Fernando Gianfrancesco

List of Publications by Year in Descending Order

76 papers	2,102 citations	24 h-index	44 g-index
88 ext. papers	2,397 ext. citations	5.9 avg, IF	3.8 L-index

#	Paper	IF	Citations
76	Early Alpine occupation backdates westward human migration in Late Glacial Europe. *Current Biology*, **2021**, 31, 2484-2493.e7	6.3	5
75	Identification of sixteen novel candidate genes for late onset Parkinson's disease. *Molecular Neurodegeneration*, **2021**, 16, 35	19	3
74	Novel autophagic vacuolar myopathies: Phenotype and genotype features. *Neuropathology and Applied Neurobiology*, **2021**, 47, 664-678	5.2	2
73	The two faces of giant cell tumor of bone. *Cancer Letters*, **2020**, 489, 1-8	9.9	6
72	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	6.3	2
71	The Loss of Profilin 1 Causes Early Onset Paget's Disease of Bone. *Journal of Bone and Mineral Research*, **2020**, 35, 1387-1398	6.3	13
70	Early posterior vitreous detachment is associated with LAMA5 dominant mutation. *Ophthalmic Genetics*, **2019**, 40, 39-42	1.2	7
69	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	4	7
68	Autosomal-dominant myopia associated to a novel P4HA2 missense variant and defective collagen hydroxylation. *Clinical Genetics*, **2018**, 93, 982-991	4	12
67	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	6.9	9
66	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	4.8	6
65	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	2.4	15
64	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	3.3	4
63	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	5.8	27
62	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	4.8	8
61	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	3.3	12
60	ZNF687 Mutations in Severe Paget Disease of Bone Associated with Giant Cell Tumor. *American Journal of Human Genetics*, **2016**, 98, 275-86	11	39

59	Hypovitaminosis D and organ damage in patients with arterial hypertension: a multicenter double blind randomised controlled trial of cholecalciferol supplementation (HYPODD) : study design, clinical procedures and treatment protocol. *High Blood Pressure and Cardiovascular Prevention*, **2015**, 22, 135-42	2.9	3
58	Clinical characteristics and evolution of giant cell tumor occurring in Paget's disease of bone. *Journal of Bone and Mineral Research*, **2015**, 30, 257-63	6.3	30
57	AB0200 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	2.4	
56	A novel GBE1 mutation and features of polyglucosan bodies autophagy in adult polyglucosan body disease. *Neuromuscular Disorders*, **2015**, 25, 247-52	2.9	6
55	Exclusion of TNFRSF11B as Candidate Gene for Otosclerosis in Campania Population. *Indian Journal of Otolaryngology and Head and Neck Surgery*, **2014**, 66, 297-301	0.6	3
54	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	6.9	26
53	Paget□ disease of bone: epidemiology, pathogenesis and pharmacotherapy. *Expert Opinion on Orphan Drugs*, **2014**, 2, 591-603	1.1	2
52	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	4.2	32
51	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	4.2	12
50	Imerslund-Gr□beck syndrome in a 25-month-old Italian girl caused by a homozygous mutation in AMN. *Italian Journal of Pediatrics*, **2013**, 39, 58	3.2	4
49	Association of a GRIA3 gene polymorphism with migraine in an Australian case-control cohort. *Headache*, **2013**, 53, 1245-9	4.2	20
48	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, 3654-66	5.6	21
47	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, 341-50	6.3	15
46	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, 2338-46	6.3	43
45	A nonsynonymous TNFRSF11A variation increases NFB activity and the severity of Paget's disease. *Journal of Bone and Mineral Research*, **2012**, 27, 443-52	6.3	31
44	Confirmation that Xq27 and Xq28 are susceptibility loci for migraine in independent pedigrees and a case-control cohort. *Neurogenetics*, **2012**, 13, 97-101	3	6
43	A functional allelic variant of the FGF23 gene is associated with renal phosphate leak in calcium nephrolithiasis. *Journal of Clinical Endocrinology and Metabolism*, **2012**, 97, E840-4	5.6	12
42	The melatonin receptor 1A (MTNR1A) gene is associated with recurrent and idiopathic calcium nephrolithiasis. *Nephrology Dialysis Transplantation*, **2012**, 27, 210-8	4.3	20

