Teresa Esposito

List of Publications by Year in descending order

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86 papers	2,919 citations	230014 27 h-index	198040 52 g-index
86 all docs	86 docs citations	86 times ranked	4081 citing authors

#	Article	IF	Citations
1	Role of Uncoupling Protein 2 Gene Polymorphisms on the Risk of Ischemic Stroke in a Sardinian Population. Life, 2022, 12, 721.	1.1	1
2	Novel autophagic vacuolar myopathies: Phenotype and genotype features. Neuropathology and Applied Neurobiology, 2021, 47, 664-678.	1.8	4
3	Rare Variants in Autophagy and Non-Autophagy Genes in Late-Onset Pompe Disease: Suggestions of Their Disease-Modifying Role in Two Italian Families. International Journal of Molecular Sciences, 2021, 22, 3625.	1.8	2
4	Analysis of Genetic and Non-genetic Predictors of Levodopa Induced Dyskinesia in Parkinsonâ€™s Disease. Frontiers in Pharmacology, 2021, 12, 640603.	1.6	8
5	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	4.4	41
6	Intrafamilial â€œDOAâ€•plusâ€•phenotype variability related to different OMI/HTRA2 expression. American Journal of Medical Genetics, Part A, 2020, 182, 176-182.	0.7	2
7	ZNF687 Mutations in an Extended Cohort of Neoplastic Transformations in Paget's Disease of Bone: Implications for Clinical Pathology. Journal of Bone and Mineral Research, 2020, 35, 1974-1980.	3.1	9
8	The Loss of Profilin 1 Causes Early Onset Paget's Disease of Bone. Journal of Bone and Mineral Research, 2020, 35, 1387-1398.	3.1	27
9	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. Ophthalmic Genetics, 2019, 40, 39-42.	0.5	10
10	Whole Exome Sequencing Study of Parkinson Disease and Related Endophenotypes in the Italian Population. Frontiers in Neurology, 2019, 10, 1362.	1.1	9
11	Successful long-term therapy with flecainide in a family with paramyotonia congenita. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1232-1234.	0.9	7
12	ZNF687 mutations are frequently found in pagetic patients from South Italy: implication in the pathogenesis of Paget's disease of bone. Clinical Genetics, 2018, 93, 1240-1244.	1.0	14
13	Autosomalâ€dominant myopia associated to a novel P4HA2 missense variant and defective collagen hydroxylation. Clinical Genetics, 2018, 93, 982-991.	1.0	21
14	Vacuolated PAS-positive lymphocytes as an hallmark of Pompe disease and other myopathies related to impaired autophagy. Journal of Cellular Physiology, 2018, 233, 5829-5837.	2.0	15
15	The identification of H3F3A mutation in giant cell tumour of the clivus and the histological diagnostic algorithm of other clival lesions permit the differential diagnosis in this location. BMC Cancer, 2018, 18, 358.	1.1	11
16	Dysregulation of the Expression of Asparagine-Linked Glycosylation 13 Short Isoform 2 Affects Nephrin Function by Altering Its N-Linked Glycosylation. Nephron, 2017, 136, 143-150.	0.9	5
17	First study on the peptidergic innervation of the brain superior sagittal sinus in humans. Neuropeptides, 2017, 65, 45-55.	0.9	12
18	Identification of the first dominant mutation of LAMA5 gene causing a complex multisystem syndrome due to dysfunction of the extracellular matrix. Journal of Medical Genetics, 2017, 54, 710-720.	1.5	35

