

# Laurence Legeai-Mallet

## List of Publications by Citations

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58  
papers

3,335  
citations

29  
h-index

57  
g-index

61  
ext. papers

3,757  
ext. citations

8.5  
avg, IF

4.73  
L-index

#	Paper	IF	Citations
58	Mutations in the gene encoding fibroblast growth factor receptor-3 in achondroplasia. <i>Nature</i> , <b>1994</b> , 371, 252-4	50.4	745
57	ADAMTS10 mutations in autosomal recessive Weill-Marchesani syndrome. <i>American Journal of Human Genetics</i> , <b>2004</b> , 75, 801-6	11	214
56	A gene for hereditary multiple exostoses maps to chromosome 19p. <i>Human Molecular Genetics</i> , <b>1994</b> , 3, 717-22	5.6	176
55	Spatio-temporal expression of FGFR 1, 2 and 3 genes during human embryo-fetal ossification. <i>Mechanisms of Development</i> , <b>1998</b> , 77, 19-30	1.7	146
54	Null leukemia inhibitory factor receptor (LIFR) mutations in Stuve-Wiedemann/Schwartz-Jampel type 2 syndrome. <i>American Journal of Human Genetics</i> , <b>2004</b> , 74, 298-305	11	141
53	Evaluation of the therapeutic potential of a CNP analog in a Fgfr3 mouse model recapitulating achondroplasia. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 1108-14	11	116
52	Fibroblast growth factor receptor 3 mutations promote apoptosis but do not alter chondrocyte proliferation in thanatophoric dysplasia. <i>Journal of Biological Chemistry</i> , <b>1998</b> , 273, 13007-14	5.4	116
51	Achondroplasia: Development, pathogenesis, and therapy. <i>Developmental Dynamics</i> , <b>2017</b> , 246, 291-309	2.9	109
50	Incomplete penetrance and expressivity skewing in hereditary multiple exostoses. <i>Clinical Genetics</i> , <b>1997</b> , 52, 12-6	4	109
49	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> , <b>2010</b> , 87, 189-98	11	107
48	A gene for achondroplasia-hypochondroplasia maps to chromosome 4p. <i>Nature Genetics</i> , <b>1994</b> , 6, 318-21	6.3	102
47	Achondroplasia. <i>Best Practice and Research in Clinical Rheumatology</i> , <b>2008</b> , 22, 3-18	5.3	92
46	Hereditary multiple exostoses and enchondromatosis. <i>Best Practice and Research in Clinical Rheumatology</i> , <b>2008</b> , 22, 45-54	5.3	78
45	A cluster of translocation breakpoints in 2q37 is associated with overexpression of NPPC in patients with a similar overgrowth phenotype. <i>Human Mutation</i> , <b>2007</b> , 28, 1183-8	4.7	72
44	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> , <b>2006</b> , 14, 1240-7	5.3	64
43	Polymerase $\beta$ mutation in a human syndrome with facial dysmorphism, immunodeficiency, livedo, and short stature ("FILS syndrome"). <i>Journal of Experimental Medicine</i> , <b>2012</b> , 209, 2323-30	16.6	55
42	Thromboxane synthase mutations in an increased bone density disorder (Ghosal syndrome). <i>Nature Genetics</i> , <b>2008</b> , 40, 284-6	36.3	55

41	Tyrosine kinase inhibitor NVP-BGJ398 functionally improves FGFR3-related dwarfism in mouse model. <i>Journal of Clinical Investigation</i> , <b>2016</b> , 126, 1871-84	15.9	51
40	Mutations in the basic domain and the loop-helix II junction of TWIST abolish DNA binding in Saethre-Chotzen syndrome. <i>FEBS Letters</i> , <b>2001</b> , 492, 112-8	3.8	45
39	FGFR3 mutation causes abnormal membranous ossification in achondroplasia. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 2914-25	5.6	44
38	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> , <b>2010</b> , 8, 2164-73	3.9	44
37	A gene for Holt-Oram syndrome maps to the distal long arm of chromosome 12. <i>Nature Genetics</i> , <b>1994</b> , 6, 405-8	36.3	44
36	Mutations of the fibroblast growth factor receptor-3 gene in achondroplasia. <i>Hormone Research</i> , <b>1996</b> , 45, 108-10		43
35	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> , <b>2009</b> , 1792, 140-7	6.9	41
34	Defective chondrocyte proliferation and differentiation in osteochondromas of MHE patients. <i>Bone</i> , <b>2006</b> , 39, 17-26	4.7	39
33	An activating Fgfr3 mutation affects trabecular bone formation via a paracrine mechanism during growth. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 2503-13	5.6	38
32	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> , <b>2012</b> , 21, 841-51	5.6	34
31	An extension of the admixture test for the study of genetic heterogeneity in hereditary multiple exostoses. <i>Human Genetics</i> , <b>1997</b> , 99, 298-302	6.3	33
30	Hyperphosphatasia with seizures, neurologic deficit, and characteristic facial features: Five new patients with Mabry syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2010</b> , 152A, 1661-9	2.5	32
29	Chondrocytes play a major role in the stimulation of bone growth by thyroid hormone. <i>Endocrinology</i> , <b>2014</b> , 155, 3123-35	4.8	29
28	Homozygosity mapping of a Weill-Marchesani syndrome locus to chromosome 19p13.3-p13.2. <i>Human Genetics</i> , <b>2002</b> , 110, 366-70	6.3	29
27	The impairment of MAGMAS function in human is responsible for a severe skeletal dysplasia. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004311	6	26
26	Constitutively-active FGFR3 disrupts primary cilium length and IFT20 trafficking in various chondrocyte models of achondroplasia. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 1-13	5.6	25
25	C-Type Natriuretic Peptide Analog as Therapy for Achondroplasia. <i>Endocrine Development</i> , <b>2016</b> , 30, 98-105		23
24	EXT 1 gene mutation induces chondrocyte cytoskeletal abnormalities and defective collagen expression in the exostoses. <i>Journal of Bone and Mineral Research</i> , <b>2000</b> , 15, 1489-500	6.3	23

