

Celine V Cluzeau

List of Publications by Year in descending order

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Version: 2024-02-01

12
papers

1,571
citations

840776

11
h-index

1199594

12
g-index

12
all docs

12
docs citations

12
times ranked

2404
citing authors

#	ARTICLE	IF	CITATIONS
1	Interleukin-36 Receptor Antagonist Deficiency and Generalized Pustular Psoriasis. <i>New England Journal of Medicine</i> , 2011, 365, 620-628.	27.0	836
2	Only four genes (EDA1, EDAR, EDARADD, and WNT10A) account for 90% of hypohidrotic/anhidrotic ectodermal dysplasia cases. <i>Human Mutation</i> , 2011, 32, 70-72.	2.5	240
3	Microarray expression analysis and identification of serum biomarkers for Niemann-Pick disease, type C1. <i>Human Molecular Genetics</i> , 2012, 21, 3632-3646.	2.9	84
4	Autosomal dominant anhidrotic ectodermal dysplasias at the EDARADD locus. <i>Human Mutation</i> , 2007, 28, 703-709.	2.5	80
5	Human and mouse neuroinflammation markers in Niemann-Pick disease, type C1. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 83-92.	3.6	71
6	Quantitative Proteomic Analysis of Niemann-Pick Disease, Type C1 Cerebellum Identifies Protein Biomarkers and Provides Pathological Insight. <i>PLoS ONE</i> , 2012, 7, e47845.	2.5	59
7	Father-to-daughter transmission of Cornelia de Lange syndrome caused by a mutation in the 5' untranslated region of the NIPBL Gene. <i>Human Mutation</i> , 2006, 27, 731-735.	2.5	58
8	Modeling Smith-Lemli-Opitz syndrome with induced pluripotent stem cells reveals a causal role for Wnt/ β -catenin defects in neuronal cholesterol synthesis phenotypes. <i>Nature Medicine</i> , 2016, 22, 388-396.	30.7	46
9	Modeling Niemann-Pick disease type C1 in zebrafish: a robust platform for <i>in vivo</i> screening of candidate therapeutic compounds. <i>DMM Disease Models and Mechanisms</i> , 2018, 11, .	2.4	38
10	Mutations in EDARADD account for a small proportion of hypohidrotic ectodermal dysplasia cases. <i>British Journal of Dermatology</i> , 2010, 162, 1044-1048.	1.5	27
11	Unusual presentation of a severe autosomal recessive anhidrotic ectodermal dysplasia with a novel mutation in the EDAR gene. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2657-2662.	1.2	22
12	Gastrointestinal Tract Pathology in a BALB/c Niemann-Pick Disease Type C1 Null Mouse Model. <i>Digestive Diseases and Sciences</i> , 2018, 63, 870-880.	2.3	10