Laurence Duplomb

List of Publications by Year
in descending order

56 papers	2,527 citations	186209 28 h-index	197736 49 g-index
56 all docs	56 docs citations	56 times ranked	4712 citing authors

#	Article	IF	Citations
1	The dual role of IL-6-type cytokines on bone remodeling and bone tumors. Cytokine and Growth Factor Reviews, 2009, 20, 19-28.	3.2	168
2	PIK3R1 Mutations Cause Syndromic Insulin Resistance with Lipoatrophy. American Journal of Human Genetics, 2013, 93, 141-149.	2.6	162
3	Interleukinâ€34 is expressed by giant cell tumours of bone and plays a key role in RANKLâ€induced osteoclastogenesis. Journal of Pathology, 2010, 221, 77-86.	2.1	136
4	Hyperleptinemia prevents lipotoxic cardiomyopathy in acyl CoA synthase transgenic mice. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 13624-13629.	3.3	133
5	Interleukin-6 Inhibits Receptor Activator of Nuclear Factor ÎºB Ligand-Induced Osteoclastogenesis by Diverting Cells into the Macrophage Lineage: Key Role of Serine727 Phosphorylation of Signal Transducer and Activator of Transcription 3. Endocrinology, 2008, 149, 3688-3697.	1.4	129
6	Osteoprotegerin: Multiple partners for multiple functions. Cytokine and Growth Factor Reviews, 2013, 24, 401-409.	3.2	115
7	TheDYRK1Agene is a cause of syndromic intellectual disability with severe microcephaly and epilepsy. Journal of Medical Genetics, 2012, 49, 731-736.	1.5	103
8	Hepatic insig-1 or -2 overexpression reduces lipogenesis in obese Zucker diabetic fatty rats and in fasted/refed normal rats. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 7106-7111.	3.3	98
9	In-Frame Mutations in Exon 1 of SKI Cause Dominant Shprintzen-Goldberg Syndrome. American Journal of Human Genetics, 2012, 91, 950-957.	2.6	95
10	Oncostatin M Regulates the Synthesis and Turnover of gp130, Leukemia Inhibitory Factor Receptor Î±, and Oncostatin M Receptor Î² by Distinct Mechanisms. Journal of Biological Chemistry, 2001, 276, 47038-47045.	1.6	87
11	Concise Review: Embryonic Stem Cells: A New Tool to Study Osteoblast and Osteoclast Differentiation. Stem Cells, 2007, 25, 544-552.	1.4	83
12	Proteoglycans: key partners in bone cell biology. BioEssays, 2007, 29, 758-771.	1.2	81
13	Cohen syndrome is associated with major glycosylation defects. Human Molecular Genetics, 2014, 23, 2391-2399.	1.4	79
14	Mannose 6-Phosphate/Insulin-like Growth Factor II Receptor Mediates Internalization and Degradation of Leukemia Inhibitory Factor but Not Signal Transduction. Journal of Biological Chemistry, 1999, 274, 24685-24693.	1.6	72
15	Gallium modulates osteoclastic bone resorption in vitro without affecting osteoblasts. British Journal of Pharmacology, 2010, 159, 1681-1692.	2.7	69
16	Stimulation of Leukemia Inhibitory Factor Receptor Degradation by Extracellular Signal-regulated Kinase. Journal of Biological Chemistry, 2000, 275, 28793-28801.	1.6	64
17	The Mannose 6-Phosphate/Insulin-like Growth Factor II Receptor Is a Nanomolar Affinity Receptor for Glycosylated Human Leukemia Inhibitory Factor. Journal of Biological Chemistry, 1998, 273, 20886-20893.	1.6	62
18	Factor VIII-von Willebrand Factor Complex Inhibits Osteoclastogenesis and Controls Cell Survival. Journal of Biological Chemistry, 2009, 284, 31704-31713.	1.6	58

