Maria Vittoria Cubellis

List of Publications by Year
in descending order

75 papers	3,224 citations	147726 31 h-index	155592 55 g-index
76 all docs	76 docs citations	76 times ranked	3220 citing authors

#	Article	IF	Citations
1	Autocrine saturation of pro-urokinase receptors on human A431 cells. Cell, 1986, 45, 675-684.	13.5	364
2	Hypomethylation at multiple maternally methylated imprinted regions including PLAGL1 and GNAS loci in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2009, 17, 611-619.	1.4	194
3	Accessibility of receptor-bound urokinase to type-1 plasminogen activator inhibitor.. Proceedings of the National Academy of Sciences of the United States of America, 1989, 86, 4828-4832.	3.3	167
4	Regulation of urokinase receptors in monocytelike U937 cells by phorbol ester phorbol myristate acetate.. Journal of Cell Biology, 1989, 108, 693-702.	2.3	143
5	The KCNQ1OT1 imprinting control region and non-coding RNA: new properties derived from the study of Beckwithâ€“Wiedemann syndrome and Silverâ€“Russell syndrome cases. Human Molecular Genetics, 2012, 21, 10-25.	1.4	135
6	Isolation and sequencing of a new Î²-galactosidase-encoding archaebacterial gene. Gene, 1990, 94, 89-94.	1.0	113
7	(Epi)genotypeâ€“phenotype correlations in Beckwithâ€“Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 183-190.	1.4	113
8	Properties of polyproline II, a secondary structure element implicated in protein-protein interactions. Proteins: Structure, Function and Bioinformatics, 2005, 58, 880-892.	1.5	94
9	CDKN1C mutations: two sides of the same coin. Trends in Molecular Medicine, 2014, 20, 614-622.	3.5	89
10	The pharmacological chaperone 1-deoxynojirimycin increases the activity and lysosomal trafficking of multiple mutant forms of acid alpha-glucosidase. Human Mutation, 2009, 30, 1683-1692.	1.1	85
11	Cloning and sequencing of the gene coding for aspartate aminotransferase from the thermoacidophilic archaebacterium Sulfolobus solfataricus. FEBS Journal, 1989, 186, 375-381.	0.2	80
12	The receptor for urokinase-plasminogen activator. Journal of Cellular Biochemistry, 1986, 32, 179-186.	1.2	79
13	The molecular function and clinical phenotype of partial deletions of the IGF2/H19 imprinting control region depends on the spatial arrangement of the remaining CTCF-binding sites. Human Molecular Genetics, 2013, 22, 544-557.	1.4	78
14	Different mechanisms cause imprinting defects at the IGF2/H19 locus in Beckwith-Wiedemann syndrome and Wilms' tumour. Human Molecular Genetics, 2008, 17, 1427-1435.	1.4	76
15	Pharmacological Chaperones: A Therapeutic Approach for Diseases Caused by Destabilizing Missense Mutations. International Journal of Molecular Sciences, 2020, 21, 489.	1.8	70
16	The urokinase receptor and regulation of cell surface plasminogen activation. Cell Differentiation and Development, 1990, 32, 247-253.	0.4	57
17	Secondary structure assignment that accurately reflects physical and evolutionary characteristics. BMC Bioinformatics, 2005, 6, S8.	1.2	53
18	Looking for protein stabilizing drugs with thermal shift assay. Drug Testing and Analysis, 2015, 7, 831-834.	1.6	53

