Annalisa Frattini

List of Publications by Year
in descending order

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Version: 2024-02-01

70 papers	5,232 citations	186265 28 h-index	91884 69 g-index
70 all docs	70 docs citations	70 times ranked	5074 citing authors

#	Article	IF	Citations
1	Mutations of Jak-3 gene in patients with autosomal severe combined immune deficiency (SCID). Nature, 1995, 377, 65-68.	27.8	864
2	Defects in TCIRG1 subunit of the vacuolar proton pump are responsible for a subset of human autosomal recessive osteopetrosis. Nature Genetics, 2000, 25, 343-346.	21.4	629
3	Partial V(D)J Recombination Activity Leads to Omenn Syndrome. Cell, 1998, 93, 885-896.	28.9	429
4	Osteoclast-poor human osteopetrosis due to mutations in the gene encoding RANKL. Nature Genetics, 2007, 39, 960-962.	21.4	346
5	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to TNFRSF11A (RANK) Mutations. American Journal of Human Genetics, 2008, 83, 64-76.	6.2	270
6	Grey-lethal mutation induces severe malignant autosomal recessive osteopetrosis in mouse and human. Nature Medicine, 2003, 9, 399-406.	30.7	245
7	Involvement of PLEKHM1 in osteoclastic vesicular transport and osteopetrosis in incisors absent rats and humans. Journal of Clinical Investigation, 2007, 117, 919-930.	8.2	204
8	Chloride Channel ClCN7 Mutations Are Responsible for Severe Recessive, Dominant, and Intermediate Osteopetrosis. Journal of Bone and Mineral Research, 2003, 18, 1740-1747.	2.8	202
9	The mutational spectrum of human malignant autosomal recessive osteopetrosis. Human Molecular Genetics, 2001, 10, 1767-1773.	2.9	201
10	Impaired gastric acidification negatively affects calcium homeostasis and bone mass. Nature Medicine, 2009, 15, 674-681.	30.7	172
11	Mutations in OSTM1 (Grey Lethal) Define a Particularly Severe Form of Autosomal Recessive Osteopetrosis With Neural Involvement. Journal of Bone and Mineral Research, 2006, 21, 1098-1105.	2.8	97
12	Molecular Cloning of ILP-2, a Novel Member of the Inhibitor of Apoptosis Protein Family. Molecular and Cellular Biology, 2001, 21, 4292-4301.	2.3	95
13	In utero transplantation of adult bone marrow decreases perinatal lethality and rescues the bone phenotype in the knockin murine model for classical, dominant osteogenesis imperfecta. Blood, 2009, 114, 459-468.	1.4	93
14	Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. Human Molecular Genetics, 2009, 18, 418-427.	2.9	92
15	TCIRG1-dependent recessive osteopetrosis: Mutation analysis, functional identification of the splicing defects, andin vitro rescue by U1 snRNA. Human Mutation, 2004, 24, 225-235.	2.5	90
16	Molecular and clinical heterogeneity in CLCN7-dependent osteopetrosis: report of 20 novel mutations. Human Mutation, 2010, 31, E1071-E1080.	2.5	77
17	Mapping of two genes encoding isoforms of the actin binding protein ABP-280, a dystrophin like protein, to Xq28 and to chromosome 7. Human Molecular Genetics, 1993, 2, 761-766.	2.9	73
18	Organization of the human CD40L gene: implications for molecular defects in X chromosome-linked hyper-IgM syndrome and prenatal diagnosis.. Proceedings of the National Academy of Sciences of the United States of America, 1994, 91, 2110-2114.	7.1	68

