Fernando Gianfrancesco

List of Publications by Year
in descending order

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83 papers	2,652 citations	293460 24 h-index	223390 49 g-index
88 all docs	88 docs citations	88 times ranked	4288 citing authors

#	Article	IF	Citations
1	The Osteoclast Traces the Route to Bone Tumors and Metastases. Frontiers in Cell and Developmental Biology, 2022, 10, .	1.8	12
2	The loss of Profilin 1 is a driver of chromosome instability in osteosarcoma. Bone Reports, 2022, 16, 101183.	0.2	0
3	Zfp687 knock-in mouse model of Pagetâ€™s disease of bone exhibits increased bone marrow adiposity preceding bone remodelling alteration. Bone Reports, 2022, 16, 101197.	0.2	0
4	Novel autophagic vacuolar myopathies: Phenotype and genotype features. Neuropathology and Applied Neurobiology, 2021, 47, 664-678.	1.8	4
5	Modulation of Endocannabinoid Tone in Osteoblastic Differentiation of MC3T3-E1 Cells and in Mouse Bone Tissue over Time. Cells, 2021, 10, 1199.	1.8	7
6	Early Alpine occupation backdates westward human migration in Late Glacial Europe. Current Biology, 2021, 31, 2484-2493.e7.	1.8	17
7	Identification of sixteen novel candidate genes for late onset Parkinsonâ€™s disease. Molecular Neurodegeneration, 2021, 16, 35.	4.4	41
8	The two faces of giant cell tumor of bone. Cancer Letters, 2020, 489, 1-8.	3.2	20
9	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
10	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
11	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. Ophthalmic Genetics, 2019, 40, 39-42.	0.5	10
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	Effect of genetic variants of OPTN in the pathophysiology of Paget's disease of bone. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 143-151.	1.8	17
15	Identification of a novel locus on chromosome 2q13, which predisposes to clinical vertebral fractures independently of bone density. Annals of the Rheumatic Diseases, 2018, 77, 378-385.	0.5	21
16	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
17	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
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	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
20	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
21	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
22	AB0200â€...The Ineffectiveness of Relaxin Treatment to Ameliorate Dermal Fibrosis in Systemic Sclerosis Could be Due to Relaxin Receptor Alterations. Annals of the Rheumatic Diseases, 2015, 74, 957.3-958.	0.5	0
23	A novel GBE1 mutation and features of polyglucosan bodies autophagy in Adult Polyglucosan Body Disease. Neuromuscular Disorders, 2015, 25, 247-252.	0.3	11
24	Hypovitaminosis D and Organ Damage In Patients With Arterial Hypertension: A Multicenter Double Blind Randomised Controlled Trial of Cholecalciferol Supplementation (HYPODD). High Blood Pressure and Cardiovascular Prevention, 2015, 22, 135-142.	1.0	4
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	Pagetâ€™s disease of bone: epidemiology, pathogenesis and pharmacotherapy. Expert Opinion on Orphan Drugs, 2014, 2, 591-603.	0.5	3
27	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
28	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
29	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
30	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
31	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
32	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
33	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
34	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
35	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
36	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

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37	The melatonin receptor 1A (MTNR1A) gene is associated with recurrent and idiopathic calcium nephrolithiasis. Nephrology Dialysis Transplantation, 2012, 27, 210-218.	0.4	23
38	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
39	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
40	piR_015520 Belongs to Piwi-Associated RNAs Regulates Expression of the Human Melatonin Receptor 1A Gene. PLoS ONE, 2011, 6, e22727.	1.1	72
41	Genome-wide association identifies three new susceptibility loci for Paget's disease of bone. Nature Genetics, 2011, 43, 685-689.	9.4	158
42	Comparison of intravenous and intramuscular neridronate regimens for the treatment of paget disease of bone. Journal of Bone and Mineral Research, 2011, 26, 512-518.	3.1	28
43	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
44	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
45	FSHR gene polymorphisms influence bone mineral density and bone turnover in postmenopausal women. European Journal of Endocrinology, 2010, 163, 165-172.	1.9	87
46	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
47	DDX11L: a novel transcript family emerging from human subtelomeric regions. BMC Genomics, 2009, 10, 250.	1.2	13
48	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
49	De novo seven extra repeat expanded mutation in the PRNP gene in an Italian patient with early onset dementia. BMJ Case Reports, 2009, 2009, bcr0820080711-bcr0820080711.	0.2	2
50	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
51	Different spectra of genomic deletions within the CCM genes between Italian and American CCM patient cohorts. Neurogenetics, 2008, 9, 25-31.	0.7	33
52	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
53	Investigation of Gamma-aminobutyric acid (GABA) A receptors genes and migraine susceptibility. BMC Medical Genetics, 2008, 9, 109.	2.1	21
54	ZPLD1 gene is disrupted in a patient with balanced translocation that exhibits cerebral cavernous malformations. Neuroscience, 2008, 155, 345-349.	1.1	30