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19	Antitumor Action of Seminal Ribonuclease, Its Dimeric Structure, and Its Resistance to the Cytosolic Ribonuclease Inhibitorâ€. Biochemistry, 2001, 40, 3492-3496.	1.2	49
20	Inherited and Sporadic Epimutations at the IGF2-H19 Locus in Beckwith-Wiedemann Syndrome and Wilms’ Tumor. Endocrine Development, 2009, 14, 1-9.	1.3	48
21	Loss-of-function maternal-effect mutations of PADI6 are associated with familial and sporadic Beckwith-Wiedemann syndrome with multi-locus imprinting disturbance. Clinical Epigenetics, 2020, 12, 139.	1.8	40
22	Protective Role of a TMPRSS2 Variant on Severe COVID-19 Outcome in Young Males and Elderly Women. Genes, 2021, 12, 596.	1.0	39
23	Characterization of aromatic aminotransferases from the hyperthermophilic archaeon Thermococcus litoralis. FEBS Journal, 1994, 220, 543-549.	0.2	38
24	An extremely thermostable aromatic aminotransferase from the hyperthermophilic archaeon Pyrococcus furiosus. BBA - Proteins and Proteomics, 1995, 1247, 90-96.	2.1	36
25	Ribonucleases and Angiogenins from Fish. Journal of Biological Chemistry, 2006, 281, 27454-27460.	1.6	36
26	Prediction of the responsiveness to pharmacological chaperones: lysosomal human alpha-galactosidase, a case of study. Orphanet Journal of Rare Diseases, 2010, 5, 36.	1.2	35
27	Conformational Response to Ligand Binding in Phosphomannomutase2. Journal of Biological Chemistry, 2014, 289, 34900-34910.	1.6	34
28	Fetal growth patterns in Beckwithâ€“Wiedemann syndrome. Clinical Genetics, 2016, 90, 21-27.	1.0	34
29	A splicing mutation of the HMGA2 gene is associated with Silverâ€“Russell syndrome phenotype. Journal of Human Genetics, 2015, 60, 287-293.	1.1	33
30	Therapy of Fabry disease with pharmacological chaperones: from in silico predictions to in vitro tests. Orphanet Journal of Rare Diseases, 2011, 6, 66.	1.2	32
31	The Analysis of Variants in the General Population Reveals That PMM2 Is Extremely Tolerant to Missense Mutations and That Diagnosis of PMM2-CDG Can Benefit from the Identification of Modifiers. International Journal of Molecular Sciences, 2018, 19, 2218.	1.8	32
32	Relaxation of Insulin-like Growth Factor 2 Imprinting and Discordant Methylation at KvDMR1 in Two First Cousins Affected by Beckwith-Wiedemann and Klippel-Trenaunay-Weber Syndromes. American Journal of Human Genetics, 2000, 66, 841-847.	2.6	31
33	Silver-Russell Syndrome and Beckwith-Wiedemann Syndrome Phenotypes Associated with 11p Duplication in a Single Family. Pediatric and Developmental Pathology, 2010, 13, 326-330.	0.5	31
34	Drug repositioning can accelerate discovery of pharmacological chaperones. Orphanet Journal of Rare Diseases, 2015, 10, 55.	1.2	29
35	Î²-Glucose-1,6-Bisphosphate Stabilizes Pathological Phophomannomutase2 Mutants In Vitro and Represents a Lead Compound to Develop Pharmacological Chaperones for the Most Common Disorder of Glycosylation, PMM2-CDG. International Journal of Molecular Sciences, 2019, 20, 4164.	1.8	29
36	Tryptophan biosynthesis genes trpEGC in the thermoacidophilic archaebacterium Sulfolobus solfataricus. Journal of Bacteriology, 1993, 175, 299-302.	1.0	28

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37	Identification of an Allosteric Binding Site on Human Lysosomal Alpha-Galactosidase Opens the Way to New Pharmacological Chaperones for Fabry Disease. PLoS ONE, 2016, 11, e0165463.	1.1	27
38	Limited proteolysis as a probe of conformational changes in aspartate aminotransferase from Sulfolobus solfataricus. FEBS Journal, 1992, 204, 1183-1189.	0.2	26
39	Fabry_CEP: a tool to identify Fabry mutations responsive to pharmacological chaperones. Orphanet Journal of Rare Diseases, 2013, 8, 111.	1.2	26
40	Heterodimerization of Two Pathological Mutants Enhances the Activity of Human Phosphomannomutase2. PLoS ONE, 2015, 10, e0139882.	1.1	26
41	Identification of a Novel Mutation in the Myosin VIIA Motor Domain in a Family with Autosomal Dominant Hearing Loss (DFNA11). Audiology and Neuro-Otology, 2006, 11, 157-164.	0.6	25
42	The Large Phenotypic Spectrum of Fabry Disease Requires Graduated Diagnosis and Personalized Therapy: A Meta-Analysis Can Help to Differentiate Missense Mutations. International Journal of Molecular Sciences, 2016, 17, 2010.	1.8	25
43	The polymorphism L412F in TLR3 inhibits autophagy and is a marker of severe COVID-19 in males. Autophagy, 2022, 18, 1662-1672.	4.3	25
44	Use of fast protein liquid chromatography for the purification of synthetic oligonucleotides. Journal of Chromatography A, 1985, 329, 406-414.	1.8	22
45	The phenotypic variations of multi-locus imprinting disturbances associated with maternal-effect variants of NLRP5 range from overt imprinting disorder to apparently healthy phenotype. Clinical Epigenetics, 2019, 11, 190.	1.8	22
46	Viable phenotype of ILNEB syndrome without nephrotic impairment in siblings heterozygous for unreported integrin alpha3 mutations. Orphanet Journal of Rare Diseases, 2016, 11, 136.	1.2	21
47	Biochemical phenotype of a common diseaseâ€causing mutation and a possible therapeutic approach for the phosphomannomutase 2â€associated disorder of glycosylation. Molecular Genetics & Genomic Medicine, 2013, 1, 32-44.	0.6	20
48	Identification and analysis of conserved pockets on protein surfaces. BMC Bioinformatics, 2013, 14, S9.	1.2	19
49	An intron-less Î²Î³-crystallin-type gene from the sponge Geodia cydonium. Gene, 2002, 299, 79-82.	1.0	18
50	Preparation and characterization of geodin.. FEBS Journal, 2005, 272, 1023-1035.	2.2	17
51	Why does SARS-CoV-2 hit in different ways? Host genetic factors can influence the acquisition or the course of COVID-19. European Journal of Medical Genetics, 2021, 64, 104227.	0.7	17
52	A thermodynamic assay to test pharmacological chaperones for Fabry disease. Biochimica Et Biophysica Acta - General Subjects, 2014, 1840, 1214-1224.	1.1	16
53	Assessment of Gene Variant Amenability for Pharmacological Chaperone Therapy with 1-Deoxygalactonojirimycin in Fabry Disease. International Journal of Molecular Sciences, 2020, 21, 956.	1.8	15
54	Proteostasis regulators modulate proteasomal activity and gene expression to attenuate multiple phenotypes in Fabry disease. Biochemical Journal, 2020, 477, 359-380.	1.7	15

