Annalisa Botta

List of Publications by Year
in descending order

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80	2,397	236612	233125
papers	citations	25 h-index	45 g-index

84	84	84	2857
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Congenital heart disease in mice deficient for the DiGeorge syndrome region. Nature, 1999, 401, 379-383.	13.7	365
2	Searching for Psoriasis Susceptibility Genes in Italy: Genome Scan and Evidence for a New Locus on Chromosome 1. Journal of Investigative Dermatology, 1999, 112, 32-35.	0.3	161
3	Title is missing!. Nature, 1999, 401, 379-383.	13.7	147
4	Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. American Journal of Human Genetics, 2016, 98, 1130-1145.	2.6	118
5	Overexpression of microRNA-206 in the skeletal muscle from myotonic dystrophy type 1 patients. Journal of Translational Medicine, 2010, 8, 48.	1.8	97
6	Evidence for differential S100 gene over-expression in psoriatic patients from genetically heterogeneous pedigrees. Human Genetics, 2002, 111, 310-313.	1.8	78
7	Normal myogenesis and increased apoptosis in myotonic dystrophy type-1 muscle cells. Cell Death and Differentiation, 2010, 17, 1315-1324.	5.0	74
8	Myotonic dystrophy type 1: role of <scp>CCG</scp>, <scp>CTC</scp> and <scp>CGG</scp> interruptions within <scp>DMPK</scp> alleles in the pathogenesis and molecular diagnosis. Clinical Genetics, 2017, 92, 355-364.	1.0	52
9	The CTG repeat expansion size correlates with the splicing defects observed in muscles from myotonic dystrophy type 1 patients. Journal of Medical Genetics, 2008, 45, 639-646.	1.5	51
10	Co-segregation of DM2 with a recessive CLCN1 mutation in juvenile onset of myotonic dystrophy type 2. Journal of Neurology, 2012, 259, 2090-2099.	1.8	47
11	Risk Prediction for Clinical Phenotype in Myotonic Dystrophy Type 1: Data from 2,650 Patients. Genetic Testing and Molecular Biomarkers, 2007, 11, 84-90.	1.7	46
12	Comparative mapping of the DiGeorge syndrome region in mouse shows inconsistent gene order and differential degree of gene conservation. Mammalian Genome, 1997, 8, 890-895.	1.0	44
13	Effect of the [CCTG]n repeat expansion on ZNF9 expression in myotonic dystrophy type II (DM2). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 329-334.	1.8	44
14	Gene Expression Analysis in Myotonic Dystrophy: Indications for a Common Molecular Pathogenic Pathway in DM1 and DM2. Gene Expression, 2006, 13, 339-351.	0.5	39
15	SCN4A mutation as modifying factor of Myotonic Dystrophy Type 2 phenotype. Neuromuscular Disorders, 2015, 25, 301-307.	0.3	39
16	Identification and characterization of 5â€² CCG interruptions in complex DMPK expanded alleles. European Journal of Human Genetics, 2017, 25, 257-261.	1.4	38
17	Association study of a promoter polymorphism of UFD1L gene with schizophrenia. American Journal of Medical Genetics Part A, 2001, 105, 529-533.	2.4	37
18	An Age-Standardized Prevalence Estimate and a Sex and Age Distribution of Myotonic Dystrophy Types 1 and 2 in the Rome Province, Italy. Neuroepidemiology, 2016, 46, 191-197.	1.1	37