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19	Sensitivity and specificity of the PAS positive lymphocyte vacuoles in the diagnostic approach to late onset Pompe disease. Neuromuscular Disorders, 2017, 27, S160-S161.	0.3	0
20	Evidence for epistatic interaction between VDR and SLC13A2 genes in the pathogenesis of hypocitraturia in recurrent calcium oxalate stone formers. Journal of Nephrology, 2017, 30, 411-418.	0.9	10
21	The distinct clinical features of giant cell tumor of bone in pagetic and non-pagetic patients are associated with genetic, biochemical and histological differences. Oncotarget, 2017, 8, 63121-63131.	0.8	15
22	ZNF687 Mutations in Severe Paget Disease of Bone Associated with Giant Cell Tumor. American Journal of Human Genetics, 2016, 98, 275-286.	2.6	61
23	A Unique Myopathy Syndrome in a Patient Disclosing Clinical, Laboratory, and Genetic Findings of Late-Onset Pompe Disease, Together with a Lack of Dysferlin on Muscle Biopsy. Journal of Neuromuscular Diseases, 2015, 2, S29-S30.	1.1	0
24	A novel GBE1 mutation and features of polyglucosan bodies autophagy in Adult Polyglucosan Body Disease. Neuromuscular Disorders, 2015, 25, 247-252.	0.3	11
25	Clinical Characteristics and Evolution of Giant Cell Tumor Occurring in Paget's Disease of Bone. Journal of Bone and Mineral Research, 2015, 30, 257-263.	3.1	38
26	A Unique Myopathy Syndrome in a Patient Disclosing Clinical, Laboratory, and Genetic Findings of Late-Onset Pompe Disease, Together with a Lack of Dysferlin on Muscle Biopsy. Journal of Neuromuscular Diseases, 2015, 2, S29-S30.	1.1	0
27	Pagetâ€™s disease of bone: epidemiology, pathogenesis and pharmacotherapy. Expert Opinion on Orphan Drugs, 2014, 2, 591-603.	0.5	3
28	Exclusion of TNFRSF11B as Candidate Gene for Otosclerosis in Campania Population. Indian Journal of Otolaryngology and Head and Neck Surgery, 2014, 66, 297-301.	0.3	3
29	Paget disease of bone-associated UBA domain mutations of SQSTM1 exert distinct effects on protein structure and function. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 992-1000.	1.8	28
30	Digenic mutational inheritance of the integrin alpha 7 and the myosin heavy chain 7B genes causes congenital myopathy with left ventricular non-compact cardiomyopathy. Orphanet Journal of Rare Diseases, 2013, 8, 91.	1.2	43
31	Distinct disease phenotypes linked to different combinations of GAA mutations in a large late-onset GSDII sibship. Orphanet Journal of Rare Diseases, 2013, 8, 159.	1.2	15
32	Imerslund-GrÃsbeck syndrome in a 25-month-old Italian girl caused by a homozygous mutation in AMN. Italian Journal of Pediatrics, 2013, 39, 58.	1.0	9
33	Association of a <scp>GRIA3</scp> Gene Polymorphism With Migraine in an <scp>A</scp>ustralian Caseâ€Control Cohort. Headache, 2013, 53, 1245-1249.	1.8	22
34	Unique X-linked familial FSGS with co-segregating heart block disorder is associated with a mutation in the NXF5 gene. Human Molecular Genetics, 2013, 22, 3654-3666.	1.4	25
35	Giant cell tumor occurring in familial Paget's disease of bone: Report of clinical characteristics and linkage analysis of a large pedigree. Journal of Bone and Mineral Research, 2013, 28, 341-350.	3.1	19
36	Common susceptibility alleles and SQSTM1 mutations predict disease extent and severity in a multinational study of patients with Paget's disease. Journal of Bone and Mineral Research, 2013, 28, 2338-2346.	3.1	50