41	piR_015520 belongs to Piwi-associated RNAs regulates expression of the human melatonin receptor 1A gene. *PLoS ONE*, **2011**, 6, e22727	3.7	62
40	Genome-wide association identifies three new susceptibility loci for Paget's disease of bone. *Nature Genetics*, **2011**, 43, 685-9	36.3	134
39	Comparison of intravenous and intramuscular neridronate regimens for the treatment of Paget disease of bone. *Journal of Bone and Mineral Research*, **2011**, 26, 512-8	6.3	20
38	FSHR gene polymorphisms influence bone mineral density and bone turnover in postmenopausal women. *European Journal of Endocrinology*, **2010**, 163, 165-72	6.5	76
37	Epidemiological, clinical, and genetic characteristics of Paget's disease of bone in a rural area of Calabria, Southern Italy. *Journal of Endocrinological Investigation*, **2010**, 33, 519-25	5.2	10
36	SQSTM1 gene analysis and gene-environment interaction in Paget's disease of bone. *Journal of Bone and Mineral Research*, **2010**, 25, 1375-84	6.3	58
35	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	34
34	DDX11L: a novel transcript family emerging from human subtelomeric regions. *BMC Genomics*, **2009**, 10, 250	4.5	10
33	Bone turnover and the osteoprotegerin-RANKL pathway in tumor-induced osteomalacia: a longitudinal study of five cases. *Calcified Tissue International*, **2009**, 85, 293-300	3.9	15
32	De novo seven extra repeat expanded mutation in the PRNP gene in an Italian patient with early onset dementia. *BMJ Case Reports*, **2009**, 2009,	0.9	2
31	ZPLD1 gene is disrupted in a patient with balanced translocation that exhibits cerebral cavernous malformations. *Neuroscience*, **2008**, 155, 345-9	3.9	24
30	Genetic variants of Y chromosome are associated with a protective lipid profile in black men. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2008**, 28, 1569-74	9.4	17
29	Vitamin D receptor gene polymorphisms predict acquired resistance to clodronate treatment in patients with Paget's disease of bone. *Calcified Tissue International*, **2008**, 83, 414-24	3.9	15
28	Different spectra of genomic deletions within the CCM genes between Italian and American CCM patient cohorts. *Neurogenetics*, **2008**, 9, 25-31	3	28
27	ATP1A2 gene mutations are not present in two sisters with basilar-type migraine associated with menses. *Neurological Sciences*, **2008**, 29, 113-5	3.5	
26	Investigation of gamma-aminobutyric acid (GABA) A receptors genes and migraine susceptibility. *BMC Medical Genetics*, **2008**, 9, 109	2.1	18
25	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, 691-5	3.5	24
24	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, 1411-3	5.5	6

23	The Molecular Genetics of Migraine: Toward the Identification of Responsible Genes. *Current Genomics*, **2006**, 7, 1-10	2.6	2
22	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, 192-5	3.7	13
21	Genes, Human Diseases and Genome Evolution in the Post-Genomic Era: Insights from Uric Acid Nephrolithiasis. *Current Genomics*, **2005**, 6, 207-214	2.6	
20	Geographic distribution of Ala62Thr variant associated to Uric Acid Nephrolithiasis from Sub-Saharan to Mediterranean area. *International Journal of Anthropology*, **2004**, 19, 277-280		2
19	Genes, diet and uric acid nephrolithiasis. *International Journal of Anthropology*, **2004**, 19, 281-288		
18	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, 318-26	4.7	73
17	The evolutionary conservation of the human chitotriosidase gene in rodents and primates. *Cytogenetic and Genome Research*, **2004**, 105, 54-6	1.9	21
16	Emergence of Talanin protein associated with human uric acid nephrolithiasis in the Hominidae lineage. *Gene*, **2004**, 339, 131-8	3.8	9
15	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, 1479-91	11	60
14	Characterization of the murine orthologue of a novel human subtelomeric multigene family. *Cytogenetic and Genome Research*, **2001**, 94, 98-100	1.9	9
13	Differential divergence of three human pseudoautosomal genes and their mouse homologs: implications for sex chromosome evolution. *Genome Research*, **2001**, 11, 2095-100	9.7	31
12	Genomic rearrangement in NEMO impairs NF-kappaB activation and is a cause of incontinentia pigmenti. The International Incontinentia Pigmenti (IP) Consortium. *Nature*, **2000**, 405, 466-72	50.4	607
11	Differentially regulated and evolved genes in the fully sequenced Xq/Yq pseudoautosomal region. *Human Molecular Genetics*, **2000**, 9, 395-401	5.6	66
10	Identification and chromosomal localisation by fluorescence in situ hybridisation of human gene of phosphoinositide-specific phospholipase C beta(1). *Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids*, **2000**, 1484, 175-82	5	27
9	A novel pseudoautosomal human gene encodes a putative protein similar to Ac-like transposases. *Human Molecular Genetics*, **1999**, 8, 61-7	5.6	24
8	Molecular cloning and fine mapping of API5L1, a novel human gene strongly related to an antiapoptotic gene. *Cytogenetic and Genome Research*, **1999**, 84, 164-6	1.9	5
7	A novel pseudoautosomal gene encoding a putative GTP-binding protein resides in the vicinity of the Xp/Yp telomere. *Human Molecular Genetics*, **1998**, 7, 407-14	5.6	29
6	Escape from X inactivation of two new genes associated with DXS6974E and DXS7020E. *Genomics*, **1997**, 43, 183-90	4.3	17

5	Mapping of 59 EST gene markers in 31 intervals spanning the human X chromosome. *Gene*, **1997**, 187, 179-84	3.8	10
4	A synaptobrevin-like gene in the Xq28 pseudoautosomal region undergoes X inactivation. *Nature Genetics*, **1996**, 13, 227-9	36.3	71
3	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, 815-6	4.3	1
2	Early Alpine occupation backdates westward human migration in Late Glacial Europe		3
1	Molecular Genetics of Paget's Disease of Bone		2