

Elena Manara

List of Publications by Year in descending order

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Version: 2024-02-01

78
papers

1,260
citations

430874

18
h-index

414414

32
g-index

85
all docs

85
docs citations

85
times ranked

1900
citing authors

#	ARTICLE	IF	CITATIONS
1	A novel complex genomic rearrangement affecting the KCNJ2 regulatory region causes a variant of Cooks syndrome. <i>Human Genetics</i> , 2022, 141, 217-227.	3.8	1
2	A Multi-Gene Panel to Identify Lipedema-Predisposing Genetic Variants by a Next-Generation Sequencing Strategy. <i>Journal of Personalized Medicine</i> , 2022, 12, 268.	2.5	11
3	Recessive multiple epiphyseal dysplasia and Stargardt disease in two sisters. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1630.	1.2	2
4	Potential role of microbiome in Chronic Fatigue Syndrome/Myalgic Encephalomyelitis (CFS/ME). <i>Scientific Reports</i> , 2021, 11, 7043.	3.3	42
5	Psychomotor Delay in a Child with FGFR3 G380R Pathogenic Mutation Causing Achondroplasia. <i>Global Medical Genetics</i> , 2021, 08, 100-103.	0.9	0
6	Genetic testing in vascular and lymphatic malformations. <i>Italian Journal of Vascular and Endovascular Surgery</i> , 2021, 28, .	1.0	1
7	Expanding the Clinical and Genetic Spectrum of RAB28-Related Cone-Rod Dystrophy: Pathogenicity of Novel Variants in Italian Families. <i>International Journal of Molecular Sciences</i> , 2021, 22, 381.	4.1	8
8	A next generation sequencing gene panel for use in the diagnosis of anorexia nervosa. <i>Eating and Weight Disorders</i> , 2021, , 1.	2.5	9
9	<i>CDH5</i> , a Possible New Candidate Gene for Genetic Testing of Lymphedema. <i>Lymphatic Research and Biology</i> , 2021, , .	1.1	3
10	Study of the effects of <i>Lemna minor</i> extracts on human immune cell populations. <i>European Review for Medical and Pharmacological Sciences</i> , 2021, 25, 43-48.	0.7	1
11	Genetics of fat deposition. <i>European Review for Medical and Pharmacological Sciences</i> , 2021, 25, 14-22.	0.7	10
12	Steroid-converting enzymes in human adipose tissues and fat deposition with a focus on AKR1C enzymes. <i>European Review for Medical and Pharmacological Sciences</i> , 2021, 25, 23-32.	0.7	13
13	In vitro and clinical studies on the efficacy of β -cyclodextrin and hydroxytyrosol against SARS-CoV-2 infection. <i>European Review for Medical and Pharmacological Sciences</i> , 2021, 25, 81-89.	0.7	4
14	Naturally-occurring and cultured bacteriophages in human therapy. <i>European Review for Medical and Pharmacological Sciences</i> , 2021, 25, 101-107.	0.7	0
15	Hydroxytyrosol: A natural compound with promising pharmacological activities. <i>Journal of Biotechnology</i> , 2020, 309, 29-33.	3.8	138
16	Electrical Stimulation in the Treatment of Lymphedema and Associated Skin Ulcers. <i>Lymphatic Research and Biology</i> , 2020, 18, 270-276.	1.1	7
17	Molecular pathways involved in lymphedema: Hydroxytyrosol as a candidate natural compound for treating the effects of lymph accumulation. <i>Journal of Biotechnology</i> , 2020, 308, 82-86.	3.8	8
18	Clinical Evaluation of a Custom Gene Panel as a Tool for Precision Male Infertility Diagnosis by Next-Generation Sequencing. <i>Life</i> , 2020, 10, 242.	2.4	12

