

Mireille Claustres

List of Publications by Year in descending order

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36
papers

1,993
citations

304743

22
h-index

345221

36
g-index

37
all docs

37
docs citations

37
times ranked

2587
citing authors

#	ARTICLE	IF	CITATIONS
1	Reclassifying inconclusive diagnosis after newborn screening for cystic fibrosis. Moving forward. <i>Journal of Cystic Fibrosis</i> , 2022, 21, 448-455.	0.7	9
2	The Study of a 231 French Patient Cohort Significantly Extends the Mutational Spectrum of the Two Major Usher Genes MYO7A and USH2A. <i>International Journal of Molecular Sciences</i> , 2021, 22, 13294.	4.1	5
3	The CYSMA web server: An example of integrative tool for in silico analysis of missense variants identified in Mendelian disorders. <i>Human Mutation</i> , 2020, 41, 375-386.	2.5	6
4	TCTEX1D1 is a genetic modifier of disease progression in Duchenne muscular dystrophy. <i>European Journal of Human Genetics</i> , 2020, 28, 815-825.	2.8	36
5	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. <i>Frontiers in Genetics</i> , 2020, 11, 605.	2.3	9
6	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	6.2	51
7	Current and future molecular approaches in the diagnosis of cystic fibrosis. <i>Expert Review of Respiratory Medicine</i> , 2018, 12, 415-426.	2.5	10
8	Thirteen years' experience of 893 PGD cycles for monogenic disorders in a publicly funded, nationally regulated regional hospital service. <i>Reproductive BioMedicine Online</i> , 2018, 36, 154-163.	2.4	9
9	A Reliable Targeted Next-Generation Sequencing Strategy for Diagnosis of Myopathies and Muscular Dystrophies, Especially for the Giant Titin and Nebulin Genes. <i>Journal of Molecular Diagnostics</i> , 2018, 20, 533-549.	2.8	28
10	The HDAC inhibitor SAHA does not rescue CFTR membrane expression in Cystic Fibrosis. <i>International Journal of Biochemistry and Cell Biology</i> , 2017, 88, 124-132.	2.8	13
11	<i>CFTR</i>-France, a national relational patient database for sharing genetic and phenotypic data associated with rare<i>CFTR</i>variants. <i>Human Mutation</i> , 2017, 38, 1297-1315.	2.5	62
12	Germline mosaicism is a pitfall in PGD for X-linked disorders. Single sperm typing detects very low frequency paternal gonadal mosaicism in a case of recurrent chondrodysplasia punctata misattributed to a maternal origin. <i>Prenatal Diagnosis</i> , 2017, 37, 201-205.	2.3	8
13	Implementation of a Reliable Next-Generation Sequencing Strategy for Molecular Diagnosis of Dystrophinopathies. <i>Journal of Molecular Diagnostics</i> , 2016, 18, 731-740.	2.8	24
14	Association Study of Exon Variants in the NF- κ B and TGF β 2 Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. <i>American Journal of Human Genetics</i> , 2016, 99, 1163-1171.	6.2	71
15	Small-scale high-throughput sequencing-based identification of new therapeutic tools in cystic fibrosis. <i>Genetics in Medicine</i> , 2015, 17, 796-806.	2.4	31
16	Should diffuse bronchiectasis still be considered a CFTR-related disorder?. <i>Journal of Cystic Fibrosis</i> , 2015, 14, 646-653.	0.7	20
17	Comprehensive CFTR gene analysis of the French cystic fibrosis screened newborn cohort: implications for diagnosis, genetic counseling, and mutation-specific therapy. <i>Genetics in Medicine</i> , 2015, 17, 108-116.	2.4	37
18	A Classification Model Relative to Splicing for Variants of Unknown Clinical Significance: Application to the<i>CFTR</i>Gene. <i>Human Mutation</i> , 2013, 34, 774-784.	2.5	23

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19	Identification of a novel duplication CFTRdup2 and functional impact of large rearrangements identified in the CFTR gene. <i>Gene</i> , 2012, 500, 194-198.	2.2	9
20	Mutation (variation) databases and registries: a rationale for coordination of efforts. <i>Nature Reviews Genetics</i> , 2011, 12, 881-881.	16.3	11
21	Four-Year Follow-up of Diagnostic Service in USH1 Patients. , 2011, 52, 4063.		47
22	UMD-CFTR: A database dedicated to CF and CFTR-related disorders. <i>Human Mutation</i> , 2010, 31, 1011-1019.	2.5	23
23	Capturing all disease-causing mutations for clinical and research use: Toward an effortless system for the Human Variome Project. <i>Genetics in Medicine</i> , 2009, 11, 843-849.	2.4	42
24	UMD-predictor, a new prediction tool for nucleotide substitution pathogenicity-application to four genes: <i>FBN1</i> , <i>FBN2</i> , <i>TGFBR1</i> , and <i>TGFBR2</i> . <i>Human Mutation</i> , 2009, 30, 952-959.	2.5	80
25	Genotype-phenotype analysis in 2,405 patients with a dystrophinopathy using the UMD-DMD database: a model of nationwide knowledgebase. <i>Human Mutation</i> , 2009, 30, 934-945.	2.5	309
26	A novel double deletion underscores the importance of characterizing end points of the CFTR large rearrangements. <i>European Journal of Human Genetics</i> , 2009, 17, 1683-1687.	2.8	10
27	WGA Allows the Molecular Characterization of a Novel Large CFTR Rearrangement in a Black South African Cystic Fibrosis Patient. <i>Journal of Molecular Diagnostics</i> , 2008, 10, 544-548.	2.8	8
28	Large genomic rearrangements in the CFTR gene contribute to CBAVD. <i>BMC Medical Genetics</i> , 2007, 8, 22.	2.1	42
29	A T3 allele in the CFTR gene exacerbates exon 9 skipping in vas deferens and epididymal cell lines and is associated with Congenital Bilateral Absence of Vas Deferens (CBAVD). <i>Human Mutation</i> , 2005, 25, 72-81.	2.5	44
30	UMD (Universal Mutation Database): 2005 update. <i>Human Mutation</i> , 2005, 26, 184-191.	2.5	101
31	Molecular pathology of the CFTR locus in male infertility. <i>Reproductive BioMedicine Online</i> , 2005, 10, 14-41.	2.4	108
32	Are p.I148T, p.R74W and p.D1270N cystic fibrosis causing mutations ?. <i>BMC Medical Genetics</i> , 2004, 5, 19.	2.1	47
33	Variation in a Repeat Sequence Determines Whether a Common Variant of the Cystic Fibrosis Transmembrane Conductance Regulator Gene Is Pathogenic or Benign. <i>American Journal of Human Genetics</i> , 2004, 74, 176-179.	6.2	227
34	Time for a Unified System of Mutation Description and Reporting: A Review of Locus-Specific Mutation Databases. <i>Genome Research</i> , 2002, 12, 680-688.	5.5	120
35	Spectrum of CFTR mutations in cystic fibrosis and in congenital absence of the vas deferens in France. <i>Human Mutation</i> , 2000, 16, 143-156.	2.5	240
36	Four adult patients with the missense mutation L206W and a mild cystic fibrosis phenotype. <i>Human Genetics</i> , 1995, 96, 717-720.	3.8	14