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55	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
56	De novo seven extra repeat expanded mutation in the PRNP gene in an Italian patient with early onset dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2007, 78, 1411-1413.	0.9	8
57	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
58	A spectrum of molecular variation in a cohort of Italian families with trimethylaminuria: Identification of three novel mutations of the FM03 gene. Molecular Genetics and Metabolism, 2006, 88, 192-195.	0.5	15
59	The Molecular Genetics of Migraine: Toward the Identification of Responsible Genes. Current Genomics, 2006, 7, 1-10.	0.7	2
60	The Molecular Genetics of Migraine: Toward the Identification of Responsible Genes. Current Genomics, 2006, 7, 1-10.	0.7	1
61	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
62	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
63	Genes, diet and uric acid nephrolithiasis. International Journal of Anthropology, 2004, 19, 281-288.	0.1	0
64	Identification of a novel candidate gene,CASC2, in a region of common allelic loss at chromosome 10q26 in human endometrial cancer. Human Mutation, 2004, 23, 318-326.	1.1	86
65	The evolutionary conservation of the human chitotriosidase gene in rodents and primates. Cytogenetic and Genome Research, 2004, 105, 54-56.	0.6	23
66	Emergence of Talanin protein associated with human uric acid nephrolithiasis in the Hominidae lineage. Gene, 2004, 339, 131-138.	1.0	11
67	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
68	Characterization of the murine orthologue of a novel human subtelomeric multigene family. Cytogenetic and Genome Research, 2001, 94, 98-100.	0.6	10
69	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
70	Genomic rearrangement in NEMO impairs NF-Î²B 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	Identification and chromosomal localisation by fluorescence in situ hybridisation of human gene of phosphoinositide-specific phospholipase C Î²1. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2000, 1484, 175-182.	1.2	31

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73	A novel pseudoautosomal human gene encodes a putative protein similar to Ac-like transposases. Human Molecular Genetics, 1999, 8, 61-67.	1.4	31
74	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
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	A synaptobrevinâ€“like gene in the Xq28 pseudoautosomal region undergoes X inactivation. Nature Genetics, 1996, 13, 227-229.	9.4	78
79	Genetic Mapping of a Gene Encoding an Atypical Protein Kinase C, Protein Kinase C Lambda, to the Proximal Region of Mouse Chromosome 3. Genomics, 1995, 29, 815-816.	1.3	4
80	Interaction between FGF23 R176W mutation and C716T nonsynonymous change (T239M, rs7955866) in FGF23 on the clinical phenotype in a family with autosomal dominant hypophosphatemic rickets. Bone Abstracts, 0, , .	0.0	1
81	A OPTN variant (rs1561570) interacts with TNFRSF11A polymorphism (rs1805034) on the clinical phenotype of sporadic Paget's disease of bone. Bone Abstracts, 0, , .	0.0	1
82	Pharmacogenomics of bisphosphonate treatment in Paget's disease of bone: retrospective and prospective analysis. Bone Abstracts, 0, , .	0.0	0
83	Conventional and Pagetic Giant Cell Tumor of bone: distinct clinical features are defined by different genetic background and histological appearance. Bone Abstracts, 0, , .	0.0	0