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19	FOXC2 Disease Mutations Identified in Lymphedema Distichiasis Patients Impair Transcriptional Activity and Cell Proliferation. <i>International Journal of Molecular Sciences</i> , 2020, 21, 5112.	4.1	10
20	Segregation Analysis of Rare NRP1 and NRP2 Variants in Families with Lymphedema. <i>Genes</i> , 2020, 11, 1361.	2.4	4
21	Aldo-Keto Reductase 1C1 (AKR1C1) as the First Mutated Gene in a Family with Nonsyndromic Primary Lipedema. <i>International Journal of Molecular Sciences</i> , 2020, 21, 6264.	4.1	27
22	Somatic Variant Analysis Identifies Targets for Tailored Therapies in Patients with Vascular Malformations. <i>Journal of Clinical Medicine</i> , 2020, 9, 3387.	2.4	6
23	Genetic contributions to the etiology of anorexia nervosa: New perspectives in molecular diagnosis and treatment. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1244.	1.2	21
24	Investigation on the role of biallelic variants in <i>VEGF</i> found in a patient affected by Milroy-like lymphedema. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1389.	1.2	6
25	Unique combination and in silico modeling of biallelic POLR3A variants as a cause of Wiedemann-Rautenstrauch syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 1675-1680.	2.8	4
26	Adult-onset glutaric aciduria type I: rare presentation of a treatable disorder. <i>Neurogenetics</i> , 2020, 21, 179-186.	1.4	12
27	Pilot study for the evaluation of safety profile of a potential inhibitor of SARS-CoV-2 endocytosis. <i>Acta Biomedica</i> , 2020, 91, e2020009.	0.3	8
28	Genetic analysis of intellectual disability and autism. <i>Acta Biomedica</i> , 2020, 91, e2020003.	0.3	18
29	A pilot study on the preventative potential of alpha-cyclodextrin and hydroxytyrosol against SARS-CoV-2 transmission. <i>Acta Biomedica</i> , 2020, 91, e2020022.	0.3	14
30	Early-onset of Frontotemporal Dementia and Amyotrophic Lateral Sclerosis in an Albanian Patient with a c.1319C>T Variant in the UBQLN2 Gene. <i>Open Medicine Journal</i> , 2020, 7, 25-31.	0.7	1
31	The Biennial report: The collaboration between MAGI Research, Diagnosis and Treatment Center of Genetic and Rare Diseases and Near East University DESAM Institute. <i>The EuroBiotech Journal</i> , 2020, 4, 167-170.	1.0	0
32	Genetic testing for autonomic dysfunction or dysautonomias. <i>Acta Biomedica</i> , 2020, 91, e2020002.	0.3	4
33	Pheromone receptors and their putative ligands: possible role in humans. <i>European Review for Medical and Pharmacological Sciences</i> , 2020, 24, 2140-2150.	0.7	5
34	Reply to the Letter - "The development of Brugada syndrome phenotype is multifactorial, combining genetic and environmental factors". <i>European Review for Medical and Pharmacological Sciences</i> , 2020, 24, 3446-3447.	0.7	0
35	Research Article Glu298Asp polymorphism in the <i>NOS3</i> gene is not associated with susceptibility to chronic heart failure in a Russian population. <i>Genetics and Molecular Research</i> , 2019, 18, .	0.2	1
36	Mutation profile of BBS genes in patients with Bardet-Biedl syndrome: an Italian study. <i>Italian Journal of Pediatrics</i> , 2019, 45, 72.	2.6	30

