Chiara Di Resta

List of Publications by Year
in descending order

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47 papers	1,372 citations	361413 20 h-index	345221 36 g-index
49 all docs	49 docs citations	49 times ranked	2298 citing authors

#	Article	IF	Citations
1	Increased Sensitivity of the Neuronal Nicotinic Receptor Î±2 Subunit Causes Familial Epilepsy with Nocturnal Wandering and Ictal Fear. American Journal of Human Genetics, 2006, 79, 342-350.	6.2	225
2	Development, evaluation, and validation of machine learning models for COVID-19 detection based on routine blood tests. Clinical Chemistry and Laboratory Medicine, 2021, 59, 421-431.	2.3	109
3	Next-generation sequencing approach for the diagnosis of human diseases: open challenges and new opportunities. Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine, 2018, 29, 4-14.	0.7	71
4	Pharmacogenomics education in medical and pharmacy schools: conclusions of a global survey. Pharmacogenomics, 2019, 20, 643-657.	1.3	65
5	Brugada syndrome genetics is associated with phenotype severity. European Heart Journal, 2021, 42, 1082-1090.	2.2	59
6	Genetics can contribute to the prognosis of Brugada syndrome: a pilot model for risk stratification. European Journal of Human Genetics, 2013, 21, 911-917.	2.8	58
7	The Gender Impact Assessment among Healthcare Workers in the SARS-CoV-2 Vaccinationâ€"An Analysis of Serological Response and Side Effects. Vaccines, 2021, 9, 522.	4.4	52
8	A Brugada syndrome mutation (p.S216L) and its modulation by p.H558R polymorphism: standard and dynamic characterization. Cardiovascular Research, 2011, 91, 606-616.	3.8	50
9	Exome sequencing and pathway analysis for identification of genetic variability relevant for bronchopulmonary dysplasia (BPD) in preterm newborns: A pilot study. Clinica Chimica Acta, 2015, 451, 39-45.	1.1	49
10	Updated clinical overview on cardiac laminopathies: an electrical and mechanical disease. Nucleus, 2018, 9, 380-391.	2.2	36
11	High-throughput genetic characterization of a cohort of Brugada syndrome patients. Human Molecular Genetics, 2015, 24, 5828-5835.	2.9	35
12	Personalized laboratory medicine: a patient-centered future approach. Clinical Chemistry and Laboratory Medicine, 2018, 56, 1981-1991.	2.3	33
13	Cardiac and Neuromuscular Features of Patients WithLMNA-Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458.	3.9	33
14	Impaired turnover of hyperfused mitochondria in severe axonal neuropathy due to a novel DRP1 mutation. Human Molecular Genetics, 2020, 29, 177-188.	2.9	30
15	Next Generation Sequencing: From Research Area to Clinical Practice. Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine, 2018, 29, 215-220.	0.7	29
16	Harmonization of six quantitative SARS-CoV-2 serological assays using sera of vaccinated subjects. Clinica Chimica Acta, 2021, 522, 144-151.	1.1	28
17	Effect of carbamazepine and oxcarbazepine on wild-type and mutant neuronal nicotinic acetylcholine receptors linked to nocturnal frontal lobe epilepsy. European Journal of Pharmacology, 2010, 643, 13-20.	3.5	24
18	Antibody Titer Kinetics and SARS-CoV-2 Infections Six Months after Administration with the BNT162b2 Vaccine. Vaccines, 2021, 9, 1357.	4.4	24

