

Tommaso Mazza

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

163 papers	2,151 citations	25 h-index	39 g-index
191 ext. papers	2,846 ext. citations	5.8 avg, IF	4.83 L-index

#	Paper	IF	Citations
163	Mirna expression profiles identify drivers in colorectal and pancreatic cancers. <i>PLoS ONE</i> , 2012 , 7, e33663.	3.7	116
162	Snazer: the simulations and networks analyzer. <i>BMC Systems Biology</i> , 2010 , 4, 1	3.5	98
161	Loss-of-Function Mutations in APPL1 in Familial Diabetes Mellitus. <i>American Journal of Human Genetics</i> , 2015 , 97, 177-85	11	91
160	Functional variants of the HMGA1 gene and type 2 diabetes mellitus. <i>JAMA - Journal of the American Medical Association</i> , 2011 , 305, 903-12	27.4	72
159	Congruency in the prediction of pathogenic missense mutations: state-of-the-art web-based tools. <i>Briefings in Bioinformatics</i> , 2013 , 14, 448-59	13.4	65
158	DNA Hypomethylation and Histone Variant macroH2A1 Synergistically Attenuate Chemotherapy-Induced Senescence to Promote Hepatocellular Carcinoma Progression. <i>Cancer Research</i> , 2016 , 76, 594-606	10.1	58
157	Metabolomic profile in pancreatic cancer patients: a consensus-based approach to identify highly discriminating metabolites. <i>Oncotarget</i> , 2016 , 7, 5815-29	3.3	56
156	Biological and clinical manifestations of juvenile Huntington's disease: a retrospective analysis. <i>Lancet Neurology</i> , 2018 , 17, 986-993	24.1	56
155	Wnt5a Drives an Invasive Phenotype in Human Glioblastoma Stem-like Cells. <i>Cancer Research</i> , 2017 , 77, 996-1007	10.1	54
154	Fasting cycles potentiate the efficacy of gemcitabine treatment in in vitro and in vivo pancreatic cancer models. <i>Oncotarget</i> , 2015 , 6, 18545-57	3.3	50
153	Functional genome-wide siRNA screen identifies KIAA0586 as mutated in Joubert syndrome. <i>ELife</i> , 2015 , 4, e06602	8.9	49
152	Mutations in B9D1 and MKS1 cause mild Joubert syndrome: expanding the genetic overlap with the lethal ciliopathy Meckel syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 72	4.2	47
151	Induction of cancer cell stemness by depletion of macrohistone H2A1 in hepatocellular carcinoma. <i>Hepatology</i> , 2018 , 67, 636-650	11.2	46
150	Dysregulation of EGFR Pathway in EphA2 Cell Subpopulation Significantly Associates with Poor Prognosis in Colorectal Cancer. <i>Clinical Cancer Research</i> , 2017 , 23, 159-170	12.9	45
149	SIRT1-metabolite binding histone macroH2A1.1 protects hepatocytes against lipid accumulation. <i>Aging</i> , 2014 , 6, 35-47	5.6	43
148	MitImpact: an exhaustive collection of pre-computed pathogenicity predictions of human mitochondrial non-synonymous variants. <i>Human Mutation</i> , 2015 , 36, E2413-22	4.7	42
147	Mutations in CEP120 cause Joubert syndrome as well as complex ciliopathy phenotypes. <i>Journal of Medical Genetics</i> , 2016 , 53, 608-15	5.8	40

146	The Biological Clock: A Pivotal Hub in Non-alcoholic Fatty Liver Disease Pathogenesis. <i>Frontiers in Physiology</i> , 2018 , 9, 193	4.6	37
145	Clinical, neuroradiological and molecular characterization of cerebellar dysplasia with cysts (Poretti-Boltshauser syndrome). <i>European Journal of Human Genetics</i> , 2016 , 24, 1262-7	5.3	32
144	Stepwise analysis of MIR9 loci identifies miR-9-5p to be involved in Oestrogen regulated pathways in breast cancer patients. <i>Scientific Reports</i> , 2017 , 7, 45283	4.9	31
143	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. <i>PLoS Computational Biology</i> , 2017 , 13, e1005628	5	31
142	Histone variant macroH2A1 rewires carbohydrate and lipid metabolism of hepatocellular carcinoma cells towards cancer stem cells. <i>Epigenetics</i> , 2018 , 13, 829-845	5.7	28
