Stefano Castellana

List of Publications by Year
in descending order

64	968	623574 14	526166 27
papers	citations	h-index	g-index

65	65	65	2223
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Evolutionary Patterns of the Mitochondrial Genome in Metazoa: Exploring the Role of Mutation and Selection in Mitochondrial Proteinâ€“Coding Genes. Genome Biology and Evolution, 2011, 3, 1067-1079.	1.1	139
2	Congruency in the prediction of pathogenic missense mutations: state-of-the-art web-based tools. Briefings in Bioinformatics, 2013, 14, 448-459.	3.2	79
3	Mutations in B9D1 and MKS1 cause mild Joubert syndrome: expanding the genetic overlap with the lethal ciliopathy Meckel syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 72.	1.2	63
4	MitImpact: an Exhaustive Collection of Pre-computed Pathogenicity Predictions of Human Mitochondrial Non-synonymous Variants. Human Mutation, 2015, 36, E2413-E2422.	1.1	61
5	High-confidence assessment of functional impact of human mitochondrial non-synonymous genome variations by APOGEE. PLoS Computational Biology, 2017, 13, e1005628.	1.5	54
6	Stepwise analysis of MIR9 loci identifies miR-9-5p to be involved in Oestrogen regulated pathways in breast cancer patients. Scientific Reports, 2017, 7, 45283.	1.6	45
7	Gene code CD274/PD-L1: from molecular basis toward cancer immunotherapy. Therapeutic Advances in Medical Oncology, 2018, 10, 175883591881559.	1.4	38
8	The RHNumtS compilation: Features and bioinformatics approaches to locate and quantify Human NumtS. BMC Genomics, 2008, 9, 267.	1.2	34
9	Expanding the mutation spectrum in 130 probands with ARPKD: identification of 62 novel PKHD1 mutations by sanger sequencing and MLPA analysis. Journal of Human Genetics, 2016, 61, 811-821.	1.1	27
10	Infantile and childhood onset <scp>PLA</scp>2G6â€associated neurodegeneration in a large North African cohort. European Journal of Neurology, 2015, 22, 178-186.	1.7	25
11	Molecular dynamics recipes for genome research. Briefings in Bioinformatics, 2018, 19, 853-862.	3.2	23
12	TRIM8-driven transcriptomic profile of neural stem cells identified glioma-related nodal genes and pathways. Biochimica Et Biophysica Acta - General Subjects, 2019, 1863, 491-501.	1.1	22
13	MitImpact 3: modeling the residue interaction network of the Respiratory Chain subunits. Nucleic Acids Research, 2021, 49, D1282-D1288.	6.5	21
14	Systematic Analysis of Mouse Genome Reveals Distinct Evolutionary and Functional Properties Among Circadian and Ultradian Genes. Frontiers in Physiology, 2018, 9, 1178.	1.3	19
15	Inflammatory Bowel Disease Meets Systems Biology: A Multi-Omics Challenge and Frontier. OMICS A Journal of Integrative Biology, 2016, 20, 692-698.	1.0	16
16	A single-center study on 140 patients with cerebral cavernous malformations: 28 new pathogenic variants and functional characterization of a PDCD10 large deletion. Human Mutation, 2018, 39, 1885-1900.	1.1	16
17	Customised next-generation sequencing multigene panel to screen a large cohort of individuals with chromatin-related disorder. Journal of Medical Genetics, 2020, 57, 760-768.	1.5	15
18	Multifaceted enrichment analysis of RNAâ€“RNA crosstalk reveals cooperating micro-societies in human colorectal cancer. Nucleic Acids Research, 2016, 44, 4025-4036.	6.5	14