23	Central nervous system malformations and deformations in FGFR2-related craniosynostosis. <i>American Journal of Medical Genetics, Part A</i> , <b>2012</b> , 158A, 2797-806	2.5	22
22	Delayed bone age due to a dual effect of FGFR3 mutation in Achondroplasia. <i>Bone</i> , <b>2010</b> , 47, 905-15	4.7	22
21	FGFR3 intracellular mutations induce tyrosine phosphorylation in the Golgi and defective glycosylation. <i>Biochimica Et Biophysica Acta - Molecular Cell Research</i> , <b>2007</b> , 1773, 502-12	4.9	22
20	Crouzon syndrome with acanthosis nigricans: a case-based update. <i>Childs Nervous System</i> , <b>2011</b> , 27, 349-54	5.4	17
19	New insight on FGFR3-related chondrodysplasias molecular physiopathology revealed by human chondrocyte gene expression profiling. <i>PLoS ONE</i> , <b>2009</b> , 4, e7633	3.7	17
18	Thanatophoric dysplasia caused by double missense FGFR3 mutations. <i>American Journal of Medical Genetics, Part A</i> , <b>2009</b> , 149A, 1296-301	2.5	16
17	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> , <b>2002</b> , 161, 1325-35	5.8	16
16	Meckel's and condylar cartilages anomalies in achondroplasia result in defective development and growth of the mandible. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 2997-3010	5.6	14
15	Novel therapeutic approaches for the treatment of achondroplasia. <i>Bone</i> , <b>2020</b> , 141, 115579	4.7	10
14	Human immortalized chondrocytes carrying heterozygous FGFR3 mutations: an in vitro model to study chondrodysplasias. <i>FEBS Letters</i> , <b>2007</b> , 581, 2593-8	3.8	9
13	FGFR3 in Periosteal Cells Drives Cartilage-to-Bone Transformation in Bone Repair. <i>Stem Cell Reports</i> , <b>2020</b> , 15, 955-967	8	7
12	Fgfr3 Is a Positive Regulator of Osteoblast Expansion and Differentiation During Zebrafish Skull Vault Development. <i>Journal of Bone and Mineral Research</i> , <b>2020</b> , 35, 1782-1797	6.3	6
11	Prevention of guanylyl cyclase-B dephosphorylation rescues achondroplastic dwarfism. <i>JCI Insight</i> , <b>2021</b> , 6,	9.9	4
10	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> , <b>2015</b> , 29, 619-41	4.2	3
9	An Fgfr3-activating mutation in immature murine osteoblasts affects the appendicular and craniofacial skeleton. <i>DMM Disease Models and Mechanisms</i> , <b>2021</b> , 14,	4.1	3
8	The impact of polyphenols on chondrocyte growth and survival: a preliminary report. <i>Food and Nutrition Research</i> , <b>2015</b> , 59, 29311	3.1	1
7	FGFR3 is a positive regulator of osteoblast expansion and differentiation during zebrafish skull vault development		1
6	The phosphatase inhibitor LB-100 acts synergistically with the NPR2 agonist BMN-111 to improve bone growth		1

- 5 Phosphatase inhibition by LB-100 enhances BMN-111 stimulation of bone growth. *JCI Insight*, **2021**, 6, 9.9 1
- 4 Growth charts in - and -related faciocraniosynostoses.. *Bone Reports*, **2022**, 16, 101524 2.6 1
- 3 Corrigendum to Hyperphosphatasia With Seizures, Neurologic Deficit, and Characteristic Facial Features: Five New Patients With Mabry Syndrome. *Am J Med Genet* 152A: 1661-1669 **2011**, 155, 1215-1215
- 2 Les exostoses : des protéines impliquées dans la biosynthèse des héparanes sulfates. *Medecine/Sciences*, **2002**, 18, 23-25
- 1 Theobroma cacao improves bone growth by modulating defective ciliogenesis in a mouse model of achondroplasia.. *Bone Research*, **2022**, 10, 8 13.3