Maria Iascone

List of Publications by Year
in descending order

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114 papers	2,510 citations	236833 25 h-index	254106 43 g-index
119 all docs	119 docs citations	119 times ranked	4948 citing authors

#	Article	IF	Citations
1	Diagnostic approach to neonatal and infantile cholestasis: A position paper by the SIGENP liver disease working group. Digestive and Liver Disease, 2022, 54, 40-53.	0.4	15
2	Progressive Clinical and Neuroradiological Findings in a Child with BCL11B Missense Mutation: Expanding the Phenotypic Spectrum of Related Disorder. Neuropediatrics, 2022, 53, 283-286.	0.3	5
3	Mucopolysaccharidosis-Plus Syndrome, a Rapidly Progressive Disease: Favorable Impact of a Very Prolonged Steroid Treatment on the Clinical Course in a Child. Genes, 2022, 13, 442.	1.0	8
4	Identical <scp>EP300</scp> variant leading to Rubinsteinâ€“Taybi syndrome with different clinical and immunologic phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 2129-2134.	0.7	2
5	Not Only Diagnostic Yield: Whole-Exome Sequencing in Infantile Cardiomyopathies Impacts on Clinical and Family Management. Journal of Cardiovascular Development and Disease, 2022, 9, 2.	0.8	12
6	Family history is key to the interpretation of exome sequencing in the prenatal context: unexpected diagnosis of Basal Cell Nevus Syndrome. Prenatal Diagnosis, 2022, , .	1.1	1
7	A novel homozygous disruptive PRF1 variant (K285Sfs*4) causes very early-onset of familial hemophagocytic lymphohystiocytosis type 2. Pediatric Hematology and Oncology, 2021, 38, 174-178.	0.3	1
8	Epilepsy and movement disorders in CDG : Report on the oldestâ€known MOGSâ€CDG patient. American Journal of Medical Genetics, Part A, 2021, 185, 219-222.	0.7	10
9	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	1.1	4
10	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499.	0.6	26
11	Lithium as a possible therapeutic strategy for Cornelia de Lange syndrome. Cell Death Discovery, 2021, 7, 34.	2.0	10
12	Reply. Journal of the American College of Cardiology, 2021, 77, 1378-1379.	1.2	0
13	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
14	Spinal cord involvement and paroxysmal events in â€œInfantile Onset Transient Hypomyelinationâ€•due to TMEM63A mutation. Journal of Human Genetics, 2021, 66, 1035-1037.	1.1	12
15	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
16	De novo variants in <scp>TCF7L2</scp> are associated with a syndromic neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 2384-2390.	0.7	13
17	La tecnologia genetica: ciÃ² che ogni pediatra dovrebbe sapere. Medico E Bambino, 2021, 40, 291-301.	0.1	2
18	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	2.6	17