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19	High variability of genomic instability and gene expression profiling in different HeLa clones. Scientific Reports, 2015, 5, 15377.	3.3	68
20	Vacuolar H+-ATPase d2 Subunit: Molecular Characterization, Developmental Regulation, and Localization to Specialized Proton Pumps in Kidney and Bone. Journal of the American Society of Nephrology: JASN, 2005, 16, 1245-1256.	6.1	59
21	Rescue of ATPa3-deficient murine malignant osteopetrosis by hematopoietic stem cell transplantation in utero. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 14629-14634.	7.1	58
22	Severe Malignant Osteopetrosis Caused by a GL Gene Mutation. Journal of Bone and Mineral Research, 2004, 19, 1194-1199.	2.8	47
23	OTX1 expression in breast cancer is regulated by p53. Oncogene, 2011, 30, 3096-3103.	5.9	41
24	Expansion Cranioplasty with Jackscrew Distracters for Craniosynostosis and Intracranial Hypertension in Transplanted Osteopetrosis. Pediatric Neurosurgery, 2007, 43, 102-106.	0.7	38
25	Nephrogenic Diabetes Insipidus. Journal of the American Society of Nephrology: JASN, 2000, 11, 1033-1043.	6.1	37
26	A nonsynonymous TNFRSF11A variation increases NFÎºB activity and the severity of Paget's disease. Journal of Bone and Mineral Research, 2012, 27, 443-452.	2.8	34
27	Polymorphisms of the CLCN7 Gene Are Associated With BMD in Women. Journal of Bone and Mineral Research, 2005, 20, 1960-1967.	2.8	31
28	The Exon-Intron Organization of the Human X-Linked Gene (FLN1) Encoding Actin-Binding Protein 280. Genomics, 1994, 21, 71-76.	2.9	29
29	Identification and genomic organization of a gene coding for a new member of the cell adhesion molecule family mapping to Xq25. Gene, 1998, 214, 1-6.	2.2	28
30	Molecular study of six families originating from the Middle-East and presenting with autosomal recessive osteopetrosis. European Journal of Medical Genetics, 2007, 50, 188-199.	1.3	28
31	Association Between a Polymorphism Affecting an AP1 Binding Site in the Promoter of the TCIRG1 Gene and Bone Mass in Women. Calcified Tissue International, 2003, 74, 35-41.	3.1	27
32	Heterogeneous gene distribution reflects human genome complexity as detected at the cytogenetic level. Cancer Genetics and Cytogenetics, 2002, 134, 168-171.	1.0	25
33	SMC1B is present in mammalian somatic cells and interacts with mitotic cohesin proteins. Scientific Reports, 2015, 5, 18472.	3.3	24
34	The human RNASET2 protein affects the polarization pattern of human macrophages in vitro. Immunology Letters, 2018, 203, 102-111.	2.5	24
35	ShwachmanâĐiamond syndrome with clonal interstitial deletion of the long arm of chromosome 20 in bone marrow: haematological features, prognosis and genomic instability. British Journal of Haematology, 2019, 184, 974-981.	2.5	24
36	ZNF75: Isolation of a cDNA Clone of the KRAB Zinc Finger Gene Subfamily Mapped in YACs 1 Mb Telomeric of HPRT. Genomics, 1993, 18, 223-229.	2.9	19

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37	Identification of a new member (ZNF183) of the Ring finger gene family in Xq24-25. Gene, 1997, 192, 291-298.	2.2	17
38	Brain lipid composition in grey-lethal mutant mouse characterized by severe malignant osteopetrosis. Glycoconjugate Journal, 2009, 26, 623-633.	2.7	17
39	A new mutation (TTR Ala-47) in the transthyretin gene associated with hereditary amyloidosis. Human Mutation, 1994, 4, 61-64.	2.5	16
40	Characterization and Fine Localization of Two New Genes in Xq28 Using the Genomic Sequence/EST Database Screening Approach. Genomics, 1996, 34, 323-327.	2.9	16
41	Comparative genomic hybridization on microarray (aâ€«scp›CGH‹/scp›) in olfactory neuroblastoma: Analysis of ten cases and review of the literature. Genes Chromosomes and Cancer, 2015, 54, 771-775.	2.8	16
42	Novel recurrent chromosome anomalies in Shwachmanâ€“Diamond syndrome. Pediatric Blood and Cancer, 2017, 64, e26454.	1.5	16
43	Genomic Organization of the Human VP16 Accessory Protein, a Housekeeping Gene (HCFC1) Mapping to Xq28. Genomics, 1994, 23, 30-35.	2.9	15
44	The Chromosome Localization and the HCF Repeats of the Human Host Cell Factor Gene (HCFC1) Are Conserved in the Mouse Homologue. Genomics, 1996, 32, 277-280.	2.9	15
45	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.	2.1	13
46	In vitro and in vivo antisense-mediated growth inhibition of a mammary adenocarcinoma from MMTV-neu transgenic mice. Gene Therapy, 1998, 5, 388-393.	4.5	13
47	Osteopetroses and immunodeficiencies in humans. Current Opinion in Allergy and Clinical Immunology, 2006, 6, 421-427.	2.3	13
48	Electron Microscopic Findings in Skin Biopsies from Patients with Infantile Osteopetrosis and Neuronal Storage Disease. Ultrastructural Pathology, 2007, 31, 333-338.	0.9	13
49	Isolation of a zinc finger motif (ZNF75) mapping on chromosome Xq26. Genomics, 1992, 13, 1231-1236.	2.9	12
50	The ZNF75 Zinc Finger Gene Subfamily: Isolation and Mapping of the Four Members in Humans and Great Apes. Genomics, 1996, 35, 312-320.	2.9	12
51	Chromosome Missegregation in Single Human Oocytes Is Related to the Age and Gene Expression Profile. International Journal of Molecular Sciences, 2020, 21, 1934.	4.1	12
52	The Dissection of Human Autosomal Recessive Osteopetrosis Identifies an Osteoclast-Poor Form Due to RANKL Deficiency. Cell Cycle, 2007, 6, 3027-3033.	2.6	11
53	Characterization of a Novel Alu-Alu Recombination-Mediated Genomic Deletion in the ‹i›TCIRG1‹/i› Gene in Five Osteopetrotic Patients. Journal of Bone and Mineral Research, 2009, 24, 162-167.	2.8	11
54	The Mammary Gland and the Homeobox Gene Otx1. Breast Journal, 2010, 16, S53-S56.	1.0	11