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55	Taming molecular flexibility to tackle rare diseases. Biochimie, 2015, 113, 54-58.	1.3	13
56	D2A sequence of the urokinase receptor induces cell growth through Î±vÎ²3 integrin and EGFR. Cellular and Molecular Life Sciences, 2018, 75, 1889-1907.	2.4	13
57	Drug Repositioning for Fabry Disease: Acetylsalicylic Acid Potentiates the Stabilization of Lysosomal Alpha-Galactosidase by Pharmacological Chaperones. International Journal of Molecular Sciences, 2022, 23, 5105.	1.8	13
58	The active site of Sulfolobus solfataricus aspartate aminotransferase. BBA - Proteins and Proteomics, 1991, 1080, 198-204.	2.1	12
59	Molecular characterization of G6PD deficiency in Southern Italy: heterogeneity, correlation genotypeâ€“phenotype and description of a new variant (G6PD Neapolis). British Journal of Haematology, 1997, 98, 41-46.	1.2	12
60	Gain of function in CDKN1C. Nature Genetics, 2012, 44, 737-738.	9.4	12
61	A mutant of phosphomannomutase1 retains full enzymatic activity, but is not activated by IMP: Possible implications for the disease PMM2-CDG. PLoS ONE, 2017, 12, e0189629.	1.1	12
62	Functional characterisation of a novel mutation affecting the catalytic domain of MMP2 in siblings with multicentric osteolysis, nodulosis and arthropathy. Journal of Human Genetics, 2014, 59, 631-637.	1.1	11
63	Mechanistic Insight into the Mode of Action of Acid Î²-Glucosidase Enhancer Ambroxol. International Journal of Molecular Sciences, 2022, 23, 3536.	1.8	11
64	In silico docking of urokinase plasminogen activator and integrins. BMC Bioinformatics, 2008, 9, S8.	1.2	10
65	Biallelic variant in cyclin B3 is associated with failure of maternal meiosis II and recurrent digynic triploidy. Journal of Medical Genetics, 2021, 58, 783-788.	1.5	9
66	Challenging popular tools for the annotation of genetic variations with a real case, pathogenic mutations of lysosomal alpha-galactosidase. BMC Bioinformatics, 2018, 19, 433.	1.2	8
67	Novel genetic variants of KHDC3L and other members of the subcortical maternal complex associated with Beckwithâ€“Wiedemann syndrome or Pseudohypoparathyroidism 1B and multi-locus imprinting disturbances. Clinical Epigenetics, 2022, 14, .	1.8	7
68	Nucleotide sequence of a cDNA coding for bovine mitochondrial aspartate aminotransferase. International Journal of Biochemistry and Cell Biology, 1995, 27, 507-511.	1.2	3
69	An open reading frame in intron seven of the sea urchin DNA-methyltransferase gene codes for a functional AP1 endonuclease. Biochemical Journal, 2002, 365, 833-840.	1.7	3
70	D2A-Ala peptide derived from the urokinase receptor exerts anti-tumoural effects in vitro and in vivo. Peptides, 2018, 101, 17-24.	1.2	3
71	Cloning and sequence analysis of a cdna encoding bovine cytosolic aspartate aminotransferase. International Journal of Biochemistry & Cell Biology, 1993, 25, 1505-1509.	0.8	2
72	The interconversion of isoforms of seminal ribonuclease: modelling key intermediates and trypsin effects 1 1Edited by J. Thorton. Journal of Molecular Biology, 2000, 301, 775-782.	2.0	2

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73	Identification of Trombospondin-1 as a Novel Amelogenin Interactor by Functional Proteomics. Frontiers in Chemistry, 2017, 5, 74.	1.8	2
74	Bioinformatics tools for marine biotechnology: a practical tutorial with a metagenomic approach. BMC Bioinformatics, 2020, 21, 348.	1.2	1
75	Data on the inhibition of cell proliferation and invasion by the D2A-Ala peptide derived from the urokinase receptor. Data in Brief, 2019, 22, 903-908.	0.5	0