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19	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	1.1	9
20	A novel mutation in COL3A1 associates to vascular Ehlersâ€“Danlos syndrome with predominant musculoskeletal involvement. Molecular Genetics & Genomic Medicine, 2021, 9, e1753.	0.6	4
21	Biallelic PI4KA variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. Brain, 2021, 144, 2659-2669.	3.7	19
22	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. Clinical Epigenetics, 2021, 13, 157.	1.8	22
23	ELP2 compound heterozygous variants associated with cortico-cerebellar atrophy, nodular heterotopia and epilepsy: Phenotype expansion and review of the literature. European Journal of Medical Genetics, 2021, 64, 104361.	0.7	2
24	A novel HIST1HE pathogenic variant in a girl with macrocephaly and intellectual disability: a new case and review of literature. Clinical Dysmorphology, 2021, 30, 39-43.	0.1	1
25	Heart transplantation in Danon disease: Long term single centre experience and review of the literature. European Journal of Medical Genetics, 2020, 63, 103645.	0.7	11
26	A novel nonsense and inactivating variant of ST3GAL3 in two infant siblings suffering severe epilepsy and expressing circulating CA19.9. Glycobiology, 2020, 30, 95-104.	1.3	19
27	ATP8A2-related disorders as recessive cerebellar ataxia. Journal of Neurology, 2020, 267, 203-213.	1.8	15
28	Human iPSC modelling of a familial form of atrial fibrillation reveals a gain of function of If and ICaL in patient-derived cardiomyocytes. Cardiovascular Research, 2020, 116, 1147-1160.	1.8	50
29	Dual genetic diagnoses: neurofibromatosis type 1 and KBG syndrome. Clinical Dysmorphology, 2020, 29, 101-103.	0.1	4
30	Development, behaviour and sensory processing in Marshallâ€“Smith syndrome and Malan syndrome: phenotype comparison in two related syndromes. Journal of Intellectual Disability Research, 2020, 64, 956-969.	1.2	13
31	Phenotypic spectrum of short-chain enoyl-Coa hydratase-1 (ECHS1) deficiency. European Journal of Paediatric Neurology, 2020, 28, 151-158.	0.7	18
32	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
33	Congenital Muscular Mitral-Aortic Discontinuity Identified in Patients With ObstructiveÂHypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2020, 76, 2238-2247.	1.2	15
34	A rare case of pediatric cardiomyopathy: AlstrÃ¶m syndrome identified by gene panel analysis. Clinical Case Reports (discontinued), 2020, 8, 3369-3373.	0.2	2
35	Clinical Profile of Cardiac Involvement in Danon Disease. Circulation Genomic and Precision Medicine, 2020, 13, e003117.	1.6	29
36	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22

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37	Missense NR2F1 variant in monozygotic twins affected with the Boschâ€“Boonstraâ€“Schaaf optic atrophy syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1278.	0.6	7
38	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	2.6	37
39	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	2.6	37
40	PIGWâ€related glycosylphosphatidylinositol deficiency: Description of a new patient and review of the literature. American Journal of Medical Genetics, Part A, 2020, 182, 1477-1482.	0.7	5
41	Intermittent granulocyte maturation arrest, hypocellular bone marrow, and episodic normal neutrophil count can be associated with SRP54 mutations causing Shwachmanâ€“Diamondâ€like syndrome. British Journal of Haematology, 2020, 189, e171-e174.	1.2	14
42	Familial Sleep Disorders in Unknown Genetic Syndrome. Journal of Pediatric Genetics, 2020, 09, 132-136.	0.3	0
43	Double homozygosity in CEP57 and DYNC2H1 genes detected by WES: Composite or expanded phenotype?. Molecular Genetics & Genomic Medicine, 2020, 8, e1064.	0.6	9
44	<scp>HNRNPH1</scp>â€related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. Clinical Genetics, 2020, 98, 91-98.	1.0	25
45	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	2.6	59
46	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	3.7	43
47	Phenotype delineation of ZNF462 related syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2075-2082.	0.7	23
48	Severe Late-Onset Fabry Cardiomyopathy Unmasked by a Multimodality Imaging Approach. Circulation: Cardiovascular Imaging, 2019, 12, e009709.	1.3	1
49	Diagnostic Yield of an Algorithm for Neonatal and Infantile Cholestasis Integrating Next-Generation Sequencing. Journal of Pediatrics, 2019, 211, 54-62.e4.	0.9	39
50	Prenatal findings in oralâ€facialâ€digital syndrome type VI: Report of three cases and literature review. Prenatal Diagnosis, 2019, 39, 652-655.	1.1	1
51	Mild phenotype in Molybdenum cofactor deficiency: A new patient and review of the literature. Molecular Genetics & Genomic Medicine, 2019, 7, e657.	0.6	22
52	Role of Preoperative Cardiovascular Magnetic Resonance in Planning Ventricular Septal Myectomy in Patients With Obstructive Hypertrophic Cardiomyopathy. American Journal of Cardiology, 2019, 123, 1517-1526.	0.7	18
53	Total loss of GM3 synthase activity by a normally processed enzyme in a novel variant and in all ST3GAL5 variants reported to cause a distinct congenital disorder of glycosylation. Glycobiology, 2019, 29, 229-241.	1.3	23
54	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	2.6	44