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37	Vascular anomalies: molecular bases, genetic testing and therapeutic approaches. <i>International Angiology</i> , 2019, 38, 157-170.	0.9	14
38	Quality assurance of genetic laboratories and the EBTNA practice certification. <i>Journal of Biotechnology</i> , 2019, 305, S7.	3.8	0
39	Taste, olfactory and texture related genes and food choices: implications on health status. <i>European Review for Medical and Pharmacological Sciences</i> , 2019, 23, 1305-1321.	0.7	19
40	PipeMAGI: an integrated and validated workflow for analysis of NGS data for clinical diagnostics. <i>European Review for Medical and Pharmacological Sciences</i> , 2019, 23, 6753-6765.	0.7	17
41	Putative role of Brugada syndrome genes in familial atrial fibrillation. <i>European Review for Medical and Pharmacological Sciences</i> , 2019, 23, 7582-7598.	0.7	3
42	Epigenetic heterogeneity affects the risk of relapse in children with t(8;21)RUNX1-RUNX1T1-rearranged AML. <i>Leukemia</i> , 2018, 32, 1124-1134.	7.2	17
43	Genetic tests in lymphatic vascular malformations and lymphedema. <i>Journal of Medical Genetics</i> , 2018, 55, 222-232.	3.2	34
44	Variant discovery in patients with Mendelian vascular anomalies by next-generation sequencing and their use in patient clinical management. <i>Journal of Vascular Surgery</i> , 2018, 67, 922-932.e11.	1.1	20
45	A novel p.(Glu111Val) missense mutation in GUCA1A associated with cone-rod dystrophy leads to impaired calcium sensing and perturbed second messenger homeostasis in photoreceptors. <i>Human Molecular Genetics</i> , 2018, 27, 4204-4217.	2.9	32
46	Moving from research to diagnostics: MAGI's experience in lymphedema. <i>Journal of Biotechnology</i> , 2018, 280, S6-S7.	3.8	0
47	Quality assurance of genetic laboratories and the EBTNA practice certification, a simple standardization assurance system for a laboratory network. <i>The EuroBiotech Journal</i> , 2018, 2, 215-222.	1.0	1
48	Characterization of children with FLT3-ITD acute myeloid leukemia: a report from the AIEOP AML-2002 study group. <i>Leukemia</i> , 2017, 31, 18-25.	7.2	29
49	NUP98-fusion transcripts characterize different biological entities within acute myeloid leukemia: a report from the AIEOP-AML group. <i>Leukemia</i> , 2017, 31, 974-977.	7.2	35
50	Clinical and molecular findings in an Albanian family with familial adenomatous polyposis. <i>Genetics and Molecular Research</i> , 2017, 16, .	0.2	0
51	Research Article A targeted NGS approach to identify a c.352C>G variant in the TWIST1 gene in an Albanian family with Saethre-Chotzen syndrome. <i>Genetics and Molecular Research</i> , 2017, 16, .	0.2	0
52	Research Article AluYb8 insertion in the WNK1 gene is not associated with hypertension in a Russian Caucasian population. <i>Genetics and Molecular Research</i> , 2017, 16, .	0.2	0
53	CREB engages C/EBP β to initiate leukemogenesis. <i>Leukemia</i> , 2016, 30, 1887-1896.	7.2	28
54	Identification of the NUP98-PHF23 fusion gene in pediatric cytogenetically normal acute myeloid leukemia by whole-transcriptome sequencing. <i>Journal of Hematology and Oncology</i> , 2015, 8, 69.	17.0	14

