Mireille Claustres

List of Publications by Year
in descending order

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36	1,993	304743	345221
papers	citations	22 h-index	36 g-index

37	37	37	2587
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genotype-phenotype analysis in 2,405 patients with a dystrophinopathy using the UMD-DMD database: a model of nationwide knowledgebase. Human Mutation, 2009, 30, 934-945.	2.5	309
2	Spectrum of CFTR mutations in cystic fibrosis and in congenital absence of the vas deferens in France. Human Mutation, 2000, 16, 143-156.	2.5	240
3	Variation in a Repeat Sequence Determines Whether a Common Variant of the Cystic Fibrosis Transmembrane Conductance Regulator Gene Is Pathogenic or Benign. American Journal of Human Genetics, 2004, 74, 176-179.	6.2	227
4	Time for a Unified System of Mutation Description and Reporting: A Review of Locus-Specific Mutation Databases. Genome Research, 2002, 12, 680-688.	5.5	120
5	Molecular pathology of the CFTR locus in male infertility. Reproductive BioMedicine Online, 2005, 10, 14-41.	2.4	108
6	UMD (Universal Mutation Database): 2005 update. Human Mutation, 2005, 26, 184-191.	2.5	101
7	UMD-predictor, a new prediction tool for nucleotide substitution pathogenicity-application to four genes:FBN1,FBN2,TGFBR1, andTGFBR2. Human Mutation, 2009, 30, 952-959.	2.5	80
8	Association Study of Exon Variants in the NF-Î²B and TGFÎ² Pathways Identifies CD40 as a Modifier of Duchenne Muscular Dystrophy. American Journal of Human Genetics, 2016, 99, 1163-1171.	6.2	71
9	CFTR-France, a national relational patient database for sharing genetic and phenotypic data associated with rareCFTRvariants. Human Mutation, 2017, 38, 1297-1315.	2.5	62
10	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
11	Are p.I148T, p.R74W and p.D1270N cystic fibrosis causing mutations ?. BMC Medical Genetics, 2004, 5, 19.	2.1	47
12	Four-Year Follow-up of Diagnostic Service in USH1 Patients. , 2011, 52, 4063.		47
13	A T3 allele in theCFTR gene exacerbates exon 9 skipping in vas deferens and epididymal cell lines and is associated with Congenital Bilateral Absence of Vas Deferens (CBAVD). Human Mutation, 2005, 25, 72-81.	2.5	44
14	Large genomic rearrangements in the CFTRgene contribute to CBAVD. BMC Medical Genetics, 2007, 8, 22.	2.1	42
15	Capturing all disease-causing mutations for clinical and research use: Toward an effortless system for the Human Variome Project. Genetics in Medicine, 2009, 11, 843-849.	2.4	42
16	Comprehensive CFTR gene analysis of the French cystic fibrosis screened newborn cohort: implications for diagnosis, genetic counseling, and mutation-specific therapy. Genetics in Medicine, 2015, 17, 108-116.	2.4	37
17	TCTEX1D1 is a genetic modifier of disease progression in Duchenne muscular dystrophy. European Journal of Human Genetics, 2020, 28, 815-825.	2.8	36
18	Small-scale high-throughput sequencingâ€“based identification of new therapeutic tools in cystic fibrosis. Genetics in Medicine, 2015, 17, 796-806.	2.4	31

#	Article	IF	Citations
19	A Reliable Targeted Next-Generation Sequencing Strategy for Diagnosis of Myopathies and Muscular Dystrophies, Especially for the Giant Titin and Nebulin Genes. Journal of Molecular Diagnostics, 2018, 20, 533-549.	2.8	28
20	Implementation of a Reliable Next-Generation Sequencing Strategy for Molecular Diagnosis of Dystrophinopathies. Journal of Molecular Diagnostics, 2016, 18, 731-740.	2.8	24
21	UMD-CFTR: A database dedicated to CF and CFTR-related disorders. Human Mutation, 2010, 31, 1011-1019.	2.5	23
22	A Classification Model Relative to Splicing for Variants of Unknown Clinical Significance: Application to theCFTRGene. Human Mutation, 2013, 34, 774-784.	2.5	23
23	Should diffuse bronchiectasis still be considered a CFTR-related disorder?. Journal of Cystic Fibrosis, 2015, 14, 646-653.	0.7	20
24	Four adult patients with the missense mutation L206W and a mild cystic fibrosis phenotype. Human Genetics, 1995, 96, 717-720.	3.8	14
25	The HDAC inhibitor SAHA does not rescue CFTR membrane expression in Cystic Fibrosis. International Journal of Biochemistry and Cell Biology, 2017, 88, 124-132.	2.8	13
26	Mutation (variation) databases and registries: a rationale for coordination of efforts. Nature Reviews Genetics, 2011, 12, 881-881.	16.3	11
27	A novel double deletion underscores the importance of characterizing end points of the CFTR large rearrangements. European Journal of Human Genetics, 2009, 17, 1683-1687.	2.8	10
28	Current and future molecular approaches in the diagnosis of cystic fibrosis. Expert Review of Respiratory Medicine, 2018, 12, 415-426.	2.5	10
29	Identification of a novel duplication CFTRdup2 and functional impact of large rearrangements identified in the CFTR gene. Gene, 2012, 500, 194-198.	2.2	9
30	Thirteen years' experience of 893 PGD cycles for monogenic disorders in a publicly funded, nationally regulated regional hospital service. Reproductive BioMedicine Online, 2018, 36, 154-163.	2.4	9
31	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	2.3	9
32	Reclassifying inconclusive diagnosis after newborn screening for cystic fibrosis. Moving forward. Journal of Cystic Fibrosis, 2022, 21, 448-455.	0.7	9
33	WGA Allows the Molecular Characterization of a Novel Large CFTR Rearrangement in a Black South African Cystic Fibrosis Patient. Journal of Molecular Diagnostics, 2008, 10, 544-548.	2.8	8
34	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. Prenatal Diagnosis, 2017, 37, 201-205.	2.3	8
35	The CYSMA web server: An example of integrative tool for in silico analysis of missense variants identified in Mendelian disorders. Human Mutation, 2020, 41, 375-386.	2.5	6
36	The Study of a 231 French Patient Cohort Significantly Extends the Mutational Spectrum of the Two Major Usher Genes MYO7A and USH2A. International Journal of Molecular Sciences, 2021, 22, 13294.	4.1	5