141	TMS follow-up study in patients with vascular cognitive impairment-no dementia. <i>Neuroscience Letters</i> , 2013 , 534, 155-9	3.3	27
140	Cyto-Sim: a formal language model and stochastic simulator of membrane-enclosed biochemical processes. <i>Bioinformatics</i> , 2007 , 23, 2800-2	7.2	26
139	Establishment of stable iPS-derived human neural stem cell lines suitable for cell therapies. <i>Cell Death and Disease</i> , 2018 , 9, 937	9.8	26
138	Hypomorphic Recessive Variants in SUFU Impair the Sonic Hedgehog Pathway and Cause Joubert Syndrome with Cranio-facial and Skeletal Defects. <i>American Journal of Human Genetics</i> , 2017 , 101, 552-563	11.1	25
137	Oral-facial-digital syndrome type VI: is C5orf42 really the major gene?. <i>Human Genetics</i> , 2015 , 134, 123-66.3	6.3	24
136	Taming the complexity of biological pathways through parallel computing. <i>Briefings in Bioinformatics</i> , 2009 , 10, 278-88	13.4	24
135	EphB2 stem-related and EphA2 progression-related miRNA-based networks in progressive stages of CRC evolution: clinical significance and potential miRNA drivers. <i>Molecular Cancer</i> , 2018 , 17, 169	42.1	24
134	On Parallel Stochastic Simulation of Diffusive Systems. <i>Lecture Notes in Computer Science</i> , 2008 , 191-210.9	0.9	21
133	AURA: Atlas of UTR Regulatory Activity. <i>Bioinformatics</i> , 2012 , 28, 142-4	7.2	20
132	Loss of histone macroH2A1 in hepatocellular carcinoma cells promotes paracrine-mediated chemoresistance and CD4CD25FoxP3 regulatory T cells activation. <i>Theranostics</i> , 2020 , 10, 910-924	12.1	20
131	Infantile and childhood onset PLA2G6-associated neurodegeneration in a large North African cohort. <i>European Journal of Neurology</i> , 2015 , 22, 178-86	6	19
130	Gene expression of muscular and neuronal pathways is cooperatively dysregulated in patients with idiopathic achalasia. <i>Scientific Reports</i> , 2016 , 6, 31549	4.9	19
129	Insights From Molecular Characterization of Adult Patients of Families With Multigenerational Diabetes. <i>Diabetes</i> , 2018 , 67, 137-145	0.9	18

128	Diagnostic and Prognostic Value of Hypermethylation and Its Clinical Significance as a Novel Circulating Cell-Free DNA Biomarker in Colorectal Cancer. <i>Cancers</i> , 2019 , 11,	6.6	18
127	Using ontologies for preprocessing and mining spectra data on the Grid. <i>Future Generation Computer Systems</i> , 2007 , 23, 55-60	7.5	17
126	TRIM8-driven transcriptomic profile of neural stem cells identified glioma-related nodal genes and pathways. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2019 , 1863, 491-501	4	17
125	MicroRNAs and Long Non-Coding RNAs as Potential Candidates to Target Specific Motifs of SARS-CoV-2. <i>Non-coding RNA</i> , 2021 , 7,	7.1	17
124	Development of a metabolites risk score for one-year mortality risk prediction in pancreatic adenocarcinoma patients. <i>Oncotarget</i> , 2016 , 7, 8968-78	3.3	16
123	COL1-related overlap disorder: A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlers-Danlos syndrome overlap. <i>Clinical Genetics</i> , 2020 , 97, 396-406	4	16
122	Analysis of clock gene-miRNA correlation networks reveals candidate drivers in colorectal cancer. <i>Oncotarget</i> , 2016 , 7, 45444-45461	3.3	15
121	Molecular dynamics recipes for genome research. <i>Briefings in Bioinformatics</i> , 2018 , 19, 853-862	13.4	14
120	Circadian transcriptome analysis in human fibroblasts from Hunter syndrome and impact of iduronate-2-sulfatase treatment. <i>BMC Medical Genomics</i> , 2013 , 6, 37	3.7	14
119	Deficiency and haploinsufficiency of histone macroH2A1.1 in mice recapitulate hematopoietic defects of human myelodysplastic syndrome. <i>Clinical Epigenetics</i> , 2019 , 11, 121	7.7	13