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55	Rare presentation and wide intrafamilial variability of Fabry disease: case report and review of the literature. Anatolian Journal of Cardiology, 2019, 22, 154-158.	0.5	0
56	Contemporary genetic testing in inherited cardiac disease. Journal of Cardiovascular Medicine, 2018, 19, 1-11.	0.6	48
57	Atypical presentation of pediatric BRAF RASopathy with acute encephalopathy. American Journal of Medical Genetics, Part A, 2018, 176, 2867-2871.	0.7	26
58	Two Missense Variants Detected in Breast Cancer Probands Preventing BRCA2-PALB2 Protein Interaction. Frontiers in Oncology, 2018, 8, 480.	1.3	11
59	Coronary pathology of inherited generalized arterial calcification of infancy: a case report. Cardiovascular Pathology, 2018, 36, 15-19.	0.7	3
60	Mitral valve abnormalities in hypertrophic cardiomyopathy: a primary expression of the disease? Getting closer to the answer. European Heart Journal Cardiovascular Imaging, 2018, 19, 1107-1108.	0.5	6
61	A novel <scp>EP</scp>300 mutation associated with Rubinsteinâ€Taybi syndrome type 2 presenting as combined immunodeficiency. Pediatric Allergy and Immunology, 2018, 29, 776-781.	1.1	4
62	Further delineation of Malan syndrome. Human Mutation, 2018, 39, 1226-1237.	1.1	42
63	P2X7R mutation disrupts the NLRP3-mediated Th program and predicts poor cardiac allograft outcomes. Journal of Clinical Investigation, 2018, 128, 3490-3503.	3.9	31
64	Intraoperative Diagnosis of Anderson-Fabry Disease in Patients With Obstructive Hypertrophic Cardiomyopathy Undergoing Surgical Myectomy. JAMA Cardiology, 2017, 2, 1147.	3.0	14
65	A novel mutation in FHL1 gene causing hypertrophic cardiomyopathy associated with myopathy. Neuromuscular Disorders, 2017, 27, S139.	0.3	0
66	Mutual epitheliumâ€macrophage dependency in liver carcinogenesis mediated by ST18. Hepatology, 2017, 65, 1708-1719.	3.6	19
67	First evidence of Smithâ€“Magenis syndrome in mother and daughter due to a novel RAI mutation. American Journal of Medical Genetics, Part A, 2017, 173, 231-238.	0.7	17
68	Atypical Clinical Presentation and Successful Treatment With Oral Cholic Acid of a Child with Defective Bile Acid Synthesis due to a Novel Mutation in the HSD3B7 Gene. Mental Illness, 2017, 9, 7266.	0.8	6
69	A Child With Ichthyosis and Liver Failure. Journal of Pediatric Gastroenterology and Nutrition, 2017, 65, e70-e73.	0.9	5
70	Reversible Dilated Cardiomyopathy: Into the Thaumaturgy of the Heartâ€”Part 1. Neurology International, 2016, 6, 5861.	0.2	0
71	Reversible dilated cardiomyopathy: into the thaumaturgy of the heart - Part 2. Neurology International, 2016, 6, .	0.2	0
72	A Novel HRAS Mutation Independently Contributes to Left Ventricular Hypertrophy in a Family with a Known MYH7 Mutation. PLoS ONE, 2016, 11, e0168501.	1.1	13