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19	A de novo microdeletion of SEMA5A in a boy with autism spectrum disorder and intellectual disability. European Journal of Human Genetics, 2016, 24, 838-843.	1.4	40
20	IntragenicCAMTA1rearrangements cause non-progressive congenital ataxia with or without intellectual disability. Journal of Medical Genetics, 2012, 49, 400-408.	1.5	39
21	Autosomal-recessive SASH1 variants associated with a new genodermatosis with pigmentation defects, palmoplantar keratoderma and skin carcinoma. European Journal of Human Genetics, 2015, 23, 957-962.	1.4	39
22	Increased expression and activity of 11Î²-HSD-1 in diabetic islets and prevention with troglitazone. Biochemical and Biophysical Research Communications, 2004, 313, 594-599.	1.0	37
23	Changing facial phenotype in Cohen syndrome: towards clues for an earlier diagnosis. European Journal of Human Genetics, 2013, 21, 736-742.	1.4	35
24	STAG1mutations cause a novel cohesinopathy characterised by unspecific syndromic intellectual disability. Journal of Medical Genetics, 2017, 54, 479-488.	1.5	35
25	De novo truncating variants in the intronless IRF2BPL are responsible for developmental epileptic encephalopathy. Genetics in Medicine, 2019, 21, 1008-1014.	1.1	34
26	Extracellular HSP110 skews macrophage polarization in colorectal cancer. OncoImmunology, 2016, 5, e1170264.	2.1	33
27	FR901228, an inhibitor of histone deacetylases, increases the cellular responsiveness to IL-6 type cytokines by enhancing the expression of receptor proteins. Oncogene, 2002, 21, 6264-6277.	2.6	32
28	Vps13b is required for acrosome biogenesis through functions in Golgi dynamic and membrane trafficking. Cellular and Molecular Life Sciences, 2020, 77, 511-529.	2.4	32
29	A GWAS in Latin Americans identifies novel face shape loci, implicating VPS13B and a Denisovan introgressed region in facial variation. Science Advances, 2021, 7, .	4.7	32
30	Differentiation of osteoblasts from mouse embryonic stem cells without generation of embryoid body. In Vitro Cellular and Developmental Biology - Animal, 2007, 43, 21-24.	0.7	30
31	Insulin response dysregulation explains abnormal fat storage and increased risk of diabetes mellitus type 2 in Cohen Syndrome. Human Molecular Genetics, 2015, 24, 6603-6613.	1.4	26
32	Long term oncostatin M treatment induces an osteocyte-like differentiation on osteosarcoma and calvaria cells. Bone, 2009, 44, 830-839.	1.4	25
33	Glycosaminoglycans inhibit the adherence and the spreading of osteoclasts and their precursors: Role in osteoclastogenesis and bone resorption. European Journal of Cell Biology, 2011, 90, 49-57.	1.6	23
34	Autosomal recessive <scp>IFT57</scp> hypomorphic mutation cause ciliary transport defect in unclassified oralâ€“facialâ€“digital syndrome with short stature and brachymesophalangia. Clinical Genetics, 2016, 90, 509-517.	1.0	20
35	Mutations in the Immunoglobulin-like Domain of gp190, the Leukemia Inhibitory Factor (LIF) Receptor, Increase or Decrease Its Affinity for LIF. Journal of Biological Chemistry, 2003, 278, 16253-16261.	1.6	18
36	Homozygous FIBP nonsense variant responsible of syndromic overgrowth, with overgrowth, macrocephaly, retinal coloboma and learning disabilities. Clinical Genetics, 2016, 89, e1-4.	1.0	18

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37	Heterozygous deletion of the LRFN2 gene is associated with working memory deficits. European Journal of Human Genetics, 2016, 24, 911-918.	1.4	18
38	Report of the first patient with a homozygous OTUD7A variant responsible for epileptic encephalopathy and related proteasome dysfunction. Clinical Genetics, 2020, 97, 567-575.	1.0	18
39	Conditioned media from mouse osteosarcoma cells promote MC3T3â€E1 cell proliferation using JAKs and PI3â€K/Akt signal crosstalk. Cancer Science, 2008, 99, 2170-2176.	1.7	15
40	Serpin B1 defect and increased apoptosis of neutrophils in Cohen syndrome neutropenia. Journal of Molecular Medicine, 2019, 97, 633-645.	1.7	15
41	Soluble Mannose 6-Phosphate/Insulin-Like Growth Factor II (IGF-II) Receptor Inhibits Interleukin-6-Type Cytokine-Dependent Proliferation by Neutralization of IGF-II. Endocrinology, 2003, 144, 5381-5389.	1.4	14
42	Demonstration of reverse fatty acid transport from rat cardiomyocytes. Journal of Lipid Research, 2004, 45, 1992-1999.	2.0	14
43	Congenital neutropenia with retinopathy, a new phenotype without intellectual deficiency or obesity secondary to VPS13B mutations. American Journal of Medical Genetics, Part A, 2014, 164, 522-527.	0.7	13
44	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	1.5	11
45	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	1.1	9
46	A constitutive BCL2 down-regulation aggravates the phenotype of PKD1-mutant-induced polycystic kidney disease. Human Molecular Genetics, 2017, 26, 4680-4688.	1.4	8
47	Identification of Agonistic and Antagonistic Antibodies against gp190, the Leukemia Inhibitory Factor Receptor, Reveals Distinct Roles for Its Two Cytokine-binding Domains. Journal of Biological Chemistry, 2001, 276, 47975-47981.	1.6	7
48	Search for a gene responsible for Floatingâ€Harbor syndrome on chromosome 12q15q21.1. American Journal of Medical Genetics, Part A, 2012, 158A, 333-339.	0.7	7
49	Excess of de novo variants in genes involved in chromatin remodelling in patients with marfanoid habitus and intellectual disability. Journal of Medical Genetics, 2020, 57, 466-474.	1.5	7
50	Cystoid maculopathy is a frequent feature of Cohen syndrome-associated retinopathy. Scientific Reports, 2021, 11, 16412.	1.6	7
51	Delineation of the 3p14.1p13 microdeletion associated with syndromic distal limb contractures. American Journal of Medical Genetics, Part A, 2014, 164, 3027-3034.	0.7	6
52	GLI3 is rarely implicated in OFD syndromes with midline abnormalities. Human Mutation, 2011, 32, 1332-1333.	1.1	5
53	Neutralization of HSF1 in cells from PIK3CA-related overgrowth spectrum patients blocks abnormal proliferation. Biochemical and Biophysical Research Communications, 2020, 530, 520-526.	1.0	5
54	Cohen Syndrome-Associated Cataract Is Explained by VPS13B Functions in Lens Homeostasis and Is Modified by Additional Genetic Factors. , 2020, 61, 18.		3

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55	Independence of hyperleptinemia-induced fat disappearance from thyroid hormone. Biochemical and Biophysical Research Communications, 2004, 323, 49-51.	1.0	2
56	Periodontal disorders in a cohort of patients with Cohen syndrome. Special Care in Dentistry, 2021, 41, 118-124.	0.4	1