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37	A Functional Allelic Variant of theFGF23Gene Is Associated with Renal Phosphate Leak in Calcium Nephrolithiasis. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E840-E844.	1.8	20
38	The melatonin receptor 1A (MTNR1A) gene is associated with recurrent and idiopathic calcium nephrolithiasis. Nephrology Dialysis Transplantation, 2012, 27, 210-218.	0.4	23
39	A nonsynonymous TNFRSF11A variation increases NFÎºB activity and the severity of Paget's disease. Journal of Bone and Mineral Research, 2012, 27, 443-452.	3.1	34
40	Confirmation that Xq27 and Xq28 are susceptibility loci for migraine in independent pedigrees and a case-control cohort. Neurogenetics, 2012, 13, 97-101.	0.7	8
41	piR_015520 Belongs to Piwi-Associated RNAs Regulates Expression of the Human Melatonin Receptor 1A Gene. PLoS ONE, 2011, 6, e22727.	1.1	72
42	SQSTM1 gene analysis and gene-environment interaction in Paget's disease of bone. Journal of Bone and Mineral Research, 2010, 25, 1375-1384.	3.1	64
43	Common variants in the regulative regions of GRIA1 and GRIA3 receptor genes are associated with migraine susceptibility. BMC Medical Genetics, 2010, 11, 103.	2.1	40
44	FSHR gene polymorphisms influence bone mineral density and bone turnover in postmenopausal women. European Journal of Endocrinology, 2010, 163, 165-172.	1.9	87
45	Epidemiological, clinical, and genetic characteristics of Pagetâ€™s disease of bone in a rural area of Calabria, Southern Italy. Journal of Endocrinological Investigation, 2010, 33, 519-525.	1.8	10
46	Bone Turnover and the Osteoprotegerinâ€“RANKL Pathway in Tumor-Induced Osteomalacia: A Longitudinal Study of Five Cases. Calcified Tissue International, 2009, 85, 293-300.	1.5	24
47	Vitamin D Receptor Gene Polymorphisms Predict Acquired Resistance to Clodronate Treatment in Patients with Pagetâ€™s Disease of Bone. Calcified Tissue International, 2008, 83, 414-424.	1.5	18
48	Different spectra of genomic deletions within the CCM genes between Italian and American CCM patient cohorts. Neurogenetics, 2008, 9, 25-31.	0.7	33
49	ATP1A2 gene mutations are not present in two sisters with basilar-type migraine associated with menses. Neurological Sciences, 2008, 29, 113-115.	0.9	0
50	Investigation of Gamma-aminobutyric acid (GABA) A receptors genes and migraine susceptibility. BMC Medical Genetics, 2008, 9, 109.	2.1	21
51	ZPLD1 gene is disrupted in a patient with balanced translocation that exhibits cerebral cavernous malformations. Neuroscience, 2008, 155, 345-349.	1.1	30
52	Genetic Variants of Y Chromosome Are Associated With a Protective Lipid Profile in Black Men. Arteriosclerosis, Thrombosis, and Vascular Biology, 2008, 28, 1569-1574.	1.1	21
53	Highly variable penetrance in subjects affected with cavernous cerebral angiomas (CCM) carrying novel CCM1 and CCM2 mutations. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 691-695.	1.1	32
54	The Molecular Genetics of Migraine: Toward the Identification of Responsible Genes. Current Genomics, 2006, 7, 1-10.	0.7	2

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55	Genes, Human Diseases and Genome Evolution in the Post-Genomic Era: Insights from Uric Acid Nephrolithiasis. Current Genomics, 2005, 6, 207-214.	0.7	0
56	Geographic distribution of Ala62Thr variant associated to Uric Acid Nephrolithiasis from Sub-Saharan to Mediterranean area. International Journal of Anthropology, 2004, 19, 277-280.	0.1	2
57	Genes, diet and uric acid nephrolithiasis. International Journal of Anthropology, 2004, 19, 281-288.	0.1	0
58	Emergence of Talanin protein associated with human uric acid nephrolithiasis in the Hominidae lineage. Gene, 2004, 339, 131-138.	1.0	11
59	Identification of a Novel Gene and a Common Variant Associated with Uric Acid Nephrolithiasis in a Sardinian Genetic Isolate. American Journal of Human Genetics, 2003, 72, 1479-1491.	2.6	65
60	Physical and Genetic Characterization Reveals a Pseudogene, an Evolutionary Junction, and Unstable Loci in Distal Xq28. Genomics, 2002, 79, 31-40.	1.3	13
61	Ï‡/Autosomal Translocations in the Ï‡q Critical Region Associated with Premature Ovarian Failure Fall within and outside Genes. Genomics, 2001, 76, 30-36.	1.3	36
62	Survival of Male Patients with Incontinentia Pigmenti Carrying a Lethal Mutation Can Be Explained by Somatic Mosaicism or Klinefelter Syndrome. American Journal of Human Genetics, 2001, 69, 1210-1217.	2.6	150
63	Characterization of the murine orthologue of a novel human subtelomeric multigene family. Cytogenetic and Genome Research, 2001, 94, 98-100.	0.6	10
64	Multiple pathogenic and benign genomic rearrangements occur at a 35 kb duplication involving the NEMO and LAGE2 genes. Human Molecular Genetics, 2001, 10, 2557-2567.	1.4	79
65	Differential Divergence of Three Human Pseudoautosomal Genes and Their Mouse Homologs: Implications for Sex Chromosome Evolution. Genome Research, 2001, 11, 2095-2100.	2.4	37
66	A recurrent deletion in the ubiquitously expressed NEMO (IKK-gamma) gene accounts for the vast majority of incontinentia pigmenti mutations. Human Molecular Genetics, 2001, 10, 2171-2179.	1.4	165
67	Human homologue of the murinebare patches/striated gene is not mutated in incontinentia pigmenti type 2. , 2000, 91, 241-244.		3
68	Filamin (FLN1),plexin (SEX), major palmitoylated proteinp55 (MPP1), and von-Hippel Lindau binding protein (VBP1) are not involved in incontinentia pigmenti type 2. American Journal of Medical Genetics Part A, 2000, 94, 79-84.	2.4	5
69	Molecular analysis of the mature T cell proliferation-1 (MTCP-1) gene in Xq28-linked incontinentia pigmenti. European Journal of Human Genetics, 2000, 8, 239-240.	1.4	0
70	Genomic rearrangement in NEMO impairs NF-ÎºB activation and is a cause of incontinentia pigmenti. Nature, 2000, 405, 466-472.	13.7	709
71	Differentially regulated and evolved genes in the fully sequenced Xq/Yq pseudoautosomal region. Human Molecular Genetics, 2000, 9, 395-401.	1.4	92
72	A novel pseudoautosomal human gene encodes a putative protein similar to Ac-like transposases. Human Molecular Genetics, 1999, 8, 61-67.	1.4	31

