

Maria Iascone

List of Publications by Year in descending order

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Version: 2024-02-01

114
papers

2,510
citations

236925
25
h-index

254184
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.	0.9	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.	0.6	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.	2.4	8
4	Identical <sc><i>EP300</i></sc> variant leading to Rubinsteinâ€Taybi syndrome with different clinical and immunologic phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 2129-2134.	1.2	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.	1.6	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, , .	2.3	1
7	A novel homozygous disruptive PRF1 variant (K285Sfs*4) causes very early-onset of familial hemophagocytic lymphohistiocytosis type 2. Pediatric Hematology and Oncology, 2021, 38, 174-178.	0.8	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.	1.2	10
9	Defining the genotypic and phenotypic spectrum of X-linked MSL3-related disorder. Genetics in Medicine, 2021, 23, 384-395.	2.4	4
10	Absent B cells, agammaglobulinemia, and hypertrophic cardiomyopathy in folliculin-interacting protein 1 deficiency. Blood, 2021, 137, 493-499.	1.4	26
11	Lithium as a possible therapeutic strategy for Cornelia de Lange syndrome. Cell Death Discovery, 2021, 7, 34.	4.7	10
12	Reply. Journal of the American College of Cardiology, 2021, 77, 1378-1379.	2.8	0
13	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
14	Spinal cord involvement and paroxysmal events in â€Infantile Onset Transient Hypomyelinationâ€due to TMEM63A mutation. Journal of Human Genetics, 2021, 66, 1035-1037.	2.3	12
15	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
16	De novo variants in <sc><i>TCF7L2</i></sc> are associated with a syndromic neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 2384-2390.	1.2	13
17	La tecnologia genetica: ciÂ² 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.	6.2	17

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19	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. <i>Genetics in Medicine</i> , 2021, 23, 1901-1911.	2.4	9
20	A novel mutation in COL3A1 associates to vascular Ehlers-Danlos syndrome with predominant musculoskeletal involvement. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1753.	1.2	4
21	Biallelic <i>PI4KA</i> variants cause a novel neurodevelopmental syndrome with hypomyelinating leukodystrophy. <i>Brain</i> , 2021, 144, 2659-2669.	7.6	19
22	Childhood-onset dystonia-causing KMT2B variants result in a distinctive genomic hypermethylation profile. <i>Clinical Epigenetics</i> , 2021, 13, 157.	4.1	22
23	ELP2 compound heterozygous variants associated with cortico-cerebellar atrophy, nodular heterotopia and epilepsy: Phenotype expansion and review of the literature. <i>European Journal of Medical Genetics</i> , 2021, 64, 104361.	1.3	2
24	A novel HIST1HE pathogenic variant in a girl with macrocephaly and intellectual disability: a new case and review of literature. <i>Clinical Dysmorphology</i> , 2021, 30, 39-43.	0.3	1
25	Heart transplantation in Danon disease: Long term single centre experience and review of the literature. <i>European Journal of Medical Genetics</i> , 2020, 63, 103645.	1.3	11
26	A novel nonsense and inactivating variant of ST3GAL3 in two infant siblings suffering severe epilepsy and expressing circulating CA19.9. <i>Glycobiology</i> , 2020, 30, 95-104.	2.5	19
27	ATP8A2-related disorders as recessive cerebellar ataxia. <i>Journal of Neurology</i> , 2020, 267, 203-213.	3.6	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. <i>Cardiovascular Research</i> , 2020, 116, 1147-1160.	3.8	50
29	Dual genetic diagnoses: neurofibromatosis type 1 and KBG syndrome. <i>Clinical Dysmorphology</i> , 2020, 29, 101-103.	0.3	4
30	Development, behaviour and sensory processing in Marshall-Smith syndrome and Malan syndrome: phenotype comparison in two related syndromes. <i>Journal of Intellectual Disability Research</i> , 2020, 64, 956-969.	2.0	13
31	Phenotypic spectrum of short-chain enoyl-CoA hydratase-1 (ECHS1) deficiency. <i>European Journal of Paediatric Neurology</i> , 2020, 28, 151-158.	1.6	18
32	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. <i>Brain</i> , 2020, 143, 2437-2453.	7.6	21
33	Congenital Muscular Mitral-Aortic Discontinuity Identified in Patients With Obstructive Hypertrophic Cardiomyopathy. <i>Journal of the American College of Cardiology</i> , 2020, 76, 2238-2247.	2.8	15
34	A rare case of pediatric cardiomyopathy: Alström syndrome identified by gene panel analysis. <i>Clinical Case Reports (discontinued)</i> , 2020, 8, 3369-3373.	0.5	2
35	Clinical Profile of Cardiac Involvement in Danon Disease. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e003117.	3.6	29
36	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. <i>Genetics in Medicine</i> , 2020, 22, 1215-1226.	2.4	22

