

Laurence Legeai-Mallet

List of Publications by Year in descending order

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Version: 2024-02-01

58
papers

4,110
citations

136950

32
h-index

155660

55
g-index

61
all docs

61
docs citations

61
times ranked

4095
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in the gene encoding fibroblast growth factor receptor-3 in achondroplasia. <i>Nature</i> , 1994, 371, 252-254.	27.8	873
2	ADAMTS10 Mutations in Autosomal Recessive Weill-Marchesani Syndrome. <i>American Journal of Human Genetics</i> , 2004, 75, 801-806.	6.2	255
3	A gene for hereditary multiple exostoses maps to chromosome 19p. <i>Human Molecular Genetics</i> , 1994, 3, 717-722.	2.9	199
4	Null Leukemia Inhibitory Factor Receptor (LIFR) Mutations in StÅ¼ve-Wiedemann/Schwartz-Jampel Type 2 Syndrome. <i>American Journal of Human Genetics</i> , 2004, 74, 298-305.	6.2	162
5	Spatio-temporal expression of FGFR 1, 2 and 3 genes during human embryo-fetal ossification. <i>Mechanisms of Development</i> , 1998, 77, 19-30.	1.7	161
6	Achondroplasia: Development, pathogenesis, and therapy. <i>Developmental Dynamics</i> , 2017, 246, 291-309.	1.8	160
7	Evaluation of the Therapeutic Potential of a CNP Analog in a Fgfr3 Mouse Model Recapitulating Achondroplasia. <i>American Journal of Human Genetics</i> , 2012, 91, 1108-1114.	6.2	155
8	Incomplete penetrance and expressivity skewing in hereditary multiple exostoses. <i>Clinical Genetics</i> , 1997, 52, 12-16.	2.0	132
9	Distinct Effects of Allelic NFIX Mutations on Nonsense-Mediated mRNA Decay Engender Either a Sotos-like or a Marshall-Smith Syndrome. <i>American Journal of Human Genetics</i> , 2010, 87, 189-198.	6.2	131
10	A gene for achondroplasiaâ€“hypochondroplasia maps to chromosome 4p. <i>Nature Genetics</i> , 1994, 6, 318-321.	21.4	128
11	Fibroblast Growth Factor Receptor 3 Mutations Promote Apoptosis but Do Not Alter Chondrocyte Proliferation in Thanatophoric Dysplasia. <i>Journal of Biological Chemistry</i> , 1998, 273, 13007-13014.	3.4	127
12	Achondroplasia. <i>Best Practice and Research in Clinical Rheumatology</i> , 2008, 22, 3-18.	3.3	117
13	Hereditary multiple exostoses and enchondromatosis. <i>Best Practice and Research in Clinical Rheumatology</i> , 2008, 22, 45-54.	3.3	104
14	Tyrosine kinase inhibitor NVP-BGJ398 functionally improves FGFR3-related dwarfism in mouse model. <i>Journal of Clinical Investigation</i> , 2016, 126, 1871-1884.	8.2	84
15	Polymerase Î¼1 mutation in a human syndrome with facial dysmorphism, immunodeficiency, livedo, and short stature (â€œFELS syndromeâ€œ). <i>Journal of Experimental Medicine</i> , 2012, 209, 2323-2330.	8.5	83
16	A cluster of translocation breakpoints in 2q37 is associated with overexpression of NPPC in patients with a similar overgrowth phenotype. <i>Human Mutation</i> , 2007, 28, 1183-1188.	2.5	79
17	Novel FGFR3 mutations creating cysteine residues in the extracellular domain of the receptor cause achondroplasia or severe forms of hypochondroplasia. <i>European Journal of Human Genetics</i> , 2006, 14, 1240-1247.	2.8	75
18	Thromboxane synthase mutations in an increased bone density disorder (Ghosal syndrome). <i>Nature Genetics</i> , 2008, 40, 284-286.	21.4	61

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19	FGFR3 mutation causes abnormal membranous ossification in achondroplasia. <i>Human Molecular Genetics</i> , 2014, 23, 2914-2925.	2.9	57
20	Mutations in the basic domain and the loop-helix II junction of TWIST abolish DNA binding in Saethre-Chotzen syndrome. <i>FEBS Letters</i> , 2001, 492, 112-118.	2.8	55
21	Synthesis and biological evaluation of a triazole-based library of pyrido[2,3-d]pyrimidines as FGFR3 tyrosine kinase inhibitors. <i>Organic and Biomolecular Chemistry</i> , 2010, 8, 2164.	2.8	53
22	A gene for Holtâ€™Oram syndrome maps to the distal long arm of chromosome 12. <i>Nature Genetics</i> , 1994, 6, 405-408.	21.4	51
23	Activating Fgfr3 Y367C mutation causes hearing loss and inner ear defect in a mouse model of chondrodysplasia. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2009, 1792, 140-147.	3.8	49
24	Mutations of the Fibroblast Growth Factor Receptor-3 Gene in Achondroplasia. <i>Hormone Research</i> , 1996, 45, 108-110.	1.8	47
25	Defective chondrocyte proliferation and differentiation in osteochondromas of MHE patients. <i>Bone</i> , 2006, 39, 17-26.	2.9	44
26	Hyperphosphatasia with seizures, neurologic deficit, and characteristic facial features: Five new patients with Mabry syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1661-1669.	1.2	42
27	Homozygosity mapping of a Weill-Marchesani syndrome locus to chromosome 19p13.3-p13.2. <i>Human Genetics</i> , 2002, 110, 366-370.	3.8	41
28	An activating Fgfr3 mutation affects trabecular bone formation via a paracrine mechanism during growth. <i>Human Molecular Genetics</i> , 2012, 21, 2503-2513.	2.9	40
29	A novel tyrosine kinase inhibitor restores chondrocyte differentiation and promotes bone growth in a gain-of-function Fgfr3 mouse model. <i>Human Molecular Genetics</i> , 2012, 21, 841-851.	2.9	40
30	Constitutively-active FGFR3 disrupts primary cilium length and IFT20 trafficking in various chondrocyte models of achondroplasia. <i>Human Molecular Genetics</i> , 2018, 27, 1-13.	2.9	37
31	An extension of the admixture test for the study of genetic heterogeneity in hereditary multiple exostoses. <i>Human Genetics</i> , 1997, 99, 298-302.	3.8	36
32	The Impairment of MAGMAS Function in Human Is Responsible for a Severe Skeletal Dysplasia. <i>PLoS Genetics</i> , 2014, 10, e1004311.	3.5	34
33	Chondrocytes Play a Major Role in the Stimulation of Bone Growth by Thyroid Hormone. <i>Endocrinology</i> , 2014, 155, 3123-3135.	2.8	34
34	C-Type Natriuretic Peptide Analog as Therapy for Achondroplasia. <i>Endocrine Development</i> , 2016, 30, 98-105.	1.3	31
35	Novel therapeutic approaches for the treatment of achondroplasia. <i>Bone</i> , 2020, 141, 115579.	2.9	31
36	FGFR3 intracellular mutations induce tyrosine phosphorylation in the Golgi and defective glycosylation. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , 2007, 1773, 502-512.	4.1	30