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19	Epstein-Barr virus infection induces miR-21 in terminally differentiated malignant B cells. International Journal of Cancer, 2015, 137, 1491-1497.	2.3	34
20	Vitamin D Receptor in Muscle Atrophy of Elderly Patients: A Key Element of Osteoporosis-Sarcopenia Connection. , 2018, 9, 952.		34
21	Altered Ca2+ Homeostasis and Endoplasmic Reticulum Stress in Myotonic Dystrophy Type 1 Muscle Cells. Genes, 2013, 4, 275-292.	1.0	33
22	Reliable and versatile immortal muscle cell models from healthy and myotonic dystrophy type 1 primary human myoblasts. Experimental Cell Research, 2016, 342, 39-51.	1.2	32
23	Expansion size and presence of CCG/CTC/CGG sequence interruptions in the expanded CTG array are independently associated to hypermethylation at the DMPK locus in myotonic dystrophy type 1 (DM1). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 2645-2652.	1.8	31
24	Prevalence and predictor factors of respiratory impairment in a large cohort of patients with Myotonic Dystrophy type 1 (DM1): A retrospective, cross sectional study. Journal of the Neurological Sciences, 2019, 399, 118-124.	0.3	31
25	Overexpression of CUGBP1 in Skeletal Muscle from Adult Classic Myotonic Dystrophy Type 1 but Not from Myotonic Dystrophy Type 2. PLoS ONE, 2013, 8, e83777.	1.1	29
26	Premature senescence in primary muscle cultures of myotonic dystrophy type 2 is not associated with p16 induction. European Journal of Histochemistry, 2014, 58, 2444.	0.6	27
27	MBNL142 and MBNL143 gene isoforms, overexpressed in DM1-patient muscle, encode for nuclear proteins interacting with Src family kinases. Cell Death and Disease, 2013, 4, e770-e770.	2.7	26
28	Ribonuclear inclusions and MBNL1 nuclear sequestration do not affect myoblast differentiation but alter gene splicing in myotonic dystrophy type 2. Neuromuscular Disorders, 2009, 19, 335-343.	0.3	25
29	Expression Analysis and Protein Localization of the Human HPC-1/Syntaxin 1A, a Gene Deleted in Williams Syndrome. Genomics, 1999, 62, 525-528.	1.3	24
30	Expression analysis of the gene encoding for the U-box-type ubiquitin ligase UBE4A in human tissues. Gene, 2004, 328, 69-74.	1.0	22
31	Validation of Sensitivity and Specificity of Tetraplet-Primed PCR (TP-PCR) in the Molecular Diagnosis of Myotonic Dystrophy Type 2 (DM2). Journal of Molecular Diagnostics, 2010, 12, 601-606.	1.2	22
32	Modelling the pathogenesis of Myotonic Dystrophy type 1 cardiac phenotype through human iPSC-derived cardiomyocytes. Journal of Molecular and Cellular Cardiology, 2018, 118, 95-109.	0.9	21
33	Two Different Therapeutic Approaches for SARS-CoV-2 in hiPSCs-Derived Lung Organoids. Cells, 2022, 11, 1235.	1.8	21
34	Use of RNA Fluorescence In Situ Hybridization in the Prenatal Molecular Diagnosis of Myotonic Dystrophy Type I. Clinical Chemistry, 2006, 52, 319-322.	1.5	20
35	Aberrant splicing and expression of the non muscle myosin heavy-chain gene MYH14 in DM1 muscle tissues. Neurobiology of Disease, 2012, 45, 264-271.	2.1	20
36	Recognition of emotions conveyed by facial expression and body postures in myotonic dystrophy (DM). Cortex, 2020, 127, 58-66.	1.1	19