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37	Missense <i>NR2F1</i> variant in monozygotic twins affected with the Bosch–Boonstra–Schaaf optic atrophy syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1278.	1.2	7
38	De Novo Variants in <i>CNOT1</i> , a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. <i>American Journal of Human Genetics</i> , 2020, 107, 164-172.	6.2	37
39	De novo <i>EIF2AK1</i> and <i>EIF2AK2</i> Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	6.2	37
40	<i>PIGW</i> -related glycosylphosphatidylinositol deficiency: Description of a new patient and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1477-1482.	1.2	5
41	Intermittent granulocyte maturation arrest, hypocellular bone marrow, and episodic normal neutrophil count can be associated with <i>SRP54</i> mutations causing Shwachman–Diamond–like syndrome. <i>British Journal of Haematology</i> , 2020, 189, e171-e174.	2.5	14
42	Familial Sleep Disorders in Unknown Genetic Syndrome. <i>Journal of Pediatric Genetics</i> , 2020, 09, 132-136.	0.7	0
43	Double homozygosity in <i>CEP57</i> and <i>DYNC2H1</i> genes detected by WES: Composite or expanded phenotype?. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1064.	1.2	9
44	<i>HNRNP1</i> -related syndromic intellectual disability: Seven additional cases suggestive of a distinct syndromic neurodevelopmental syndrome. <i>Clinical Genetics</i> , 2020, 98, 91-98.	2.0	25
45	DNA Methylation Signature for <i>EZH2</i> Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. <i>American Journal of Human Genetics</i> , 2020, 106, 596-610.	6.2	59
46	Cohesin complex-associated holoprosencephaly. <i>Brain</i> , 2019, 142, 2631-2643.	7.6	43
47	Phenotype delineation of <i>ZNF462</i> related syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2075-2082.	1.2	23
48	Severe Late-Onset Fabry Cardiomyopathy Unmasked by a Multimodality Imaging Approach. <i>Circulation: Cardiovascular Imaging</i> , 2019, 12, e009709.	2.6	1
49	Diagnostic Yield of an Algorithm for Neonatal and Infantile Cholestasis Integrating Next-Generation Sequencing. <i>Journal of Pediatrics</i> , 2019, 211, 54-62.e4.	1.8	39
50	Prenatal findings in oral–facial–digital syndrome type VI: Report of three cases and literature review. <i>Prenatal Diagnosis</i> , 2019, 39, 652-655.	2.3	1
51	Mild phenotype in Molybdenum cofactor deficiency: A new patient and review of the literature. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e657.	1.2	22
52	Role of Preoperative Cardiovascular Magnetic Resonance in Planning Ventricular Septal Myectomy in Patients With Obstructive Hypertrophic Cardiomyopathy. <i>American Journal of Cardiology</i> , 2019, 123, 1517-1526.	1.6	18
53	Total loss of GM3 synthase activity by a normally processed enzyme in a novel variant and in all <i>ST3GAL5</i> variants reported to cause a distinct congenital disorder of glycosylation. <i>Glycobiology</i> , 2019, 29, 229-241.	2.5	23
54	De Novo Variants in <i>MAPK8IP3</i> Cause Intellectual Disability with Variable Brain Anomalies. <i>American Journal of Human Genetics</i> , 2019, 104, 203-212.	6.2	44

