Alix de Becdelievre

List of Publications by Year
in descending order

25 papers	609 citations	566801 15 h-index	610482 24 g-index
25 all docs	25 docs citations	25 times ranked	1017 citing authors

#	Article	IF	Citations
1	New syndromic form of benign hereditary chorea is associated with a deletion of TITF-1 and PAX-9 contiguous genes. Movement Disorders, 2006, 21, 2237-2240.	2.2	82
2	COMMD1-Mediated Ubiquitination Regulates CFTR Trafficking. PLoS ONE, 2011, 6, e18334.	1.1	56
3	Heterogeneity of lung disease associated with NK2 homeobox 1 mutations. Respiratory Medicine, 2017, 129, 16-23.	1.3	54
4	Correlation between PABPN1 genotype and disease severity in oculopharyngeal muscular dystrophy. Neurology, 2017, 88, 359-365.	1.5	53
5	Alternative Splicing at a NAGNAG Acceptor Site as a Novel Phenotype Modifier. PLoS Genetics, 2010, 6, e1001153.	1.5	49
6	Bethlem myopathy: long-term follow-up identifies COL6 mutations predicting severe clinical evolution. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 1337-1346.	0.9	34
7	Diaphragmatic dysfunction in Collagen VI myopathies. Neuromuscular Disorders, 2014, 24, 125-133.	0.3	32
8	CFTR p.Arg117His associated with CBAVD and other CFTR-related disorders. Journal of Medical Genetics, 2013, 50, 220-227.	1.5	31
9	Mosaicism for Dominant Collagen 6 Mutations as a Cause for Intrafamilial Phenotypic Variability. Human Mutation, 2015, 36, 48-56.	1.1	28
10	Slowly progressive spinocerebellar ataxia with extrapyramidal signs and mild cognitive impairment (SCA21). Cerebellum, 2008, 7, 179-183.	1.4	27
11	Combined Computational-Experimental Analyses ofCFTRExon Strength Uncover Predictability of Exon-Skipping Level. Human Mutation, 2013, 34, 873-881.	1.1	27
12	Relationship of nonâ€visualization of the fetal gallbladder and amniotic fluid digestive enzymes analysis to outcome. Prenatal Diagnosis, 2012, 32, 423-426.	1.1	23
13	A recurrent deep-intronic splicing CF mutation emphasizes the importance of mRNA studies in clinical practice. Journal of Cystic Fibrosis, 2011, 10, 479-482.	0.3	19
14	CFTRmutation combinations producing frequent complex alleles with different clinical and functional outcomes. Human Mutation, 2012, 33, 1557-1565.	1.1	19
15	Comprehensive description of CFTR genotypes and ultrasound patterns in 694 cases of fetal bowel anomalies: a revised strategy. Human Genetics, 2011, 129, 387-396.	1.8	17
16	Amniotic Fluid Digestive Enzyme Analysis Is Useful for Identifying CFTR Gene Mutations of Unclear Significance. Clinical Chemistry, 2009, 55, 2214-2217.	1.5	12
17	Alternative Splicing of In-Frame Exon Associated with Premature Termination Codons: Implications for Readthrough Therapies. Human Mutation, 2013, 34, 287-291.	1.1	11
18	Hereditary sensory autonomic neuropathy type II: Report of two novel mutations in the FAM134B gene. Journal of the Peripheral Nervous System, 2019, 24, 354-358.	1.4	9

#	Article	IF	Citations
19	Notable contribution of large CFTR gene rearrangements to the diagnosis of cystic fibrosis in fetuses with bowel anomalies. European Journal of Human Genetics, 2010, 18, 1166-1169.	1.4	7
20	COMMD1 modulates noxious inflammation in cystic fibrosis. International Journal of Biochemistry and Cell Biology, 2013, 45, 2402-2409.	1.2	6
21	The most frequent ABCA3 nonsense mutation -p.Tyr1515* (Y1515X) causing lethal neonatal respiratory failure in a term neonate. Annals of Thoracic Medicine, 2017, 12, 213.	0.7	5
22	Prenatal Ultrasound Suspicion of Cystic Fibrosis in a Multiethnic Population: Is Extensive CFTR Genotyping Needed?. Genes, 2021, 12, 670.	1.0	4
23	Authors' reply regarding â€œRelationship of nonâ€visualization of the fetal gallbladder and amniotic fluid digestive enzymes analysis to outcomeâ€. Prenatal Diagnosis, 2012, 32, 1121-1121.	1.1	2
24	Identification of a Novel 5â€² AlternativeCFTRmRNA Isoform in a Patient with Nasal Polyposis andCFTRMutations. Human Mutation, 2014, 35, 805-808.	1.1	1
25	SCN10A variants associated with congenital Harlequin Syndrome. British Journal of Dermatology, 2022, , .	1.4	1