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55	Minimal residual disease monitored after induction therapy by RQ-PCR can contribute to tailor treatment of patients with t(8;21) RUNX1-RUNX1T1 rearrangement. <i>Haematologica</i> , 2015, 100, e99-e101.	3.5	35
56	Abstract LB-211: NUP98-PHF23 is a novel fusion gene in pediatric cytogenetically normal acute myeloid leukemia. , 2015, , .		0
57	CREB Controls C/EBpd to Impair Myeloid Differentiation Process in Acute Myeloid Leukemia. <i>Blood</i> , 2015, 126, 4801-4801.	1.4	0
58	Clinical and Biological Characterization of Children with FLT3ITD Mutated Acute Myeloid Leukemia (AML): A Report from the AIEOP AML-2002 Study Group. <i>Blood</i> , 2015, 126, 3845-3845.	1.4	0
59	Core-binding factor acute myeloid leukemia in pediatric patients enrolled in the AIEOP AML 2002/01 trial: screening and prognostic impact of c-KIT mutations. <i>Leukemia</i> , 2014, 28, 1132-1134.	7.2	40
60	Whole transcriptome sequencing of a paediatric case of <i>de novo</i> acute myeloid leukaemia with del(5q) reveals <i>RUNX1</i> and <i>USP42</i> and <i>PRDM16</i> <i>SKL</i> fusion transcripts. <i>British Journal of Haematology</i> , 2014, 166, 449-452.	2.5	12
61	MLL-AF6 fusion oncogene sequesters AF6 into the nucleus to trigger RAS activation in myeloid leukemia. <i>Blood</i> , 2014, 124, 263-272.	1.4	41
62	NUP98 Fusion Proteins Are Recurrent Aberrancies in Childhood Acute Myeloid Leukemia: A Report from the AIEOP AML-2001-02 Study Group. <i>Blood</i> , 2014, 124, 1025-1025.	1.4	3
63	CBFA2T3-GLIS2 fusion transcript is a novel common feature in pediatric, cytogenetically normal AML, not restricted to FAB M7 subtype. <i>Blood</i> , 2013, 121, 3469-3472.	1.4	119
64	MicroRNA-34b promoter hypermethylation induces CREB overexpression and contributes to myeloid transformation. <i>Haematologica</i> , 2013, 98, 602-610.	3.5	42
65	DHH-RHEBL1 fusion transcript: a novel recurrent feature in the new landscape of pediatric CBFA2T3-GLIS2-positive acute myeloid leukemia. <i>Oncotarget</i> , 2013, 4, 1712-1720.	1.8	23
66	Core Binding Factor Acute Myeloid Leukemia In Pediatric Patients Enrolled In The AIEOP AML 2002/01 Trial: The Impact Of Minimal Residual Disease On Patient Outcome. <i>Blood</i> , 2013, 122, 3884-3884.	1.4	14
67	Core Binding Factor Acute Myeloid Leukemia In Pediatric Patients Of The AIEOP AML 2002/01 Trial: Screening and Prognostic Impact Of cKIT Mutations. <i>Blood</i> , 2013, 122, 2655-2655.	1.4	0
68	Sox4 cooperates with CREB in myeloid transformation. <i>Blood</i> , 2012, 120, 155-165.	1.4	43
69	Presence of high-ERG expression is an independent unfavorable prognostic marker in MLL-rearranged childhood myeloid leukemia. <i>Blood</i> , 2012, 119, 1086-1087.	1.4	16
70	Screening of novel genetic aberrations in pediatric acute myeloid leukemia: a report from the AIEOP AML-2002 study group. <i>Blood</i> , 2012, 120, 3860-3862.	1.4	11
71	BAG1 Overexpression Restrains the Anti-Apoptotic BCL2, MCL1 and HSP70 Proteins in Acute Myeloid Leukemia.. <i>Blood</i> , 2012, 120, 2492-2492.	1.4	0
72	NOVEL Recurrent Genetic Aberrations in Pediatric AML: An AIEOP AML-2002 Study Group.. <i>Blood</i> , 2012, 120, 2494-2494.	1.4	0

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73	The cAMP Response Element Binding Protein (CREB) Overexpression Induces Myeloid Transformation in Zebrafish. <i>Blood</i> , 2012, 120, 4727-4727.	1.4	0
74	ICER Evokes Dusp1-p38 Pathway Enhancing Chemotherapy Sensitivity in Myeloid Leukemia. <i>Clinical Cancer Research</i> , 2011, 17, 742-752.	7.0	6
75	MiR-34b Promoter Methylation and Regulation of CREB Expression In Myeloid Transformation. <i>Blood</i> , 2010, 116, 538-538.	1.4	1
76	miR-34b Targets Cyclic AMPâ€“Responsive Element Binding Protein in Acute Myeloid Leukemia. <i>Cancer Research</i> , 2009, 69, 2471-2478.	0.9	127
77	ICER expression inhibits leukemia phenotype and controls tumor progression. <i>Leukemia</i> , 2008, 22, 2217-2225.	7.2	20
78	Analyses of the Inducible Cyclic Adenosine 3â€²,5â€²-Monophosphate Early Repressor (ICER) and cAMP Response Element Binding Protein (CREB) in HL60 Cells: New Insight Leukemogenesis?.. <i>Blood</i> , 2006, 108, 2251-2251.	1.4	0