118	Novel ßActin Gene Mutation p.(Ala21Val) Causing Familial Hypertrophic Cardiomyopathy, Myocardial Noncompaction, and Transmural Crypts. Clinical-Pathologic Correlation. <i>Journal of the American Heart Association</i> , 2018 , 7,	6	13
117	Inflammatory Bowel Disease Meets Systems Biology: A Multi-Omics Challenge and Frontier. <i>OMICS A Journal of Integrative Biology</i> , 2016 , 20, 692-698	3.8	13
116	Clinical Significance of Circulating miR-1273g-3p and miR-122-5p in Pancreatic Cancer. <i>Frontiers in Oncology</i> , 2020 , 10, 44	5.3	12
115	The association of variants in PNPLA3 and GRP78 and the risk of developing hepatocellular carcinoma in an Italian population. <i>Oncotarget</i> , 2016 , 7, 86791-86802	3.3	12
114	Between SCA5 and SCAR14: delineation of the SPTBN2 p.R480W-associated phenotype. <i>European Journal of Human Genetics</i> , 2018 , 26, 928-929	5.3	12
113	Hsa-miR-210-3p expression in breast cancer and its putative association with worse outcome in patients treated with Docetaxel. <i>Scientific Reports</i> , 2019 , 9, 14913	4.9	11
112	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a PDCD10 large deletion. <i>Human Mutation</i> , 2018 , 39, 1885-1900	4.7	11
111	Heterozygous missense mutations in NFATC1 are associated with atrioventricular septal defect. <i>Human Mutation</i> , 2018 , 39, 1428-1441	4.7	10

110	Systematic Analysis of Mouse Genome Reveals Distinct Evolutionary and Functional Properties Among Circadian and Ultradian Genes. <i>Frontiers in Physiology</i> , 2018 , 9, 1178	4.6	10
109	Are Gaming-Enabled Graphic Processing Unit Cards Convenient for Molecular Dynamics Simulation?. <i>Evolutionary Bioinformatics</i> , 2019 , 15, 1176934319850144	1.9	9
108	A Role for the Biological Clock in Liver Cancer. <i>Cancers</i> , 2019 , 11,	6.6	9
107	t(15;21) translocations leading to the concurrent downregulation of RUNX1 and its transcription factor partner genes SIN3A and TCF12 in myeloid disorders. <i>Molecular Cancer</i> , 2015 , 14, 211	42.1	9
106	Estimating the divisibility of complex biological networks by sparseness indices. <i>Briefings in Bioinformatics</i> , 2010 , 11, 364-74	13.4	9
105	Age-related obesity and type 2 diabetes dysregulate neuronal associated genes and proteins in humans. <i>Oncotarget</i> , 2015 , 6, 29818-32	3.3	9
104	Efficient Parallel Statistical Model Checking of Biochemical Networks. <i>Electronic Proceedings in Theoretical Computer Science</i> , EPTCS , 14, 47-61		9
103	TRIM8 interacts with KIF11 and KIFC1 and controls bipolar spindle formation and chromosomal stability. <i>Cancer Letters</i> , 2020 , 473, 98-106	9.9	9
102	Multifaceted enrichment analysis of RNA-RNA crosstalk reveals cooperating micro-societies in human colorectal cancer. <i>Nucleic Acids Research</i> , 2016 , 44, 4025-36	20.1	9
101	Genetic characterization of suspected MODY patients in Tunisia by targeted next-generation sequencing. <i>Acta Diabetologica</i> , 2019 , 56, 515-523	3.9	9
100	Impact of genetic polymorphisms on the pathogenesis of idiopathic achalasia: Association with IL33 gene variant. <i>Human Immunology</i> , 2014 , 75, 364-9	2.3	8
99	Studying Irreversible Transitions in a Model of Cell Cycle Regulation. <i>Electronic Notes in Theoretical Computer Science</i> , 2009 , 232, 39-53	0.7	8
98	A primary tumor gene expression signature identifies a crucial role played by tumor stroma myofibroblasts in lymph node involvement in oral squamous cell carcinoma. <i>Oncotarget</i> , 2017 , 8, 104913-104927	3.3	8
97	Hsa-miR-155-5p Up-Regulation in Breast Cancer and Its Relevance for Treatment With Poly[ADP-Ribose] Polymerase 1 (PARP-1) Inhibitors. <i>Frontiers in Oncology</i> , 2020 , 10, 1415	5.3	8
