

# Teresa Casals

## List of Publications by Year in descending order

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54  
papers

3,847  
citations

236612

25  
h-index

197535

49  
g-index

55  
all docs

55  
docs citations

55  
times ranked

2964  
citing authors

#	ARTICLE	IF	CITATIONS
1	Assessing the residual CFTR gene expression in human nasal epithelium cells bearing CFTR splicing mutations causing cystic fibrosis. <i>European Journal of Human Genetics</i> , 2014, 22, 784-791.	1.4	24
2	GUSB and ATP2B4 are suitable reference genes for CFTR gene expression data normalization in nasal epithelium cells. <i>Journal of Cystic Fibrosis</i> , 2012, 11, 398-404.	0.3	4
3	Optimizing strategies for CFTR molecular testing. <i>Journal of Cystic Fibrosis</i> , 2011, 10, 143-144.	0.3	0
4	Common CFTR haplotypes and susceptibility to chronic pancreatitis and congenital bilateral absence of the vas deferens. <i>Human Mutation</i> , 2011, 32, 912-920.	1.1	22
5	Independent Contribution of Common CFTR Variants to Chronic Pancreatitis. <i>Pancreas</i> , 2010, 39, 209-215.	0.5	30
6	Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. <i>Human Mutation</i> , 2010, 31, 421-428.	1.1	31
7	CFTR Rearrangements in Spanish Cystic Fibrosis Patients: First New Duplication (35kb) Characterised in the Mediterranean Countries. <i>Annals of Human Genetics</i> , 2010, 74, 463-469.	0.3	10
8	Association of cystic fibrosis genetic modifiers with congenital bilateral absence of the vas deferens. <i>Fertility and Sterility</i> , 2010, 94, 2122-2127.	0.5	20
9	The p.Arg258Gly Mutation in Intracellular Loop 2 of CFTR is Associated with CFTR-Related Disorders. <i>Genetic Testing and Molecular Biomarkers</i> , 2009, 13, 765-768.	0.3	3
10	Best practice guidelines for molecular genetic diagnosis of cystic fibrosis and CFTR-related disorders – updated European recommendations. <i>European Journal of Human Genetics</i> , 2009, 17, 51-65.	1.4	207
11	CFTR mutations in cystic fibrosis patients from Murcia region (southeastern Spain): implications for genetic testing. <i>Clinical Genetics</i> , 2009, 76, 577-579.	1.0	5
12	Common mutations in cuban cystic fibrosis patients. <i>Journal of Cystic Fibrosis</i> , 2009, 8, 47-49.	0.3	14
13	N-terminal CFTR missense variants severely affect the behavior of the CFTR chloride channel. <i>Human Mutation</i> , 2008, 29, 738-749.	1.1	25
14	Consensus on the use and interpretation of cystic fibrosis mutation analysis in clinical practice. <i>Journal of Cystic Fibrosis</i> , 2008, 7, 179-196.	0.3	493
15	Cystic fibrosis in a southern Brazilian population: characteristics of 90% of the alleles. <i>Clinical Genetics</i> , 2007, 72, 218-223.	1.0	13
16	Spectrum of Mutations in the CFTR Gene in Cystic Fibrosis Patients of Spanish Ancestry. <i>Annals of Human Genetics</i> , 2007, 71, 194-201.	0.3	53
17	Mutational spectrum of cystic fibrosis patients from Córdoba province and its zone of influence: Implications of molecular diagnosis in Argentina. <i>Molecular Genetics and Metabolism</i> , 2006, 87, 370-375.	0.5	8
18	Gross genomic rearrangements involving deletions in the CFTR gene: characterization of six new events from a large cohort of hitherto unidentified cystic fibrosis chromosomes and meta-analysis of the underlying mechanisms. <i>European Journal of Human Genetics</i> , 2006, 14, 567-576.	1.4	77