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19	A novel mutation in CDH11, encoding cadherinâ€11, cause Branchioskeletogenital (Elsahyâ€Waters) syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 2028-2033.	0.7	13
20	Are Gaming-Enabled Graphic Processing Unit Cards Convenient for Molecular Dynamics Simulation?. Evolutionary Bioinformatics, 2019, 15, 117693431985014.	0.6	13
21	A primary tumor gene expression signature identifies a crucial role played by tumor stroma myofibroblasts in lymph node involvement in oral squamous cell carcinoma. Oncotarget, 2017, 8, 104913-104927.	0.8	12
22	Exon-Trapping Assay Improves Clinical Interpretation of COL11A1 and COL11A2 Intronic Variants in Stickler Syndrome Type 2 and Otospondylomegaepiphyseal Dysplasia. Genes, 2020, 11, 1513.	1.0	11
23	Expanding the phenotype associated to KMT2A variants: overlapping clinical signs between Wiedemannâ€“Steiner and Rubinsteinâ€“Taybi syndromes. European Journal of Human Genetics, 2021, 29, 88-98.	1.4	11
24	Pyntacle: a parallel computing-enabled framework for large-scale network biology analysis. GigaScience, 2020, 9, .	3.3	11
25	Identification of p53-target genes in Danio rerio. Scientific Reports, 2016, 6, 32474.	1.6	10
26	Sudden cardiac death in J wave syndrome with short QT associated to a novel mutation in Nav 1.8 coding gene SCN10A: First case report for a possible pharmacogenomic role. Journal of Electrocardiology, 2018, 51, 809-813.	0.4	10
27	Sudden death in mild hypertrophic cardiomyopathy with compound DSG2/DSC2/MYH6 mutations: Revisiting phenotype after genetic assessment in a master runner athlete. Journal of Electrocardiology, 2019, 53, 95-99.	0.4	10
28	A Multi-Layered Study on Harmonic Oscillations in Mammalian Genomics and Proteomics. International Journal of Molecular Sciences, 2019, 20, 4585.	1.8	9
29	Mechanisms of pathogenesis of missense mutations on the KDM6A-H3 interaction in type 2 Kabuki Syndrome. Computational and Structural Biotechnology Journal, 2020, 18, 2033-2042.	1.9	9
30	Microbiome Analysis of Mucosal Ileoanal Pouch in Ulcerative Colitis Patients Revealed Impairment of the Pouches Immunometabolites. Cells, 2021, 10, 3243.	1.8	9
31	Real-time, computer-aided, detection-assisted colonoscopy eliminates differences in adenoma detection rate between trainee and experienced endoscopists. Endoscopy International Open, 2022, 10, E616-E621.	0.9	9
32	A solid quality-control analysis of AB SOLiD short-read sequencing data. Briefings in Bioinformatics, 2013, 14, 684-695.	3.2	8
33	Putative TMPRSS3/GJB2 digenic inheritance of hearing loss detected by targeted resequencing. Molecular and Cellular Probes, 2017, 33, 24-27.	0.9	8
34	Whole Exome Sequencing Reveals a Novel AUTS2 In-Frame Deletion in a Boy with Global Developmental Delay, Absent Speech, Dysmorphic Features, and Cerebral Anomalies. Genes, 2021, 12, 229.	1.0	8
35	The Biological Clock and the Molecular Basis of Lysosomal Storage Diseases. JIMD Reports, 2014, 18, 93-105.	0.7	7
36	A Broad Overview of Computational Methods for Predicting the Pathophysiological Effects of Non-synonymous Variants. Methods in Molecular Biology, 2016, 1415, 423-440.	0.4	7