96	Association Between MICA Gene Variants and the Risk of Hepatitis C Virus-Induced Hepatocellular Cancer in a Sicilian Population Sample. <i>OMICS A Journal of Integrative Biology</i> , 2018 , 22, 274-282	3.8	7
95	A solid quality-control analysis of AB SOLiD short-read sequencing data. <i>Briefings in Bioinformatics</i> , 2013 , 14, 684-95	13.4	7
94	The Relevance of Topology in Parallel Simulation of Biological Networks. <i>IEEE/ACM Transactions on Computational Biology and Bioinformatics</i> , 2012 , 9, 911-923	3	7
93	Preprocessing of mass spectrometry proteomics data on the grid		7

92	"Fork and bracket" syndrome expands the spectrum of SBF1-related sensory motor polyneuropathies. <i>Neurology: Genetics</i> , 2016 , 2, e61	3.8	7
91	Cardiac valvular Ehlers-Danlos syndrome is a well-defined condition due to recessive null variants in COL1A2. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 846-851	2.5	7
90	Sudden death in mild hypertrophic cardiomyopathy with compound DSG2/DSC2/MYH6 mutations: Revisiting phenotype after genetic assessment in a master runner athlete. <i>Journal of Electrocardiology</i> , 2019 , 53, 95-99	1.4	7
89	Mitotic Oscillators as MP Graphs. <i>Lecture Notes in Computer Science</i> , 2006 , 382-394	0.9	7
88	Epigenetically induced ectopic expression of UNCX impairs the proliferation and differentiation of myeloid cells. <i>Haematologica</i> , 2017 , 102, 1204-1214	6.6	6
87	Clinical and functional characterization of a novel RASopathy-causing SHOC2 mutation associated with prenatal-onset hypertrophic cardiomyopathy. <i>Human Mutation</i> , 2019 , 40, 1046-1056	4.7	6
86	Identification of p53-target genes in Danio rerio. <i>Scientific Reports</i> , 2016 , 6, 32474	4.9	6
85	Sudden cardiac death in J wave syndrome with short QT associated to a novel mutation in Na 1.8 coding gene SCN10A: First case report for a possible pharmacogenomic role. <i>Journal of Electrocardiology</i> , 2018 , 51, 809-813	1.4	6
84	Functional Impact of Autophagy-Related Genes on the Homeostasis and Dynamics of Pancreatic Cancer Cell Lines. <i>IEEE/ACM Transactions on Computational Biology and Bioinformatics</i> , 2015 , 12, 667-783	3	6
83	New Method for Measuring Angle-Resolved Phases in Photoemission. <i>Physical Review X</i> , 2020 , 10,	9.1	6
82	GDF11 induces mild hepatic fibrosis independent of metabolic health. <i>Aging</i> , 2020 , 12, 20024-20046	5.6	6
81	A new case of SMABF2 diagnosed in stillbirth expands the prenatal presentation and mutational spectrum of ASCC1. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 508-512	2.5	6
80	A Broad Overview of Computational Methods for Predicting the Pathophysiological Effects of Non-synonymous Variants. <i>Methods in Molecular Biology</i> , 2016 , 1415, 423-40	1.4	6
79	Novel Variants in Two Italian Patients with Classical-Like Ehlers-Danlos Syndrome. <i>Genes</i> , 2019 , 10,	4.2	6
78	HbA1c levels in patients with gestational diabetes mellitus: Relationship with pre-pregnancy BMI and pregnancy outcome. <i>Journal of Endocrinological Investigation</i> , 2013 , 36, 1038-45	5.2	6
77	The striatal-enriched protein Rhes is a critical modulator of cocaine-induced molecular and behavioral responses. <i>Scientific Reports</i> , 2019 , 9, 15294	4.9	5
76	The biological clock and the molecular basis of lysosomal storage diseases. <i>JIMD Reports</i> , 2015 , 18, 93-105	5	
75	Estimating the global density of graphs by a sparseness index. <i>Applied Mathematics and Computation</i> , 2013 , 224, 346-357	2.7	5

74	When to test fetuses for RASopathies? Proposition from a systematic analysis of 352 multicenter cases and a postnatal cohort. <i>Genetics in Medicine</i> , 2021 , 23, 1116-1124	8.1	5