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73	Clinical and molecular characterization of two patients with palmoplantar keratoderma-congenital alopecia syndrome type 2. Clinical and Experimental Dermatology, 2016, 41, 632-635.	0.6	8
74	Temporomandibular joint ankylosis as part of the clinical spectrum of Careyâ€“Finemanâ€“Ziter syndrome?. American Journal of Medical Genetics, Part A, 2016, 170, 2191-2195.	0.7	3
75	Neonatal Jaundice with Splenomegaly: Not a Common Pick. Fetal and Pediatric Pathology, 2016, 35, 108-111.	0.4	8
76	Microcephaly, ectodermal dysplasia, multiple skeletal anomalies and distinctive facial appearance: Delineation of cerebroâ€dermatoâ€osseousâ€dysplasia. American Journal of Medical Genetics, Part A, 2015, 167, 842-851.	0.7	1
77	Endocardial Fibroelastosis Is Caused by Aberrant Endothelial to Mesenchymal Transition. Circulation Research, 2015, 116, 857-866.	2.0	98
78	Loss-of-FunctionFANCLMutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. Human Mutation, 2015, 36, 562-568.	1.1	23
79	Cavotricuspid Isthmus Ablation and Subcutaneous Monitoring Device Implantation in a 2â€Yearâ€Old Baby with 2 SCN5A Mutations, Sinus Node Dysfunction, Atrial Flutter Recurrences, and Drug Induced Longâ€QT Syndrome: A Tricky Case of Pediatric Overlap Syndrome?. Journal of Cardiovascular Electrophysiology, 2015, 26, 346-349.	0.8	3
80	Transaortic Chordal Cutting. Journal of the American College of Cardiology, 2015, 66, 1687-1696.	1.2	141
81	McCune Albright syndrome and neonatal cholestasis: A new association. Digestive and Liver Disease, 2015, 47, e256.	0.4	1
82	Improving molecular diagnosis in epilepsy by a dedicated high-throughput sequencing platform. European Journal of Human Genetics, 2015, 23, 354-362.	1.4	64
83	Identification of a novel de novo deletion in RAF1 associated with biventricular hypertrophy in Noonan syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2069-2073.	0.7	5
84	Identification of a human splenic marginal zone B cell precursor with NOTCH2-dependent differentiation properties. Journal of Experimental Medicine, 2014, 211, 987-1000.	4.2	113
85	Novel Î±-Actinin 2 Variant Associated With Familial Hypertrophic Cardiomyopathy and Juvenile Atrial Arrhythmias. Circulation: Cardiovascular Genetics, 2014, 7, 741-750.	5.1	74
86	Significance of Sarcomere Gene Mutations Analysis in the End-Stage Phase of Hypertrophic Cardiomyopathy. American Journal of Cardiology, 2014, 114, 769-776.	0.7	76
87	Alagille syndrome: Is it always cholestasis?. Digestive and Liver Disease, 2014, 46, e106.	0.4	1
88	Extensive Arterial Tortuosity and Severe Aortic Dilation in a Newborn With an EFEMP2 Mutation. Circulation, 2012, 126, 2764-2768.	1.6	20
89	Fish Oil and Postoperative Atrial Fibrillation. JAMA - Journal of the American Medical Association, 2012, 308, 2001.	3.8	201
90	A contemporary European experience with surgical septal myectomy in hypertrophic cardiomyopathy. European Heart Journal, 2012, 33, 2080-2087.	1.0	88