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19	Molecular evaluation of CFTR sequence variants in male infertility of testicular origin. Journal of Developmental and Physical Disabilities, 2005, 28, 284-290.	3.6	26
20	Spectrum of mutations in CFTR in Finland: 18 years follow-up study and identification of two novel mutations. Journal of Cystic Fibrosis, 2005, 4, 233-237.	0.3	23
21	Genotype-phenotype correlation for pulmonary function in cystic fibrosis. Thorax, 2005, 60, 558-563.	2.7	81
22	Multimutational Analysis of Eleven Cystic Fibrosis Mutations Common in the Mediterranean Areas. Clinical Chemistry, 2004, 50, 2155-2157.	1.5	5
23	Bronchiectasis in adult patients: an expression of heterozygosity for CFTR gene mutations?. Clinical Genetics, 2004, 65, 490-495.	1.0	81
24	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.	2.6	227
25	Different CFTR Mutational Spectrum in Alcoholic and Idiopathic Chronic Pancreatitis?. Pancreas, 2004, 28, 374-379.	0.5	34
26	Suitability of oligonucleotide-mediated cystic fibrosis gene repair in airway epithelial cells. Journal of Gene Medicine, 2003, 5, 625-639.	1.4	5
27	Low incidence of PRSS1 and SPINK1 mutations in patients with chronic pancreatitis in Spain. Gastroenterology, 2003, 124, A584.	0.6	0
28	New type of disease causing mutations: the example of the composite exonic regulatory elements of splicing in CFTR exon 12. Human Molecular Genetics, 2003, 12, 1111-1120.	1.4	171
29	Can a Place of Origin of the Main Cystic Fibrosis Mutations Be Identified?. American Journal of Human Genetics, 2002, 70, 257-264.	2.6	37
30	Non-viral vector-mediated uptake, distribution, and stability of chimeraplasts in human airway epithelial cells. Journal of Gene Medicine, 2002, 4, 308-322.	1.4	15
31	Quantitative assessment of chimeraplast stability in biological fluids by polyacrylamide gel electrophoresis and laser-assisted fluorescence analysis. Pharmaceutical Research, 2002, 19, 914-918.	1.7	1
32	ATB0/SLC1A5 gene. Fine localisation and exclusion of association with the intestinal phenotype of cystic fibrosis. European Journal of Human Genetics, 2001, 9, 860-866.	1.4	10
33	Adenosine Triphosphate-Binding Cassette Superfamily Transporter Gene Expression in Severe Male Infertility1. Biology of Reproduction, 2001, 65, 394-400.	1.2	20
34	Heterogeneity for mutations in the CFTR gene and clinical correlations in patients with congenital absence of the vas deferens. Human Reproduction, 2000, 15, 1476-1483.	0.4	131
35	Detection of a cystic fibrosis modifier locus for meconium ileus on human chromosome 19q13. Nature Genetics, 1999, 22, 128-129.	9.4	216
36	Missense mutations in the cystic fibrosis gene in adult patients with asthma. , 1999, 14, 510-519.		51

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37	Allele Frequencies in a Worldwide Survey of a CA Repeat in the First Intron of the CFTR Gene. <i>Human Heredity</i> , 1999, 49, 15-20.	0.4	13
38	High heterogeneity for cystic fibrosis in Spanish families: 75 mutations account for 90% of chromosomes. <i>Human Genetics</i> , 1997, 101, 365-370.	1.8	94
39	SSCP analysis: A blind sensitivity trial. , 1997, 10, 65-70.		60
40	PRENATAL DIAGNOSIS OF CYSTIC FIBROSIS IN A HIGHLY HETEROGENEOUS POPULATION. , 1996, 16, 215-222.		12
41	Mutations in the Cystic Fibrosis Gene in Patients with Congenital Absence of the Vas Deferens. <i>New England Journal of Medicine</i> , 1995, 332, 1475-1480.	13.9	959
42	Analysis of the CFTR gene confirms the high genetic heterogeneity of the Spanish population: 43 mutations account for only 78% of CF chromosomes. <i>Human Genetics</i> , 1994, 93, 447-51.	1.8	65
43	Analysis of the CFTR gene in the Spanish population: SSCP-screening for 60 known mutations and identification of four new mutations (Q30X, A120T, 1812-1 Gâ†’A, and 3667del14). <i>Human Mutation</i> , 1994, 3, 223-230.	1.1	31
44	Molecular and clinical analyses of cystic fibrosis in the South of Spain. <i>Clinical Genetics</i> , 1994, 46, 287-290.	1.0	5
45	Cystic fibrosis in Spain: high frequency of mutation G542X in the Mediterranean coastal area. <i>Human Genetics</i> , 1993, 91, 66-70.	1.8	35
46	Novel alleles, hemizyosity and deletions at an Alu-repeat within the neurofibromatosis type 1 (NF1) gene. <i>Human Molecular Genetics</i> , 1993, 2, 725-730.	1.4	47
47	Microsatellite haplotypes for cystic fibrosis: mutation frameworks and evolutionary tracers. <i>Human Molecular Genetics</i> , 1993, 2, 1015-1022.	1.4	97
48	Haplotype analysis to determine the position of a mutation among closely linked DNA markers. <i>Human Molecular Genetics</i> , 1993, 2, 1007-1014.	1.4	19
49	Identification of a new missense mutation (P205S) in the first transmembrane domain of the CFTR gene associated with a mild cystic fibrosis phenotype. <i>Human Molecular Genetics</i> , 1993, 2, 1741-1742.	1.4	18
50	Identification of a 31-bp insertion (3860ins31) in exon 20 of the cysticfibrosis (CFTR) gene. <i>Human Molecular Genetics</i> , 1993, 2, 1317-1318.	1.4	7
51	Uniparental inheritance of microsatellite alleles of the cystic fibrosis gene (CFTR): identification of a 50 kilobase deletion. <i>Human Molecular Genetics</i> , 1993, 2, 677-681.	1.4	32
52	Identification of a frameshift mutation (1609delCA) in exon 10 of the CFTR gene in seven Spanish cystic fibrosis patients. <i>Human Mutation</i> , 1992, 1, 75-76.	1.1	12
53	CAGT Microsatellite alleles within the cystic fibrosis transmembrane conductance regulator (CFTR) gene are not generated by unequal crossingover. <i>Genomics</i> , 1991, 10, 692-698.	1.3	129
54	Recurrence of a nonsense mutation in the NF1 gene causing classical neurofibromatosis type 1. <i>Human Genetics</i> , 1991, 88, 185-8.	1.8	39