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73	Molecular cloning and fine mapping of API5L1, a novel human gene strongly related to an antiapoptotic gene. Cytogenetic and Genome Research, 1999, 84, 164-166.	0.6	5
74	Smith-Lemli-Opitz syndrome: evidence of T93M as a common mutation of Î”7-sterol reductase in Italy and report of three novel mutations. European Journal of Human Genetics, 1999, 7, 937-940.	1.4	37
75	A novel pseudoautosomal gene encoding a putative GTP-binding protein resides in the vicinity of the Xp/Yp telomere. Human Molecular Genetics, 1998, 7, 407-414.	1.4	37
76	Escape from X Inactivation of Two New Genes Associated with DXS6974E and DXS7020E. Genomics, 1997, 43, 183-190.	1.3	17
77	Mapping of 59 EST gene markers in 31 intervals spanning the human X chromosome. Gene, 1997, 187, 179-184.	1.0	14
78	Expressed STSs and transcription of human Xq28. Gene, 1997, 187, 185-191.	1.0	2
79	Isolation, physical mapping, and Northern analysis of the X-linked human gene encoding methyl CpG-binding protein, MECP2. Mammalian Genome, 1996, 7, 533-535.	1.0	116
80	A synaptobrevinâ€“like gene in the Xq28 pseudoautosomal region undergoes X inactivation. Nature Genetics, 1996, 13, 227-229.	9.4	78
81	Human protein kinase C iota gene (PRKCI) is closely linked to the BTK gene in Xq21.3. Genomics, 1995, 26, 629-631.	1.3	9
82	Sequence-tagged sites (STSs) from YAC insert-ends and X-specific flow-sorted chromosomes. Mammalian Genome, 1994, 5, 511-514.	1.0	2
83	YAC Contig Organization and CpG Island Analysis in Xq28. Genomics, 1994, 24, 149-158.	1.3	44
84	Type 2 Vasopressin Receptor Gene, the Gene Responsible for Nephrogenic Diabetes Insipidus, Maps to XQ28 Close to the L1CAM Gene. Biochemical and Biophysical Research Communications, 1993, 193, 864-871.	1.0	13
85	YAC-assisted cloning of transcribed sequences from the human chromosome 3p21 region. Human Molecular Genetics, 1993, 2, 791-796.	1.4	13
86	Pharmacogenomics of bisphosphonate treatment in Paget's disease of bone: retrospective and prospective analysis. Bone Abstracts, 0, , .	0.0	0