Carine Le Goff

List of Publications by Year
in descending order

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33 papers	1,684 citations	331670 21 h-index	395702 33 g-index
35 all docs	35 docs citations	35 times ranked	2510 citing authors

#	Article	IF	Citations
1	ADAMTSL2 mutations in geleophysic dysplasia demonstrate a role for ADAMTS-like proteins in TGF-Î² bioavailability regulation. Nature Genetics, 2008, 40, 1119-1123.	21.4	211
2	Mutations in the TGFÎ² Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromicric and Geleophysic Dysplasias. American Journal of Human Genetics, 2011, 89, 7-14.	6.2	199
3	Mutations at a single codon in Mad homology 2 domain of SMAD4 cause Myhre syndrome. Nature Genetics, 2012, 44, 85-88.	21.4	125
4	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. American Journal of Human Genetics, 2012, 90, 740-745.	6.2	115
5	Regulation of procollagen amino-propeptide processing during mouse embryogenesis by specialization of homologous ADAMTS proteases: insights on collagen biosynthesis and dermatosparaxis. Development (Cambridge), 2006, 133, 1587-1596.	2.5	94
6	The ADAMTS(L) family and human genetic disorders. Human Molecular Genetics, 2011, 20, R163-R167.	2.9	93
7	Adamts9 is widely expressed during mouse embryo development. Gene Expression Patterns, 2005, 5, 609-617.	0.8	87
8	Identification of CANT1 Mutations in Desbuquois Dysplasia. American Journal of Human Genetics, 2009, 85, 706-710.	6.2	81
9	Adamts5, the gene encoding a proteoglycan-degrading metalloprotease, is expressed by specific cell lineages during mouse embryonic development and in adult tissues. Gene Expression Patterns, 2009, 9, 314-323.	0.8	77
10	From tall to short: The role of TGFÎ² signaling in growth and its disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 145-153.	1.6	74
11	ADAMTS-like 2 (ADAMTSL2) is a secreted glycoprotein that is widely expressed during mouse embryogenesis and is regulated during skeletal myogenesisâ˜†. Matrix Biology, 2007, 26, 431-441.	3.6	50
12	Mutations inLTBP3cause acromicric dysplasia and geleophysic dysplasia. Journal of Medical Genetics, 2016, 53, 457-464.	3.2	50
13	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	2.5	47
14	Positive role of cell wall anchored proteinase PrtP in adhesion of lactococci. BMC Microbiology, 2007, 7, 36.	3.3	45
15	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	3.2	45
16	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	2.8	38
17	Heterozygous Mutations in MAP3K7 , Encoding TGF-Î²-Activated Kinase 1, Cause Cardiospondylocarpofacial Syndrome. American Journal of Human Genetics, 2016, 99, 407-413.	6.2	33
18	Genetic and molecular aspects of acromelic dysplasia. Pediatric Endocrinology Reviews, 2009, 6, 418-23.	1.2	29

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19	Not All Floating-Harbor Syndrome Cases are Due to Mutations in Exon 34 ofSRCAP. Human Mutation, 2013, 34, 88-92.	2.5	28
20	Impairment of chondrogenesis and microfibrillar network in Adamtsl2 deficiency. FASEB Journal, 2019, 33, 2707-2718.	0.5	26
21	Pathogenic variants in THSD4, encoding the ADAMTS-like 6 protein, predispose to inherited thoracic aortic aneurysm. Genetics in Medicine, 2021, 23, 111-122.	2.4	25
22	In vitro readthrough of termination codons by gentamycin in the StÃ¼veâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2010, 18, 130-132.	2.8	19
23	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	2.8	18
24	Hypoalphalipoproteinemia andBRAF^{V600E}Mutation Are Major Predictors of Aortic Infiltration in the Erdheim-Chester Disease. Arteriosclerosis, Thrombosis, and Vascular Biology, 2018, 38, 1913-1925.	2.4	15
25	Unsuspected somatic mosaicism for FBN1 gene contributes to Marfan syndrome. Genetics in Medicine, 2021, 23, 865-871.	2.4	14
26	Novel Mutations in Geleophysic Dysplasia Type 1. Pediatric and Developmental Pathology, 2014, 17, 209-216.	1.0	11
27	Orthopedics management of acromicric dysplasia: Follow up of nine patients. American Journal of Medical Genetics, Part A, 2014, 164, 331-337.	1.2	10
28	A new mutational hotspot in the SKI gene in the context of MFS/TAA molecular diagnosis. Human Genetics, 2020, 139, 461-472.	3.8	8
29	Chondrodysplasias and TGFÎ² signaling. BoneKEy Reports, 2015, 4, 642.	2.7	6
30	ADAMTS Proteins and Vascular Remodeling in Aortic Aneurysms. Biomolecules, 2022, 12, 12.	4.0	6
31	The critical role of the TB5 domain of fibrillin-1 in endochondral ossification. Human Molecular Genetics, 2022, 31, 3777-3788.	2.9	3
32	Metalloproteinases and their inhibitors in the pathophysiology of heritable connective tissue disorders: current evidence. Metalloproteinases in Medicine, 2016, , 1.	1.0	1
33	ADAMTS10, ADAMTS17, and FBN1: The Weill-Marchesani Syndrome. , 2016, , 1335-1337.		0