73	Food Web Topology and Nested Keystone Species Complexes. <i>Complexity</i> , 2018 , 2018, 1-8	1.6	5
72	Putative TMPRSS3/GJB2 digenic inheritance of hearing loss detected by targeted resequencing. <i>Molecular and Cellular Probes</i> , 2017 , 33, 24-27	3.3	4
71	Genomic and physiological resilience in extreme environments are associated with a secure attachment style. <i>Translational Psychiatry</i> , 2020 , 10, 185	8.6	4
70	Customised next-generation sequencing multigene panel to screen a large cohort of individuals with chromatin-related disorder. <i>Journal of Medical Genetics</i> , 2020 , 57, 760-768	5.8	4
69	Very mild features of dysequilibrium syndrome associated with a novel VLDLR missense mutation. <i>Neurogenetics</i> , 2016 , 17, 191-5	3	4
68	Agenesis of the putamen and globus pallidus caused by recessive mutations in the homeobox gene GSX2. <i>Brain</i> , 2019 , 142, 2965-2978	11.2	4
67	Association of a homozygous GCK missense mutation with mild diabetes. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e00728	2.3	4
66	Cell Cycle and Tumor Growth in Membrane Systems with Peripheral Proteins. <i>Electronic Notes in Theoretical Computer Science</i> , 2009 , 227, 127-141	0.7	4
65	Managing ontologies for Grid computing. <i>Multiagent and Grid Systems</i> , 2006 , 2, 29-44	0.5	4
64	MicroRNA co-expression networks exhibit increased complexity in pancreatic ductal compared to Vater's papilla adenocarcinoma. <i>Oncotarget</i> , 2017 , 8, 105320-105339	3.3	4
63	Pyntacle: a parallel computing-enabled framework for large-scale network biology analysis. <i>GigaScience</i> , 2020 , 9,	7.6	4
62	Double missense mutations in cardiac myosin-binding protein C and myopalladin genes: A case report with diffuse coronary disease, complete atrioventricular block, and progression to dilated cardiomyopathy. <i>Annals of Noninvasive Electrophysiology</i> , 2020 , 25, e12687	1.5	4
61	A Lipidomic Signature Complements Stemness Features Acquisition in Liver Cancer Cells. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	4
60	Early-Onset Diabetes as Risk Factor for Pancreatic Cancer: miRNA Expression Profiling in Plasma Uncovers a Role for miR-20b-5p, miR-29a, and miR-18a-5p in Diabetes of Recent Diagnosis. <i>Frontiers in Oncology</i> , 2020 , 10, 1567	5.3	4
59	MitImpact 3: modeling the residue interaction network of the Respiratory Chain subunits. <i>Nucleic Acids Research</i> , 2021 , 49, D1282-D1288	20.1	4
58	Mono-ADP-Ribosylhydrolase MACROD2 Is Dispensable for Murine Responses to Metabolic and Genotoxic Insults. <i>Frontiers in Genetics</i> , 2018 , 9, 654	4.5	4
57	A novel mutation in CDH11, encoding cadherin-11, cause Branchioskeletogenital (Elsahy-Waters) syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 2028-2033	2.5	4

56	A Multi-Layered Study on Harmonic Oscillations in Mammalian Genomics and Proteomics. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	3
55	The recurrent SETBP1 c.2608G > A, p.(Gly870Ser) variant in a patient with Schinzel-Giedion syndrome: an illustrative case of the utility of whole exome sequencing in a critically ill neonate. <i>Italian Journal of Pediatrics</i> , 2020 , 46, 74	3.2	3
54	Analysis of MTNR1B gene polymorphisms in relationship with IRS2 gene variants, epicardial fat thickness, glucose homeostasis and cognitive performance in the elderly. <i>Chronobiology International</i> , 2017 , 34, 1083-1093	3.6	3
53	On the Preprocessing of Mass Spectrometry Proteomics Data. <i>Lecture Notes in Computer Science</i> , 2006 , 127-131	0.9	3
52	Nociceptin/orphanin FQ opioid receptor (NOP) selective ligand MCOPPB links anxiolytic and senolytic effects. <i>GeroScience</i> , 2021 , 1	8.9	3