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91	Letter by Iascone et al Regarding Article, â€œPopulation-Based Variation in Cardiomyopathy Genesâ€• Circulation: Cardiovascular Genetics, 2012, 5, e57; author reply e58.	5.1	0
92	De novo Deletion of 1q31.1â€“q32.1 in a Patient with Developmental Delay and Behavioral Disorders. Cytogenetic and Genome Research, 2012, 136, 167-170.	0.6	4
93	A new mutational mechanism for hypertrophic cardiomyopathy. Gene, 2012, 507, 165-169.	1.0	10
94	Identification of de novo mutations and rare variants in hypoplastic left heart syndrome. Clinical Genetics, 2012, 81, 542-554.	1.0	97
95	GAMES identifies and annotates mutations in next-generation sequencing projects. Bioinformatics, 2011, 27, 9-13.	1.8	28
96	Toward the development of a fully elastic mitral ring: Preliminary, acute, in vivo evaluation of physiomechanical behavior. Journal of Thoracic and Cardiovascular Surgery, 2009, 137, 174-179.	0.4	6
97	Novel human pathological mutations. Gene symbol: MYBPC3. Disease: cardiomyopathy, hypertrophic. Human Genetics, 2009, 126, 351.	1.8	1
98	Novel human pathological mutations. Human Genetics, 2008, 123, 537-555.	1.8	11
99	Gene symbol: LAMP2. Disease: Danon disease. Human Genetics, 2008, 123, 537.	1.8	7
100	Implantation of an Elastic Ring at Equator of the Left Ventricle Influences Cardiac Mechanics in Experimental Acute Ventricular Dysfunction. Journal of the American College of Cardiology, 2007, 50, 1791-1798.	1.2	6
101	The Titan can help titin: from micro to macro myocardial elasticity. Journal of Cardiovascular Medicine, 2006, 7, 153-158.	0.6	3
102	Left main stem patch plasty and aortic root homograft in Takayasu's disease. Annals of Thoracic Surgery, 2004, 77, 314-317.	0.7	9
103	Association between 5,10-Methylenetetrahydrofolate Reductase C677T and A1298C Polymorphisms and Conotruncal Heart Defects. Clinical Chemistry and Laboratory Medicine, 2003, 41, 276-80.	1.4	53
104	Analysis of the variation in the hsp70-1 and hsp90Î± mRNA expression in human myocardial tissue that has undergone surgical stress. Cell Stress and Chaperones, 2003, 8, 18.	1.2	5
105	Molecular characterization of 22q11 deletion in a three-generation family with maternal transmission. American Journal of Medical Genetics Part A, 2002, 108, 319-321.	2.4	21
106	Molecular Characterization of Chromosome 22 Deletions by Short Tandem Repeat Polymorphism (STRP) in Patients with Conotruncal Heart Defects. Clinical Chemistry and Laboratory Medicine, 2001, 39, 1249-58.	1.4	16
107	Familial occurrence of isolated right ventricular hypoplasia. , 2000, 90, 356-357.		13
108	Measurement of brain natriuretic peptide in plasma samples and cardiac tissue extracts by means of an immunoradiometric assay method. Scandinavian Journal of Clinical and Laboratory Investigation, 2000, 60, 81-90.	0.6	44

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109	A rapid procedure for the quantitation of natriuretic peptide RNAs by competitive RT-PCR in congenital heart defects. Journal of Endocrinological Investigation, 1999, 22, 835-842.	1.8	4
110	Acute enoximone effect on systemic and renal hemodynamics in patients with heart failure. Cardiovascular Drugs and Therapy, 1996, 10, 81-87.	1.3	4
111	Preparation of mono-radioiodinated tracers for study of the in vivo metabolism of atrial natriuretic peptide in humans. European Journal of Nuclear Medicine and Molecular Imaging, 1995, 22, 997-1004.	2.2	15
112	In vivo measurement of ANP overall turnover and identification of its main metabolic pathways under steady state conditions in humans. Journal of Endocrinological Investigation, 1995, 18, 194-204.	1.8	8
113	A Neuro-metabolic Syndrome that Needs to Be Discovered: A Child with Late Onset Asparagine Synthetase Deficiency. Journal of Pediatric Epilepsy, 0, , .	0.1	0
114	PRENATAL ULTRASOUND FINDINGS ASSOCIATED WITH PIGW VARIANTS:: ONE MORE PIECE IN THE FRYNS SYNDROME PUZZLE? PIGW â€related prenatal findings. Prenatal Diagnosis, 0, , .	1.1	1