Annalisa Frattini

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2953256/publications.pdf

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	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
2	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
3	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
4	Paracrine effect of human adipose-derived stem cells on lymphatic endothelial cells. Regenerative Medicine, 2020, 15, 2085-2098.	1.7	11
5	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
6	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
7	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
8	The human RNASET2 protein affects the polarization pattern of human macrophages in vitro. Immunology Letters, 2018, 203, 102-111.	2.5	24
9	Novel recurrent chromosome anomalies in Shwachmanâ€“Diamond syndrome. Pediatric Blood and Cancer, 2017, 64, e26454.	1.5	16
10	Shwachman-Diamond syndrome: diagnosis, pathogenesis and prognosis. Expert Opinion on Orphan Drugs, 2017, 5, 753-767.	0.8	4
11	SMC1B is present in mammalian somatic cells and interacts with mitotic cohesin proteins. Scientific Reports, 2015, 5, 18472.	3.3	24
12	High variability of genomic instability and gene expression profiling in different HeLa clones. Scientific Reports, 2015, 5, 15377.	3.3	68
13	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
14	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
15	A nonsynonymous ‹i›TNFRSF11A‹/i› variation increases NFÎºB activity and the severity of Paget's disease. Journal of Bone and Mineral Research, 2012, 27, 443-452.	2.8	34
16	OTX1 expression in breast cancer is regulated by p53. Oncogene, 2011, 30, 3096-3103.	5.9	41
17	A new familial sclerosing bone dysplasia. Journal of Bone and Mineral Research, 2010, 25, 676-680.	2.8	6
18	Molecular and clinical heterogeneity in CLCN7-dependent osteopetrosis: report of 20 novel mutations. Human Mutation, 2010, 31, E1071-E1080.	2.5	77

#	Article	IF	Citations
19	The Mammary Gland and the Homeobox Gene Otx1. Breast Journal, 2010, 16, S53-S56.	1.0	11
20	Brain lipid composition in grey-lethal mutant mouse characterized by severe malignant osteopetrosis. Glycoconjugate Journal, 2009, 26, 623-633.	2.7	17
21	Impaired gastric acidification negatively affects calcium homeostasis and bone mass. Nature Medicine, 2009, 15, 674-681.	30.7	172
22	Characterization of a Novel Alu-Alu Recombination-Mediated Genomic Deletion in the TCIRG1 Gene in Five Osteopetrotic Patients. Journal of Bone and Mineral Research, 2009, 24, 162-167.	2.8	11
23	Cornelia de Lange syndrome mutations in SMC1A or SMC3 affect binding to DNA. Human Molecular Genetics, 2009, 18, 418-427.	2.9	92
24	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
25	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
26	Human Osteoclast-Poor Osteopetrosis with Hypogammaglobulinemia due to TNFRSF11A (RANK) Mutations. American Journal of Human Genetics, 2008, 83, 64-76.	6.2	270
27	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
28	Expansion Cranioplasty with Jackscrew Distracters for Craniosynostosis and Intracranial Hypertension in Transplanted Osteopetrosis. Pediatric Neurosurgery, 2007, 43, 102-106.	0.7	38
29	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
30	Electron Microscopic Findings in Skin Biopsies from Patients with Infantile Osteopetrosis and Neuronal Storage Disease. Ultrastructural Pathology, 2007, 31, 333-338.	0.9	13
31	Osteoclast-poor human osteopetrosis due to mutations in the gene encoding RANKL. Nature Genetics, 2007, 39, 960-962.	21.4	346
32	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
33	Osteopetroses and immunodeficiencies in humans. Current Opinion in Allergy and Clinical Immunology, 2006, 6, 421-427.	2.3	13
34	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
35	Polymorphisms of the CLCN7 Gene Are Associated With BMD in Women. Journal of Bone and Mineral Research, 2005, 20, 1960-1967.	2.8	31
36	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

#	Article	IF	Citations
37	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
38	The genetics of dominant osteopetrosis. Drug Discovery Today Disease Mechanisms, 2005, 2, 503-509.	0.8	2
39	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
40	Severe Malignant Osteopetrosis Caused by a GL Gene Mutation. Journal of Bone and Mineral Research, 2004, 19, 1194-1199.	2.8	47
41	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
42	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
43	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
44	Grey-lethal mutation induces severe malignant autosomal recessive osteopetrosis in mouse and human. Nature Medicine, 2003, 9, 399-406.	30.7	245
45	Heterogeneous gene distribution reflects human genome complexity as detected at the cytogenetic level. Cancer Genetics and Cytogenetics, 2002, 134, 168-171.	1.0	25
46	The mutational spectrum of human malignant autosomal recessive osteopetrosis. Human Molecular Genetics, 2001, 10, 1767-1773.	2.9	201
47	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
48	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
49	Nephrogenic Diabetes Insipidus. Journal of the American Society of Nephrology: JASN, 2000, 11, 1033-1043.	6.1	37
50	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
51	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
52	Partial V(D)J Recombination Activity Leads to Omenn Syndrome. Cell, 1998, 93, 885-896.	28.9	429
53	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
54	Identification of a new member (ZNF183) of the Ring finger gene family in Xq24-25. Gene, 1997, 192, 291-298.	2.2	17

#	Article	IF	Citations
55	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
56	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
57	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
58	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
59	Computer Gene Mapping byEagI-Based STSs. Genomics, 1996, 38, 87-91.	2.9	5
60	Mutations of Jak-3 gene in patients with autosomal severe combined immune deficiency (SCID). Nature, 1995, 377, 65-68.	27.8	864
61	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
62	A new mutation (TTR Ala-47) in the transthyretin gene associated with hereditary amyloidosis. Human Mutation, 1994, 4, 61-64.	2.5	16
63	The Exon-Intron Organization of the Human X-Linked Gene (FLN1) Encoding Actin-Binding Protein 280. Genomics, 1994, 21, 71-76.	2.9	29
64	Genomic Organization of the Human VP16 Accessory Protein, a Housekeeping Gene (HCFC1) Mapping to Xq28. Genomics, 1994, 23, 30-35.	2.9	15
65	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
66	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
67	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
68	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
69	Isolation of a zinc finger motif (ZNF75) mapping on chromosome Xq26. Genomics, 1992, 13, 1231-1236.	2.9	12
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