51	Towards a Complete Covering of SBML Functionalities 2007 , 353-366		3
50	Prenatal whole exome sequencing detects a new homozygous fukutin (FKTN) mutation in a fetus with an ultrasound suspicion of familial Dandy-Walker malformation. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1054	2.3	3
49	Epigenetic Scanning of CpG Sites Uncovers New Molecular-Driven Patterns in Lung Adeno and Squamous Cell Carcinomas. <i>Antioxidants</i> , 2020 , 9,	7.1	3
48	Mechanisms of pathogenesis of missense mutations on the KDM6A-H3 interaction in type 2 Kabuki Syndrome. <i>Computational and Structural Biotechnology Journal</i> , 2020 , 18, 2033-2042	6.8	3
47	Refining the mutational spectrum and gene-phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. <i>Journal of Medical Genetics</i> , 2021 ,	5.8	3
46	Systemic depletion of histone macroH2A1.1 boosts hippocampal synaptic plasticity and social behavior in mice. <i>FASEB Journal</i> , 2021 , 35, e21793	0.9	3
45	Expanding the phenotype associated to KMT2A variants: overlapping clinical signs between Wiedemann-Steiner and Rubinstein-Taybi syndromes. <i>European Journal of Human Genetics</i> , 2021 , 29, 88-98	5.3	3
44	Whole Exome Sequencing Reveals a Novel In-Frame Deletion in a Boy with Global Developmental Delay, Absent Speech, Dysmorphic Features, and Cerebral Anomalies. <i>Genes</i> , 2021 , 12,	4.2	3
43	Potential Prognostic Role of Methylation in Non-Small-Cell Lung Cancer. <i>Cells</i> , 2020 , 9,	7.9	2
42	Concurrent chromothripsis events in a case of TP53 depleted acute myeloid leukemia with myelodysplasia-related changes. <i>Cancer Genetics</i> , 2019 , 237, 63-68	2.3	2
41	Molecular diagnostic workflow, clinical interpretation of sequence variants, and data repository procedures in 140 individuals with familial cerebral cavernous malformations. <i>Human Mutation</i> , 2019 , 40, e24-e36	4.7	2
40	Affinity analysis of differentially expressed genes in hepatocytes expressing HCV core genotype 1b or 3a. <i>BioSystems</i> , 2013 , 114, 64-8	1.9	2
39	Predicting the Effects of Parameters Changes in Stochastic Models through Parallel Synthetic Experiments and Multivariate Analysis 2010 ,		2

38	Algorithms and databases in bioinformatics: towards a proteomic ontology 2005 ,		2
37	Familial Hemiplegic Migraine: A New Gene in an Italian Family. <i>Archives of Clinical and Medical Case Reports</i> , 2019 , 03,	1.3	2
36	Stability Analysis of Biological Network Topologies during Stochastic Simulation 2011 ,		2
35	microRNA-mRNA network model in patients with achalasia. <i>Neurogastroenterology and Motility</i> , 2020 , 32, e13764	4	2
34	GDF11 rapidly increases lipid accumulation in liver cancer cells through ALK5-dependent signaling. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 2021 , 1866, 158920	5	2
33	GDF5 mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. <i>Bone</i> , 2021 , 144, 115803	4.7	2
32	Beyond COVID-19 pandemic: Topology-aware optimization of vaccination strategy for minimizing virus spreading. <i>Computational and Structural Biotechnology Journal</i> , 2022 , 20, 2664-2671	6.8	2
31	Insights into the molecular pathogenesis of cardiospondylocarpofacial syndrome: MAP3K7 c.737-7A>G variant alters the TGF β -mediated β 5MA cytoskeleton assembly and autophagy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2020 , 1866, 165742	6.9	1
30	Compound Phenotype Due to Recessive Variants in and Genes Disclosed by an Integrated Approach of SNP-Array and Whole Exome Sequencing. <i>Genes</i> , 2020 , 11,	4.2	1
29	Multi-Sided Compression Performance Assessment of ABI SOLiD WES Data. <i>Algorithms</i> , 2013 , 6, 309-318	1.8	1
28	A (Natural) Computing Perspective on Cellular Processes 2010 , 115-140		1