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37	Double missense mutations in cardiac myosinâ€binding protein C and myopalladin genes: A case report with diffuse coronary disease, complete atrioventricular block, and progression to dilated cardiomyopathy. Annals of Noninvasive Electrocardiology, 2020, 25, e12687.	0.5	7
38	Epigenetic Scanning of KEAP1 CpG Sites Uncovers New Molecular-Driven Patterns in Lung Adeno and Squamous Cell Carcinomas. Antioxidants, 2020, 9, 904.	2.2	7
39	Healthy and pro-inflammatory gut ecology plays a crucial role in the digestion and tolerance of a novel Gluten Friendlyâ„¢ bread in celiac subjects: a randomized, double blind, placebo controlin vivostudy. Food and Function, 2022, 13, 1299-1315.	2.1	7
40	The recurrent SETBP1 c.2608Gâ€‰>â€‰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. Italian Journal of Pediatrics, 2020, 46, 74.	1.0	6
41	Germline Alterations in Patients With IBD-associated Colorectal Cancer. Inflammatory Bowel Diseases, 2022, 28, 447-454.	0.9	6
42	Association of a homozygous GCK missense mutation with mild diabetes. Molecular Genetics & Genomic Medicine, 2019, 7, e00728.	0.6	5
43	Rare Somatic MEN1 Gene Pathogenic Variant in a Patient Affected by Atypical Parathyroid Adenoma. International Journal of Endocrinology, 2020, 2020, 1-5.	0.6	4
44	Phenotypic Variability of a Pathogenic PKP2 Mutation in an Italian Family Affected by Arrhythmogenic Cardiomyopathy and Juvenile Sudden Death: Considerations From Molecular Autopsy to Sport Restriction. Frontiers in Cardiovascular Medicine, 2021, 8, 635141.	1.1	4
45	A comparative benchmark of classic DNA motif discovery tools on synthetic data. Briefings in Bioinformatics, 2021, 22, .	3.2	4
46	Novel TONSL variants cause SPONASTRIME dysplasia and associate with spontaneous chromosome breaks, defective cell proliferation and apoptosis. Human Molecular Genetics, 2020, 29, 3122-3131.	1.4	3
47	Transcriptome and Gene Fusion Analysis of Synchronous Lesions Reveals lncMRPS31P5 as a Novel Transcript Involved in Colorectal Cancer. International Journal of Molecular Sciences, 2020, 21, 7120.	1.8	3
48	Compound Phenotype Due to Recessive Variants in LARP7 and OTOG Genes Disclosed by an Integrated Approach of SNP-Array and Whole Exome Sequencing. Genes, 2020, 11, 379.	1.0	3
49	Genome-wide DNA methylation profiling and exome sequencing resolved a long-time misdiagnosed case. Journal of Human Genetics, 2022, 67, 547-551.	1.1	3
50	KDM6A missense variants hamper H3 histone demethylation in lung squamous cell carcinoma. Computational and Structural Biotechnology Journal, 2022, 20, 3151-3160.	1.9	3
51	Affinity analysis of differentially expressed genes in hepatocytes expressing HCV core genotype 1b or 3a. BioSystems, 2013, 114, 64-68.	0.9	2
52	Multi-Sided Compression Performance Assessment of ABI SOLiD WES Data. Algorithms, 2013, 6, 309-318.	1.2	2
53	REDOXI-miRNA of Keap1/Nrf2 axis in lung tumors. Annals of Oncology, 2019, 30, ii5.	0.6	2
54	Novel Pathogenic Variants of the AIRE Gene in Two Autoimmune Polyendocrine Syndrome Type I Cases with Atypical Presentation: Role of the NGS in Diagnostic Pathway and Review of the Literature. Biomedicines, 2020, 8, 631.	1.4	2

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55	Novel STAG1 Frameshift Mutation in a Patient Affected by a Syndromic Form of Neurodevelopmental Disorder. Genes, 2021, 12, 1116.	1.0	2
56	Familial Hemiplegic Migraine: A New Gene in an Italian Family. Archives of Clinical and Medical Case Reports, 2019, 03, .	0.0	2
57	Expanding the Clinical and Molecular Spectrum of PSMD12-Related Neurodevelopmental Syndrome: An Additional Patient and Review. Archives of Clinical and Medical Case Reports, 2019, 03, .	0.0	2
58	Review of clinical and molecular variability in autosomal recessive cutis laxa 2A. American Journal of Medical Genetics, Part A, 2021, 185, 955-965.	0.7	2
59	Pharmacogenomics of Pediatric Cardiac Arrest: Cisplatin Treatment Worsened by a Ryanodine Receptor 2 Gene Mutation. Neurology International, 2022, 12, 80-88.	0.2	1
60	P844 NGS to investigate genetic predisposition to colon cancer in patients with colon cancer over inflammatory bowel disease. Journal of Crohn's and Colitis, 2018, 12, S543-S543.	0.6	0
61	OC.04.3 GENETIC PREDISPOSITION TO COLON CANCER IN PATIENTS WITH INFLAMMATORY BOWEL DISEASE: NGS APPROACH TO IDENTIFY HIGH RISK PATIENTS. Digestive and Liver Disease, 2019, 51, e87.	0.4	0
62	On the impact of short-reads quality on variants detection. EMBnet Journal, 2012, 18, 22.	0.2	0
63	REAL-TIME COMPUTER AIDED DETECTION-ASSISTED COLONOSCOPY ELIMINATES DIFFERENCES IN ADENOMA DETECTION RATE BETWEEN TRAINEE AND EXPERIENCED ENDOSCOPISTS. Endoscopy, 2022, 54, .	1.0	0
64	T.05.8 REAL-TIME ARTIFICIAL INTELLIGENCE-AIDED COLONOSCOPY ELIMINATES DIFFERENCES IN ADENOMA DETECTION RATE BETWEEN TRAINEES AND EXPERIENCED ENDOSCOPISTS IN TANDEM-COLONOSCOPIES. Digestive and Liver Disease, 2022, 54, S135-S136.	0.4	0