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37	EXT 1 Gene Mutation Induces Chondrocyte Cytoskeletal Abnormalities and Defective Collagen Expression in the Exostoses. <i>Journal of Bone and Mineral Research</i> , 2000, 15, 1489-1500.	2.8	28
38	FGFR3 in Periosteal Cells Drives Cartilage-to-Bone Transformation in Bone Repair. <i>Stem Cell Reports</i> , 2020, 15, 955-967.	4.8	27
39	Delayed bone age due to a dual effect of FGFR3 mutation in Achondroplasia. <i>Bone</i> , 2010, 47, 905-915.	2.9	23
40	Central nervous system malformations and deformations in <i>FGFR2</i> -related craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2797-2806.	1.2	23
41	Crouzon syndrome with acanthosis nigricans: a case-based update. <i>Child's Nervous System</i> , 2011, 27, 349-354.	1.1	22
42	Meckel's and condylar cartilages anomalies in achondroplasia result in defective development and growth of the mandible. <i>Human Molecular Genetics</i> , 2016, 25, ddw153.	2.9	21
43	Parathyroid Hormone Receptor Type 1/Indian Hedgehog Expression Is Preserved in the Growth Plate of Human Fetuses Affected with Fibroblast Growth Factor Receptor Type 3 Activating Mutations. <i>American Journal of Pathology</i> , 2002, 161, 1325-1335.	3.8	20
44	Thanatophoric dysplasia caused by double missense <i>FGFR3</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 1296-1301.	1.2	19
45	New Insight on FGFR3-Related Chondrodysplasias Molecular Physiopathology Revealed by Human Chondrocyte Gene Expression Profiling. <i>PLoS ONE</i> , 2009, 4, e7633.	2.5	19
46	<i>Fgfr3</i> Is a Positive Regulator of Osteoblast Expansion and Differentiation During Zebrafish Skull Vault Development. <i>Journal of Bone and Mineral Research</i> , 2020, 35, 1782-1797.	2.8	18
47	Prevention of guanylyl cyclase β dephosphorylation rescues achondroplastic dwarfism. <i>JCI Insight</i> , 2021, 6, .	5.0	12
48	Human immortalized chondrocytes carrying heterozygous FGFR3 mutations: An in vitro model to study chondrodysplasias. <i>FEBS Letters</i> , 2007, 581, 2593-2598.	2.8	10
49	Phosphatase inhibition by LB-100 enhances BMN-111 stimulation of bone growth. <i>JCI Insight</i> , 2021, 6, .	5.0	9
50	An <i>Fgfr3</i> -activating mutation in immature murine osteoblasts affects the appendicular and craniofacial skeleton. <i>DMM Disease Models and Mechanisms</i> , 2021, 14, .	2.4	6
51	Molecular modeling study of the induced-fit effect on kinase inhibition: the case of fibroblast growth factor receptor 3 (FGFR3). <i>Journal of Computer-Aided Molecular Design</i> , 2015, 29, 619-641.	2.9	3
52	Corrigendum to "Hyperphosphatasia With Seizures, Neurologic Deficit, and Characteristic Facial Features: Five New Patients With Mabry Syndrome" <i>Am J Med Genet 152A</i> : 1661-1669. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1215-1215.	1.2	2
53	Growth charts in FGFR2- and FGFR3-related faciocraniosynostoses. <i>Bone Reports</i> , 2022, 16, 101524.	0.4	2
54	FGFR3 overactivation in the brain is responsible for memory impairments in Crouzon syndrome mouse model.. <i>Journal of Experimental Medicine</i> , 2022, 219, .	8.5	2

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55	The impact of polyphenols on chondrocyte growth and survival: a preliminary report. Food and Nutrition Research, 2015, 59, 29311.	2.6	1
56	Key challenges in the treatment of rare pediatric skeletal genetic disorders: from bench to bedside. Drug Discovery Today, 2015, 20, 781-783.	6.4	1
57	Les exostosines : des protéines impliquées dans la biosynthèse des héparanes sulfates. Medecine/Sciences, 2002, 18, 23-25.	0.2	0
58	Theobroma cacao improves bone growth by modulating defective ciliogenesis in a mouse model of achondroplasia. Bone Research, 2022, 10, 8.	11.4	0