.6	16
33	Novel Mutations in the GH Gene (GH1) Uncover Putative Splicing Regulatory Elements. Endocrinology, 2014, 155, 1786-1792.	2.8	8
34	Unexpectedly high prevalence of rare genetic disorders in kidney transplant recipients with an unknown causal nephropathy. Clinical Transplantation, 2014, 28, 995-1003.	1.6	26
35	Genetic Variants Associated with Increased Risk of Malignant Pleural Mesothelioma: A Genome-Wide Association Study. PLoS ONE, 2013, 8, e61253.	2.5	71
36	The W520X mutation in the TSHR gene brings on subclinical hypothyroidism through an haploinsufficiency mechanism. Journal of Endocrinological Investigation, 2013, 36, 716-21.	3.3	3

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37	Dielsâ€“Alder Reactions of 2-Vinylindoles with Cyclic Dienophiles: Synthesis of [c]-Annulated Tetrahydrocarbazoles. Synlett, 2012, 23, 2913-2918.	1.8	13
38	Functional SNPs within the Intron 1 of the PROP1 Gene Contribute to Combined Growth Hormone Deficiency (CPHD). Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1791-E1797.	3.6	3
39	Association of the (CA)_n repeat polymorphism of insulinâ€like growth factorâ€I and â^'202 A/C IGFâ€binding proteinâ€3 promoter polymorphism with adult height in patients with severe growth hormone deficiency. Clinical Endocrinology, 2012, 76, 683-690.	2.4	11
40	XRCC1 and ERCC1 variants modify malignant mesothelioma risk: A caseâ€“control study. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2011, 708, 11-20.	1.0	34
41	A novel HESX1 splice mutation causes isolated GH deficiency by interfering with mRNA processing. European Journal of Endocrinology, 2011, 164, 705-713.	3.7	14
42	Immunoproteasome LMP2 60HH Variant Alters MBP Epitope Generation and Reduces the Risk to Develop Multiple Sclerosis in Italian Female Population. PLoS ONE, 2010, 5, e9287.	2.5	56
43	The case of the solitary sick kidney. Kidney International, 2010, 77, 257-258.	5.2	1
44	A Recurrent Signal Peptide Mutation in the Growth Hormone Releasing Hormone Receptor with Defective Translocation to the Cell Surface and Isolated Growth Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 3939-3947.	3.6	25
45	A Functional Common Polymorphism in the Vitamin D-Responsive Element of theGH1Promoter Contributes to Isolated Growth Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 1005-1012.	3.6	21
46	ICOS gene haplotypes correlate with IL10 secretion and multiple sclerosis evolution. Journal of Neuroimmunology, 2007, 186, 193-198.	2.3	24
47	A variation in a Pit-1 site in the growth hormone gene (GH1) promoter induces a differential transcriptional activity. Molecular and Cellular Endocrinology, 2006, 249, 51-57.	3.2	12
48	A family-based study does not confirm the association of MYO9B with celiac disease in the Italian population. Genes and Immunity, 2006, 7, 606-608.	4.1	28
49	Polymorphisms in DNA repair genes as risk factors for asbestos-related malignant mesothelioma in a general population study. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2006, 599, 124-134.	1.0	52
50	A Novel Deletion in theGH1Gene Including the IVS3 Branch Site Responsible for Autosomal Dominant Isolated Growth Hormone Deficiency. Journal of Clinical Endocrinology and Metabolism, 2006, 91, 980-986.	3.6	22
51	Concordance, disease progression, and heritability of coeliac disease in Italian twins. Gut, 2006, 55, 803-808.	12.1	155
52	Osteopontin gene haplotypes correlate with multiple sclerosis development and progression. Journal of Neuroimmunology, 2005, 163, 172-178.	2.3	66
53	Two single-nucleotide polymorphisms in the 5? and 3? ends of the osteopontin gene contribute to susceptibility to systemic lupus erythematosus. Arthritis and Rheumatism, 2005, 52, 539-547.	6.7	94
54	Genetic defects in GH synthesis and secretion. European Journal of Endocrinology, 2004, 151 Suppl 1, S3-S9.	3.7	16

