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	ADAMTS Proteins and Vascular Remodeling in Aortic Aneurysms. Biomolecules, 2022, 12, 12.	4.0	6
2	The critical role of the TB5 domain of fibrillin-1 in endochondral ossification. Human Molecular Genetics, 2022, 31, 3777-3788.	2.9	3
3	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
4	Unsuspected somatic mosaicism for FBN1 gene contributes to Marfan syndrome. Genetics in Medicine, 2021, 23, 865-871.	2.4	14
5	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
6	Impairment of chondrogenesis and microfibrillar network in Adamtsl2 deficiency. FASEB Journal, 2019, 33, 2707-2718.	0.5	26
7	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
8	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
9	Metalloproteinases and their inhibitors in the pathophysiology of heritable connective tissue disorders: current evidence. Metalloproteinases in Medicine, 2016, , 1.	1.0	1
10	Mutations inLTBP3cause acromicric dysplasia and geleophysic dysplasia. Journal of Medical Genetics, 2016, 53, 457-464.	3.2	50
11	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
12	ADAMTS10, ADAMTS17, and FBN1: The Weill-Marchesani Syndrome. , 2016, , 1335-1337.		0
13	Chondrodysplasias and TGFÎ² signaling. BoneKEy Reports, 2015, 4, 642.	2.7	6
14	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	2.8	38
15	Orthopedics management of acromicric dysplasia: Follow up of nine patients. American Journal of Medical Genetics, Part A, 2014, 164, 331-337.	1.2	10
16	Novel Mutations in Geleophysic Dysplasia Type 1. Pediatric and Developmental Pathology, 2014, 17, 209-216.	1.0	11
17	Not All Floating-Harbor Syndrome Cases are Due to Mutations in Exon 34 ofSRCAP. Human Mutation, 2013, 34, 88-92.	2.5	28
18	Mutations at a single codon in Mad homology 2 domain of SMAD4 cause Myhre syndrome. Nature Genetics, 2012, 44, 85-88.	21.4	125

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19	Further delineation of CANT1 phenotypic spectrum and demonstration of its role in proteoglycan synthesis. Human Mutation, 2012, 33, 1261-1266.	2.5	47
20	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
21	Exome Sequencing Identifies PDE4D Mutations as Another Cause of Acrodysostosis. American Journal of Human Genetics, 2012, 90, 740-745.	6.2	115
22	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
23	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
24	The ADAMTS(L) family and human genetic disorders. Human Molecular Genetics, 2011, 20, R163-R167.	2.9	93
25	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
26	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
27	Identification of CANT1 Mutations in Desbuquois Dysplasia. American Journal of Human Genetics, 2009, 85, 706-710.	6.2	81
28	Genetic and molecular aspects of acromelic dysplasia. Pediatric Endocrinology Reviews, 2009, 6, 418-23.	1.2	29
29	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
30	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
31	Positive role of cell wall anchored proteinase PrtP in adhesion of lactococci. BMC Microbiology, 2007, 7, 36.	3.3	45
32	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
33	Adamts9 is widely expressed during mouse embryo development. Gene Expression Patterns, 2005, 5, 609-617.	0.8	87