27	Fastest parallel molecular algorithms for the elliptic curve discrete logarithm problem over GF(2n) 2009 ,		1
26	Modeling and Simulation of Smart and Green Computing Systems. <i>Computer</i> , 2012 , 45, 22-23	1.6	1
25	COVID-19 Specific Immune Markers Revealed by Single Cell Phenotypic Profiling.. <i>Biomedicines</i> , 2021 , 9,	4.8	1
24	Malate Dehydrogenase 2 (MDH2) as a New Diabetogene Causing Hyperglycemia in Families with Multigenerational Diabetes. <i>Diabetes</i> , 2018 , 67, 262-OR	0.9	1
23	Gain of function of Malate Dehydrogenase 2 (MDH2) and familial hyperglycemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 ,	5.6	1
22	Statistical Model Checking of Membrane Systems with Peripheral Proteins: Quantifying the Role of Estrogen in Cellular Mitosis and DNA Damage. <i>Emergence, Complexity and Computation</i> , 2014 , 43-63	0.1	1
21	Tracing the mutated HTT and haplotype of the African ancestor who spread Huntington disease into the Middle East. <i>Genetics in Medicine</i> , 2020 , 22, 1903-1908	8.1	1

20	Heterozygous nonsense ARX mutation in a family highlights the complexity of clinical and molecular diagnosis in case of chromosomal and single gene disorder co-inheritance. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1336	2.3	1
19	Incidence and prevalence of Huntington disease (HD) in the Sultanate of Oman: the first Middle East post- service-based study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020 , 91, 1359-1360	5.5	1
18	Neuroendocrine-Related Circulating Transcripts in Small-Cell Lung Cancers: Detection Methods and Future Perspectives. <i>Cancers</i> , 2021 , 13,	6.6	1
17	Phenotypic Variability of a Pathogenic Mutation in an Italian Family Affected by Arrhythmogenic Cardiomyopathy and Juvenile Sudden Death: Considerations From Molecular Autopsy to Sport Restriction. <i>Frontiers in Cardiovascular Medicine</i> , 2021 , 8, 635141	5.4	1
16	A Serum Resistin and Multicytokine Inflammatory Pathway Is Linked With and Helps Predict All-cause Death in Diabetes. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, e4350-e4359	5.6	1
15	Genome-Wide DNA Methylation Analysis of a Cohort of 41 Patients Affected by Oculo-Auriculo-Vertebral Spectrum (OAVS). <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	1
14	Germline Alterations in Patients With IBD-associated Colorectal Cancer. <i>Inflammatory Bowel Diseases</i> , 2021 ,	4.5	1
13	The Histone Variant MacroH2A1 Impacts Circadian Gene Expression and Cell Phenotype in an In Vitro Model of Hepatocellular Carcinoma. <i>Biomedicines</i> , 2021 , 9,	4.8	1
12	Histone Variant macroH2A1.1 Enhances Nonhomologous End Joining-dependent DNA Double-strand-break Repair and Reprogramming Efficiency of Human iPSCs.. <i>Stem Cells</i> , 2022 , 40, 35-48	5.8	0
11	Pathogenic variants of MODY-genes in adult patients with early-onset type 2 diabetes.. <i>Acta Diabetologica</i> , 2022 , 1	3.9	0
10	Mining potentially actionable kinase gene fusions in cancer cell lines with the KuNG FU database. <i>Scientific Data</i> , 2020 , 7, 420	8.2	0
9	Novel Frameshift Mutation in a Patient Affected by a Syndromic Form of Neurodevelopmental Disorder. <i>Genes</i> , 2021 , 12,	4.2	0
8	Contribution of rare variants in monogenic diabetes-genes to early-onset type 2 diabetes.. <i>Diabetes and Metabolism</i> , 2022 , 101353	5.4	0
7	High performance computational systems biology. <i>IEEE/ACM Transactions on Computational Biology and Bioinformatics</i> , 2012 , 9, 641-2	3	
6	Using PROTEUS for Modeling Data Mining Analysis of Proteomics Experiments on the Grid. <i>Lecture Notes in Computer Science</i> , 2004 , 232-243	0.9	
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