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19	Introduction to Ion Channels. Advances in Experimental Medicine and Biology, 2010, 674, 9-21.	1.6	21
20	Late gadolinium enhancement role in arrhythmic risk stratification of patients with LMNA cardiomyopathy: results from a long-term follow-up multicentre study. Europace, 2020, 22, 1864-1872.	1.7	21
21	Long-term antibody persistence and exceptional vaccination response on previously SARS-CoV-2 infected subjects. Vaccine, 2021, 39, 4256-4260.	3.8	20
22	Transcriptional role of androgen receptor in the expression of long non-coding RNA Sox2OT in neurogenesis. PLoS ONE, 2017, 12, e0180579.	2.5	19
23	Is laboratory medicine ready for the era of personalized medicine? A survey addressed to laboratory directors of hospitals/academic schools of medicine in Europe. Clinical Chemistry and Laboratory Medicine, 2015, 53, 981-8.	2.3	18
24	New molecular approaches to Alzheimer's disease. Clinical Biochemistry, 2019, 72, 81-86.	1.9	18
25	Evaluation of three advanced methodologies, COLD-PCR, microarray and ddPCR, for identifying the mutational status by liquid biopsies in metastatic colorectal cancer patients. Clinica Chimica Acta, 2019, 489, 136-143.	1.1	18
26	Comparable clinical characteristics in Brugada syndrome patients harboring SCN5A or novel SCN10A variants. Europace, 2019, 21, 1550-1558.	1.7	15
27	SCN5A Nonsense Mutation and NF1 Frameshift Mutation in a Family With Brugada Syndrome and Neurofibromatosis. Frontiers in Genetics, 2019, 10, 50.	2.3	12
28	Genotype/Phenotype Relationship in a Consanguineal Family With Brugada Syndrome Harboring the R1632C Missense Variant in the SCN5A Gene. Frontiers in Physiology, 2019, 10, 666.	2.8	11
29	A novel homozygous mutation in the TRDN gene causes a severe form of pediatric malignant ventricular arrhythmia. Heart Rhythm, 2020, 17, 296-304.	0.7	11
30	Immunosuppressive therapy in childhoodâ€onset arrhythmogenic inflammatory cardiomyopathy. PACE - Pacing and Clinical Electrophysiology, 2021, 44, 552-556.	1.2	11
31	Quantitative serological evaluation as a valuableÂtool in the COVID-19 vaccination campaign. Clinical Chemistry and Laboratory Medicine, 2021, 59, 2019-2026.	2.3	11
32	Novel SCN5A Frameshift Mutation in Brugada Syndrome Associated With Complex Arrhythmic Phenotype. Frontiers in Genetics, 2019, 10, 547.	2.3	10
33	Is laboratory medicine ready for the era of personalized medicine? A survey addressed to laboratory directors of hospitals/academic schools of medicine in Europe. Drug Metabolism and Personalized Therapy, 2015, 30, 121-128.	0.6	9
34	Evaluation of damaging effects of splicing mutations: Validation of an in vitro method for diagnostic laboratories. Clinica Chimica Acta, 2014, 436, 276-282.	1.1	7
35	Novel SCN5A p.W697X Nonsense Mutation Segregation in a Family with Brugada Syndrome. International Journal of Molecular Sciences, 2019, 20, 4920.	4.1	7
36	Integration of multigene panels for the diagnosis of hereditary retinal disorders using Next Generation Sequencing and bioinformatics approaches. Electronic Journal of the International Federation of Clinical Chemistry and Laboratory Medicine, 2018, 29, 15-25.	0.7	7

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37	Current scenario of the genetic testing for rare neurological disorders exploiting next generation sequencing. Neural Regeneration Research, 2021, 16, 475.	3.0	6
38	Exploratory assessment of serological tests to determine antibody titer against SARSâ€CoVâ€2: Appropriateness and limits. Journal of Clinical Laboratory Analysis, 2022, 36, e24363.	2.1	6
39	Implementation of a companion diagnostic in the clinical laboratory: The BRAF example in melanoma. Clinica Chimica Acta, 2015, 439, 128-136.	1.1	5
40	Novel SCN5A p.V1429M Variant Segregation in a Family with Brugada Syndrome. International Journal of Molecular Sciences, 2020, 21, 5902.	4.1	5
41	Genetic background of mitral valve prolapse. Reviews in Cardiovascular Medicine, 2022, 23, 096.	1.4	5
42	Current Updates on Expanded Carrier Screening: New Insights in the Omics Era. Medicina (Lithuania), 2022, 58, 455.	2.0	5
43	Evaluation of antibody titer kinetics and SARS-CoV-2 infections in a large cohort of healthcare professionals ten months after administration of the BNT162b2 vaccine. Journal of Immunological Methods, 2022, 506, 113293.	1.4	4
44	Health technology assessment to employ COVID-19 serological tests as companion diagnostics in the vaccination campaign against SARS-CoV-2. Clinical Chemistry and Laboratory Medicine, 2022, .	2.3	2
45	Genetic testing in neurology exploiting next generation sequencing: state of art. Neural Regeneration Research, 2020, 15, 265.	3.0	1
46	Generation of a Triadin KnockOut Syndrome Zebrafish Model. International Journal of Molecular Sciences, 2021, 22, 9720.	4.1	0
47	Evidence of significant difference in key COVID-19 biomarkers during the Italian lockdown strategy. A retrospective study on patients admitted to a hospital emergency department in Northern Italy. Acta Biomedica, 2020, 91, e2020156.	0.3	0