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55	Rare presentation and wide intrafamilial variability of Fabry disease: case report and review of the literature. <i>Anatolian Journal of Cardiology</i> , 2019, 22, 154-158.	0.9	0
56	Contemporary genetic testing in inherited cardiac disease. <i>Journal of Cardiovascular Medicine</i> , 2018, 19, 1-11.	1.5	48
57	Atypical presentation of pediatric <i>BRAF</i> RASopathy with acute encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2867-2871.	1.2	26
58	Two Missense Variants Detected in Breast Cancer Probands Preventing BRCA2-PALB2 Protein Interaction. <i>Frontiers in Oncology</i> , 2018, 8, 480.	2.8	11
59	Coronary pathology of inherited generalized arterial calcification of infancy: a case report. <i>Cardiovascular Pathology</i> , 2018, 36, 15-19.	1.6	3
60	Mitral valve abnormalities in hypertrophic cardiomyopathy: a primary expression of the disease? Getting closer to the answer. <i>European Heart Journal Cardiovascular Imaging</i> , 2018, 19, 1107-1108.	1.2	6
61	A novel <i><sc>EP</sc>300</i> mutation associated with Rubinsteinâ€Taybi syndrome type 2 presenting as combined immunodeficiency. <i>Pediatric Allergy and Immunology</i> , 2018, 29, 776-781.	2.6	4
62	Further delineation of Malan syndrome. <i>Human Mutation</i> , 2018, 39, 1226-1237.	2.5	42
63	P2X7R mutation disrupts the NLRP3-mediated Th program and predicts poor cardiac allograft outcomes. <i>Journal of Clinical Investigation</i> , 2018, 128, 3490-3503.	8.2	31
64	Intraoperative Diagnosis of Anderson-Fabry Disease in Patients With Obstructive Hypertrophic Cardiomyopathy Undergoing Surgical Myectomy. <i>JAMA Cardiology</i> , 2017, 2, 1147.	6.1	14
65	A novel mutation in FHL1 gene causing hypertrophic cardiomyopathy associated with myopathy. <i>Neuromuscular Disorders</i> , 2017, 27, S139.	0.6	0
66	Mutual epitheliumâ€macrophage dependency in liver carcinogenesis mediated by ST18. <i>Hepatology</i> , 2017, 65, 1708-1719.	7.3	19
67	First evidence of Smithâ€Magenis syndrome in mother and daughter due to a novel RAI mutation. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 231-238.	1.2	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. <i>Mental Illness</i> , 2017, 9, 7266.	0.8	6
69	A Child With Ichthyosis and Liver Failure. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2017, 65, e70-e73.	1.8	5
70	Reversible Dilated Cardiomyopathy: Into the Thaumaturgy of the Heartâ€Part 1. <i>Neurology International</i> , 2016, 6, 5861.	0.5	0
71	Reversible dilated cardiomyopathy: into the thaumaturgy of the heart - Part 2. <i>Neurology International</i> , 2016, 6, .	0.5	0
72	A Novel HRAS Mutation Independently Contributes to Left Ventricular Hypertrophy in a Family with a Known MYH7 Mutation. <i>PLoS ONE</i> , 2016, 11, e0168501.	2.5	13