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37	Progression of muscle histopathology but not of spliceopathy in myotonic dystrophy type 2. Neuromuscular Disorders, 2014, 24, 1042-1053.	0.3	18
38	Generation of Human Induced Pluripotent Stem Cells from Extraembryonic Tissues of Fetuses Affected by Monogenic Diseases. Cellular Reprogramming, 2015, 17, 275-287.	0.5	18
39	Cutaneous features of myotonic dystrophy types 1 and 2: Implication of premature aging and vitamin D homeostasis. Neuromuscular Disorders, 2017, 27, 163-169.	0.3	18
40	A long PCR-based molecular protocol for detecting normal and expanded ZNF9 alleles in myotonic dystrophy type 2. Diagnostic Molecular Pathology, 2004, 13, 164-6.	2.1	18
41	DNA Methylation Signatures of Bone Metabolism in Osteoporosis and Osteoarthritis Aging-Related Diseases: An Updated Review. International Journal of Molecular Sciences, 2021, 22, 4244.	1.8	16
42	The myotonic dystrophy type 2 (DM2) gene product zinc finger protein 9 (ZNF9) is associated with sarcomeres and normally localized in DM2 patients' muscles. Neuropathology and Applied Neurobiology, 2010, 36, 275-284.	1.8	15
43	SCN4A as modifier gene in patients with myotonic dystrophy type 2. Scientific Reports, 2018, 8, 11058.	1.6	15
44	Identification of Aberrantly-Expressed Long Non-Coding RNAs in Osteoblastic Cells from Osteoporotic Patients. Biomedicines, 2020, 8, 65.	1.4	15
45	Transmission ratio distortion in the spinal muscular atrophy locus: Data from 314 prenatal tests. Neurology, 2005, 65, 1631-1635.	1.5	14
46	Activation of the interferon type I response rather than autophagy contributes to myogenesis inhibition in congenital DM1 myoblasts. Cell Death and Disease, 2018, 9, 1071.	2.7	14
47	Exclusion of the elastin gene in the pathogenesis of Costello syndrome. American Journal of Medical Genetics Part A, 2001, 98, 286-287.	2.4	13
48	A 34-year longitudinal study on long-term cardiac outcomes in DM1 patients with normal ECG at baseline at an Italian clinical centre. Journal of Neurology, 2018, 265, 885-895.	1.8	13
49	Early subclinical cochlear dysfunction in myotonic dystrophy type 1. European Journal of Neurology, 2011, 18, 1412-1416.	1.7	12
50	Expanded [CCTG]n repetitions are not associated with abnormal methylation at the CNBP locus in myotonic dystrophy type 2 (DM2) patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 917-924.	1.8	12
51	Circulating Long Non-Coding RNA GAS5 Is Overexpressed in Serum from Osteoporotic Patients and Is Associated with Increased Risk of Bone Fragility. International Journal of Molecular Sciences, 2020, 21, 6930.	1.8	12
52	Evaluation of mexiletine effect on conduction delay and bradyarrhythmic complications in patients with myotonic dystrophy type 1 over long-term follow-up. Heart Rhythm, 2020, 17, 1944-1950.	0.3	12
53	Characterization of FMR1 Repeat Expansion and Intragenic Variants by Indirect Sequence Capture. Frontiers in Genetics, 2021, 12, 743230.	1.1	12
54	Simultaneous detection of Î”F508, G542X, N1303K, G551D, and 1717-1Gâ†'A cystic fibrosis alleles by a multiplex DNA enzyme immunoassay. International Journal of Clinical and Laboratory Research, 1995, 25, 142-145.	1.0	11

