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	SCN10A variants associated with congenital Harlequin Syndrome. British Journal of Dermatology, 2022, , .	1.4	1
2	Prenatal Ultrasound Suspicion of Cystic Fibrosis in a Multiethnic Population: Is Extensive CFTR Genotyping Needed?. Genes, 2021, 12, 670.	1.0	4
3	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
4	Heterogeneity of lung disease associated with NK2 homeobox 1 mutations. Respiratory Medicine, 2017, 129, 16-23.	1.3	54
5	Correlation between PABPN1 genotype and disease severity in oculopharyngeal muscular dystrophy. Neurology, 2017, 88, 359-365.	1.5	53
6	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
7	Mosaicism for Dominant Collagen 6 Mutations as a Cause for Intrafamilial Phenotypic Variability. Human Mutation, 2015, 36, 48-56.	1.1	28
8	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
9	Diaphragmatic dysfunction in Collagen VI myopathies. Neuromuscular Disorders, 2014, 24, 125-133.	0.3	32
10	Identification of a Novel 5â€² AlternativeCFTRmRNA Isoform in a Patient with Nasal Polyposis andCFTRMutations. Human Mutation, 2014, 35, 805-808.	1.1	1
11	CFTR p.Arg117His associated with CBAVD and other CFTR-related disorders. Journal of Medical Genetics, 2013, 50, 220-227.	1.5	31
12	Alternative Splicing of In-Frame Exon Associated with Premature Termination Codons: Implications for Readthrough Therapies. Human Mutation, 2013, 34, 287-291.	1.1	11
13	COMMD1 modulates noxious inflammation in cystic fibrosis. International Journal of Biochemistry and Cell Biology, 2013, 45, 2402-2409.	1.2	6
14	Combined Computational-Experimental Analyses ofCFTRExon Strength Uncover Predictability of Exon-Skipping Level. Human Mutation, 2013, 34, 873-881.	1.1	27
15	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
16	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
17	CFTRmutation combinations producing frequent complex alleles with different clinical and functional outcomes. Human Mutation, 2012, 33, 1557-1565.	1.1	19
18	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

#	Article	IF	Citations
19	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
20	COMMD1-Mediated Ubiquitination Regulates CFTR Trafficking. PLoS ONE, 2011, 6, e18334.	1.1	56
21	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
22	Alternative Splicing at a NAGNAG Acceptor Site as a Novel Phenotype Modifier. PLoS Genetics, 2010, 6, e1001153.	1.5	49
23	Amniotic Fluid Digestive Enzyme Analysis Is Useful for Identifying CFTR Gene Mutations of Unclear Significance. Clinical Chemistry, 2009, 55, 2214-2217.	1.5	12
24	Slowly progressive spinocerebellar ataxia with extrapyramidal signs and mild cognitive impairment (SCA21). Cerebellum, 2008, 7, 179-183.	1.4	27
25	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