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55	Paracrine effect of human adipose-derived stem cells on lymphatic endothelial cells. Regenerative Medicine, 2020, 15, 2085-2098.	1.7	11
56	The frequent and clinically benign anomalies of chromosomes 7 and 20 in Shwachman-diamond syndrome may be subject to further clonal variations. Molecular Cytogenetics, 2021, 14, 54.	0.9	10
57	The human genes encoding renin-binding protein and host cell factor are closely linked in Xq28 and transcribed in the same direction. Gene, 1995, 155, 237-239.	2.2	8
58	Chloride Channel CICN7 Mutations in a Korean Patient with Infantile Malignant Osteopetrosis Initially Presenting with Neonatal Thrombocytopenia. Journal of Perinatology, 2004, 24, 312-314.	2.0	8
59	Microarray expression studies on bone marrow of patients with Shwachman-Diamond syndrome in relation to deletion of the long arm of chromosome 20, other chromosome anomalies or normal karyotype. Molecular Cytogenetics, 2020, 13, 1.	0.9	8
60	A Low-Copy Repeat in Xq26 Represents a Novel Putatively Prenylated Protein Gene (CXX1) and Its Pseudogenes (DXS9914, DXS9915, and DXS9916). Genomics, 1997, 46, 167-169.	2.9	7
61	A Homozygous Contiguous Gene Deletion in Chromosome 16p13.3 Leads to Autosomal Recessive Osteopetrosis in a Jordanian Patient. Calcified Tissue International, 2012, 91, 250-254.	3.1	7
62	Toxicogenomics applied to in vitro Cell Transformation Assay reveals mechanisms of early response to cadmium. Toxicology in Vitro, 2018, 48, 232-243.	2.4	7
63	A new familial sclerosing bone dysplasia. Journal of Bone and Mineral Research, 2010, 25, 676-680.	2.8	6
64	Computer Gene Mapping byEagI-Based STSs. Genomics, 1996, 38, 87-91.	2.9	5
65	Prognostic potential of precise molecular diagnosis of Autosomal Recessive Osteopetrosis with respect to the outcome of bone marrow transplantation. Cytotechnology, 2008, 58, 57-62.	1.6	5
66	The Complete Sequence of the Host Cell Factor 1 (HCFC1) Gene and Its Promoter: A Role for YY1 Transcription Factor in the Regulation of Its Expression. Genomics, 1996, 34, 85-91.	2.9	4
67	Shwachman-Diamond syndrome: diagnosis, pathogenesis and prognosis. Expert Opinion on Orphan Drugs, 2017, 5, 753-767.	0.8	4
68	Enhanced p53 Levels Are Involved in the Reduced Mineralization Capacity of Osteoblasts Derived from Shwachmanâ€“Diamond Syndrome Subjects. International Journal of Molecular Sciences, 2021, 22, 13331.	4.1	3
69	The genetics of dominant osteopetrosis. Drug Discovery Today Disease Mechanisms, 2005, 2, 503-509.	0.8	2
70	Fidelity of a YAC clone in the region of human MCF-2 gene. Biochemical and Biophysical Research Communications, 1991, 181, 877-883.	2.1	0