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73	Clinical and molecular characterization of two patients with palmoplantar keratoderma-congenital alopecia syndrome type 2. <i>Clinical and Experimental Dermatology</i> , 2016, 41, 632-635.	1.3	8
74	Temporomandibular joint ankylosis as part of the clinical spectrum of Careyâ€“Finemanâ€“Ziter syndrome?. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2191-2195.	1.2	3
75	Neonatal Jaundice with Splenomegaly: Not a Common Pick. <i>Fetal and Pediatric Pathology</i> , 2016, 35, 108-111.	0.7	8
76	Microcephaly, ectodermal dysplasia, multiple skeletal anomalies and distinctive facial appearance: Delineation of cerebroâ€“dermatoâ€“osseousâ€“dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 842-851.	1.2	1
77	Endocardial Fibroelastosis Is Caused by Aberrant Endothelial to Mesenchymal Transition. <i>Circulation Research</i> , 2015, 116, 857-866.	4.5	98
78	Loss-of-Function <i>FANCL</i> Mutations Associate with Severe Fanconi Anemia Overlapping the VACTERL Association. <i>Human Mutation</i> , 2015, 36, 562-568.	2.5	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?. <i>Journal of Cardiovascular Electrophysiology</i> , 2015, 26, 346-349.	1.7	3
80	Transaortic Chordal Cutting. <i>Journal of the American College of Cardiology</i> , 2015, 66, 1687-1696.	2.8	141
81	McCune Albright syndrome and neonatal cholestasis: A new association. <i>Digestive and Liver Disease</i> , 2015, 47, e256.	0.9	1
82	Improving molecular diagnosis in epilepsy by a dedicated high-throughput sequencing platform. <i>European Journal of Human Genetics</i> , 2015, 23, 354-362.	2.8	64
83	Identification of a novel de novo deletion in <i>RAF1</i> associated with biventricular hypertrophy in Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2069-2073.	1.2	5
84	Identification of a human splenic marginal zone B cell precursor with NOTCH2-dependent differentiation properties. <i>Journal of Experimental Medicine</i> , 2014, 211, 987-1000.	8.5	113
85	Novel Î±-Actinin 2 Variant Associated With Familial Hypertrophic Cardiomyopathy and Juvenile Atrial Arrhythmias. <i>Circulation: Cardiovascular Genetics</i> , 2014, 7, 741-750.	5.1	74
86	Significance of Sarcomere Gene Mutations Analysis in the End-Stage Phase of Hypertrophic Cardiomyopathy. <i>American Journal of Cardiology</i> , 2014, 114, 769-776.	1.6	76
87	Alagille syndrome: Is it always cholestasis?. <i>Digestive and Liver Disease</i> , 2014, 46, e106.	0.9	1
88	Extensive Arterial Tortuosity and Severe Aortic Dilation in a Newborn With an <i>EFEMP2</i> Mutation. <i>Circulation</i> , 2012, 126, 2764-2768.	1.6	20
89	Fish Oil and Postoperative Atrial Fibrillation. <i>JAMA - Journal of the American Medical Association</i> , 2012, 308, 2001.	7.4	201
90	A contemporary European experience with surgical septal myectomy in hypertrophic cardiomyopathy. <i>European Heart Journal</i> , 2012, 33, 2080-2087.	2.2	88

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91	Letter by Iascone et al Regarding Article, "Population-Based Variation in Cardiomyopathy Genes", 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.	1.1	4
93	A new mutational mechanism for hypertrophic cardiomyopathy. Gene, 2012, 507, 165-169.	2.2	10
94	Identification of <i>de novo</i> mutations and rare variants in hypoplastic left heart syndrome. Clinical Genetics, 2012, 81, 542-554.	2.0	97
95	GAMES identifies and annotates mutations in next-generation sequencing projects. Bioinformatics, 2011, 27, 9-13.	4.1	28
96	Toward the development of a fully elastic mitral ring: Preliminary, acute, in vivo evaluation of biomechanical behavior. Journal of Thoracic and Cardiovascular Surgery, 2009, 137, 174-179.	0.8	6
97	Novel human pathological mutations. Gene symbol: MYBPC3. Disease: cardiomyopathy, hypertrophic. Human Genetics, 2009, 126, 351.	3.8	1
98	Novel human pathological mutations. Human Genetics, 2008, 123, 537-555.	3.8	11
99	Gene symbol: LAMP2. Disease: Danon disease. Human Genetics, 2008, 123, 537.	3.8	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.	2.8	6
101	The Titan can help titin: from micro to macro myocardial elasticity. Journal of Cardiovascular Medicine, 2006, 7, 153-158.	1.5	3
102	Left main stem patch plasty and aortic root homograft in Takayasu's disease. Annals of Thoracic Surgery, 2004, 77, 314-317.	1.3	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.	2.3	53
104	Analysis of the variation in the hsp70-1 and hsp90 α mRNA expression in human myocardial tissue that has undergone surgical stress. Cell Stress and Chaperones, 2003, 8, 18.	2.9	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.	2.3	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.	1.2	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.	3.3	4
110	Acute enoximone effect on systemic and renal hemodynamics in patients with heart failure. Cardiovascular Drugs and Therapy, 1996, 10, 81-87.	2.6	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.	2.1	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.	3.3	8
113	A Neuro-metabolic Syndrome that Needs to Be Discovered: A Child with Late Onset Asparagine Synthetase Deficiency. Journal of Pediatric Epilepsy, 0, , .	0.2	0
114	PRENATAL ULTRASOUND FINDINGS ASSOCIATED WITH <i>PIGW</i> VARIANTS:: ONE MORE PIECE IN THE FRYNS SYNDROME PUZZLE? <i>PIGW</i> -related prenatal findings. Prenatal Diagnosis, 0, , .	2.3	1