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55	Cloning and characterization of the gene encoding human NPL4, a protein interacting with the ubiquitin fusion-degradation protein (UFD1L). Gene, 2001, 275, 39-46.	1.0	11
56	Prenatal Diagnosis of Cockayne Syndrome Type A Based on the Identification of Two Novel Mutations in the ERCC8 Gene. Genetic Testing and Molecular Biomarkers, 2009, 13, 127-131.	0.3	11
57	A pilot study of lncRNAs expression profile in serum of progressive multiple sclerosis patients. European Review for Medical and Pharmacological Sciences, 2020, 24, 3267-3273.	0.5	11
58	Targeted Next Generation Sequencing in patients with Myotonia Congenita. Clinica Chimica Acta, 2017, 470, 1-7.	0.5	10
59	The Role of PTX3 in Mineralization Processes and Aging-Related Bone Diseases. Frontiers in Immunology, 2020, 11, 622772.	2.2	10
60	The long pentraxin PTX3: a novel serum marker to improve the prediction of osteoporosis and osteoarthritis bone-related phenotypes. Journal of Orthopaedic Surgery and Research, 2021, 16, 288.	0.9	10
61	Circulating MicroRNAs as Biomarkers of Osteoporosis and Fragility Fractures. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 2267-2285.	1.8	10
62	Causes of the phenotypeâ€“genotype dissociation in DiGeorge syndrome: clues from mouse models. Trends in Genetics, 2001, 17, 551-554.	2.9	8
63	TNNT2 Missplicing in Skeletal Muscle as a Cardiac Biomarker in Myotonic Dystrophy Type 1 but Not in Myotonic Dystrophy Type 2. Frontiers in Neurology, 2019, 10, 992.	1.1	8
64	Epigenetics of Myotonic Dystrophies: A Minireview. International Journal of Molecular Sciences, 2021, 22, 12594.	1.8	8
65	Carrier frequency of CFTR variants in the nonâ€Caucasian populations by genome aggregation database (gnomAD)â€based analysis. Annals of Human Genetics, 2020, 84, 463-468.	0.3	7
66	Workload measurement for molecular genetics laboratory: A survey study. PLoS ONE, 2018, 13, e0206855.	1.1	6
67	AFM nanoâ€mechanical study of the beating profile of hiPSCâ€derived cardiomyocytes beating bodies WT and DM1. Journal of Molecular Recognition, 2018, 31, e2725.	1.1	6
68	Cloning and molecular characterization of three Ubiquitin Fusion Degradation 1 (Ufd1) ortholog genes from Xenopus laevis, Gallus gallus andDrosophila melanogaster. Cytogenetic and Genome Research, 2001, 92, 279-282.	0.6	5
69	Functional characterization of the 5? flanking region of human ubiquitin fusion degradation 1 like gene (UFD1L). Cell Biochemistry and Function, 2002, 20, 163-170.	1.4	5
70	Validation of the Nine Hole Peg Test as a measure of dexterity in myotonic dystrophy type 1. Neuromuscular Disorders, 2018, 28, 947-951.	0.3	5
71	Genetic variability in noncoding RNAs: involvement of miRNAs and long noncoding RNAs in osteoporosis pathogenesis. Epigenomics, 2020, 12, 2035-2049.	1.0	4
72	Characterization of a single nucleotide polymorphism in the ZNF9 gene and analysis of association with myotonic dystrophy type II (DM2) in the Italian population. Molecular and Cellular Probes, 2005, 19, 71-74.	0.9	3

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73	Generation and Neuronal Differentiation of hiPSCs From Patients With Myotonic Dystrophy Type 2. Frontiers in Physiology, 2018, 9, 967.	1.3	3
74	The Role of Motor System in Mental Rotation: New Insights from Myotonic Dystrophy Type 1. Journal of the International Neuropsychological Society, 2020, 26, 492-502.	1.2	3
75	A 14-Year Italian Experience in DM2 Genetic Testing: Frequency and Distribution of Normal and Premutated CNBP Alleles. Frontiers in Genetics, 2021, 12, 668094.	1.1	3
76	Identification, molecular characterization and segregation analysis of a variant pre-mutation allele in a three-generation Italian family. Acta Myologica, 2020, 39, 13-18.	1.5	3
77	Deregulated Clusterin as a Marker of Bone Fragility: New Insights into the Pathophysiology of Osteoporosis. Genes, 2022, 13, 652.	1.0	3
78	Therapeutic Strategies for the Treatment of Spinal Muscular Atrophy (SMA) Disease. Current Genomics, 2006, 7, 381-386.	0.7	1
79	Molecular investigation of riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency (RR-MAD) patients. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 54.	0.5	0
80	Reply to the letter entitled â€œPredictors of respiratory impairment in patients with myotonic dystrophy type 1â€• Journal of the Neurological Sciences, 2019, 403, 166-167.	0.3	0