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55	Maternal effect in multiple sclerosis. Lancet, The, 2004, 363, 1748-1749.	13.7	5
56	A whole genome screen for linkage disequilibrium in multiple sclerosis performed in a continental Italian population. Journal of Neuroimmunology, 2003, 143, 97-100.	2.3	17
57	CD45 and multiple sclerosis: the exon 4 C77G polymorphism (additional studies and meta-analysis) and new markers. Journal of Neuroimmunology, 2003, 140, 216-221.	2.3	27
58	Two new PROP1 gene mutations responsible for compound pituitary hormone deficiency. Clinical Genetics, 2003, 64, 142-147.	2.0	24
59	Prolactin and prolactin receptor gene polymorphisms in multiple sclerosis and systemic lupus erythematosus. Human Immunology, 2003, 64, 274-284.	2.4	34
60	High levels of osteopontin associated with polymorphisms in its gene are a risk factor for development of autoimmunity/lymphoproliferation. Blood, 2003, 103, 1376-1382.	1.4	90
61	IL12B Polymorphism and Type 1 Diabetes in the Italian Population: A Case-Control Study. Diabetes, 2002, 51, 1649-1650.	0.6	24
62	Genetics of Multiple Sclerosis. Molecular Diagnosis and Therapy, 2002, 2, 37-58.	3.3	37
63	Identification of single nucleotide variations in the coding and regulatory regions of the myelin-associated glycoprotein gene and study of their association with multiple sclerosis. Journal of Neuroimmunology, 2002, 126, 196-204.	2.3	11
64	The IL12B gene does not confer susceptibility to coeliac disease. Tissue Antigens, 2002, 59, 70-72.	1.0	17
65	Association tests with systemic lupus erythematosus (SLE) of IL10 markers indicate a direct involvement of a CA repeat in the 5â€² regulatory region. Genes and Immunity, 2002, 3, 454-463.	4.1	36
66	Determination of SNP allele frequencies in pooled DNAs by primer extension genotyping and denaturing high-performance liquid chromatography. Journal of Proteomics, 2001, 47, 101-110.	2.4	46
67	Detection of AGXT gene mutations by denaturing high-performance liquid chromatography for diagnosis of hyperoxyluria type 1. Clinical and Experimental Medicine, 2001, 1, 99-104.	3.6	2
68	New polymorphisms in the IL-10 promoter region. Genes and Immunity, 2000, 1, 231-233.	4.1	98
69	Rapid Method for Detection of Extra (TA) in the Promoter of the Bilirubin-UDP-Glucuronosyl Transferase 1 Gene Associated with Gilbert Syndrome. Clinical Chemistry, 2000, 46, 129-131.	3.2	26
70	Study of Two Ectopeptidases in the Susceptibility to Celiac Disease: Two Newly Identified Polymorphisms of Dipeptidylpeptidase IV. Journal of Pediatric Gastroenterology and Nutrition, 2000, 30, 464-466.	1.8	7
71	Identification by Denaturing High-Performance Liquid Chromatography of Numerous Polymorphisms in a Candidate Region for Multiple Sclerosis Susceptibility. Genomics, 1999, 56, 247-253.	2.9	66
72	Evidence for gene conversion in the generation of extensive polymorphism in the promoter of the growth hormone gene. Human Genetics, 1997, 100, 249-255.	3.8	61

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73	Reassessment of the Specificity of Lens Opacities in Myotonic Dystrophy. Ophthalmic Research, 1996, 28, 224-229.	1.9	11
74	Homozygous tandem duplication within the gene encoding the Î²-subunit of rod phosphodiesterase as a cause for autosomal recessive retinitis pigmentosa. Human Mutation, 1995, 5, 228-234.	2.5	68
75	HLA supratypes in an Italian population. Immunogenetics, 1994, 39, 114-20.	2.4	12
76	Influence of ancestral gender on transmission of familial amyotrophic lateral sclerosis. Lancet, The, 1994, 344, 1639.	13.7	3
77	Gametic association of HSP70â€1 promoter region alleles and their inclusion in extended HLA haplotypes. Tissue Antigens, 1993, 42, 62-66.	1.0	9
78	Editorial: Novel Insights Into the Genetics of Growth Disorders. Frontiers in Genetics, 0, 13, .	2.3	0