# Teresa Casals

## List of Publications by Year in descending order

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| 54 | 3,847 | 236612 | 197535 |
|---|---|---|---|
| papers | citations | 25 h-index | 49 g-index |

| 55 | 55 | 55 | 2964 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Mutations in the Cystic Fibrosis Gene in Patients with Congenital Absence of the Vas Deferens. New England Journal of Medicine, 1995, 332, 1475-1480. | 13.9 | 959 |
| 2 | Consensus on the use and interpretation of cystic fibrosis mutation analysis in clinical practice. Journal of Cystic Fibrosis, 2008, 7, 179-196. | 0.3 | 493 |
| 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. | 2.6 | 227 |
| 4 | Detection of a cystic fibrosis modifier locus for meconium ileus on human chromosome 19q13. Nature Genetics, 1999, 22, 128-129. | 9.4 | 216 |
| 5 | Best practice guidelines for molecular genetic diagnosis of cystic fibrosis and CFTR-related disorders â€“ updated European recommendations. European Journal of Human Genetics, 2009, 17, 51-65. | 1.4 | 207 |
| 6 | 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 |
| 7 | 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 |
| 8 | CAGT Microsatellite alleles within the cystic fibrosis transmembrane conductance regulator (CFTR) gene are not generated by unequal crossingover. Genomics, 1991, 10, 692-698. | 1.3 | 129 |
| 9 | Microsatellite haplotypes for cystic fibrosis: mutation frameworks and evolutionary tracers. Human Molecular Genetics, 1993, 2, 1015-1022. | 1.4 | 97 |
| 10 | High heterogeneity for cystic fibrosis in Spanish families: 75 mutations account for 90% of chromosomes. Human Genetics, 1997, 101, 365-370. | 1.8 | 94 |
| 11 | Bronchiectasis in adult patients: an expression of heterozygosity for CFTR gene mutations?. Clinical Genetics, 2004, 65, 490-495. | 1.0 | 81 |
| 12 | Genotype-phenotype correlation for pulmonary function in cystic fibrosis. Thorax, 2005, 60, 558-563. | 2.7 | 81 |
| 13 | 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. European Journal of Human Genetics, 2006, 14, 567-576. | 1.4 | 77 |
| 14 | Analysis of the CFTR gene confirms the high genetic heterogeneity of the Spanish population: 43 mutations account for only 78% of CF chromosomes. Human Genetics, 1994, 93, 447-51. | 1.8 | 65 |
| 15 | SSCP analysis: A blind sensitivity trial. , 1997, 10, 65-70. | | 60 |
| 16 | Spectrum of Mutations in the CFTR Gene in Cystic Fibrosis Patients of Spanish Ancestry. Annals of Human Genetics, 2007, 71, 194-201. | 0.3 | 53 |
| 17 | Missense mutations in the cystic fibrosis gene in adult patients with asthma. , 1999, 14, 510-519. | | 51 |
| 18 | Novel alleles, hemizygosity and deletions at an Alu-repeat within the neurofibromatosis type 1 (NF1) gene. Human Molecular Genetics, 1993, 2, 725-730. | 1.4 | 47 |

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|---|---|---|---|
| 19 | Recurrence of a nonsense mutation in the NF1 gene causing classical neurofibromatosis type 1. Human Genetics, 1991, 88, 185-8. | 1.8 | 39 |
| 20 | 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 |
| 21 | Cystic fibrosis in Spain: high frequency of mutation G542X in the Mediterranean coastal area. Human Genetics, 1993, 91, 66-70. | 1.8 | 35 |
| 22 | Different CFTR Mutational Spectrum in Alcoholic and Idiopathic Chronic Pancreatitis?. Pancreas, 2004, 28, 374-379. | 0.5 | 34 |
| 23 | Uniparental inheritance of microsatellite alleles of the cystic fibrosis gene (CFTR): identification of a 50 kilobase deletion. Human Molecular Genetics, 1993, 2, 677-681. | 1.4 | 32 |
| 24 | 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 3667de14). Human Mutation, 1994, 3, 223-230. | 1.1 | 31 |
| 25 | Complete ascertainment of intragenic copy number mutations (CNMs) in the CFTR gene and its implications for CNM formation at other autosomal loci. Human Mutation, 2010, 31, 421-428. | 1.1 | 31 |
| 26 | Independent Contribution of Common CFTR Variants to Chronic Pancreatitis. Pancreas, 2010, 39, 209-215. | 0.5 | 30 |
| 27 | 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 |
| 28 | N-terminal CFTR missense variants severely affect the behavior of the CFTR chloride channel. Human Mutation, 2008, 29, 738-749. | 1.1 | 25 |
| 29 | Assessing the residual CFTR gene expression in human nasal epithelium cells bearing CFTR splicing mutations causing cystic fibrosis. European Journal of Human Genetics, 2014, 22, 784-791. | 1.4 | 24 |
| 30 | 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 |
| 31 | Common CFTR haplotypes and susceptibility to chronic pancreatitis and congenital bilateral absence of the vas deferens. Human Mutation, 2011, 32, 912-920. | 1.1 | 22 |
| 32 | Adenosine Triphosphate-Binding Cassette Superfamily Transporter Gene Expression in Severe Male Infertility1. Biology of Reproduction, 2001, 65, 394-400. | 1.2 | 20 |
| 33 | Association of cystic fibrosis genetic modifiers with congenital bilateral absence of the vas deferens. Fertility and Sterility, 2010, 94, 2122-2127. | 0.5 | 20 |
| 34 | Haplotype analysis to determine the position of a mutation among closely linked DNA markers. Human Molecular Genetics, 1993, 2, 1007-1014. | 1.4 | 19 |
| 35 | Identification of a new missense mutation (P205S) in the first transmembrane domain of the CFTR gene associated with a mild cystic fibrosis phenotype. Human Molecular Genetics, 1993, 2, 1741-1742. | 1.4 | 18 |
| 36 | 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 |

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|---|---|---|---|
| 37 | Common mutations in cuban cystic fibrosis patients. Journal of Cystic Fibrosis, 2009, 8, 47-49. | 0.3 | 14 |
| 38 | Allele Frequencies in a Worldwide Survey of a CA Repeat in the First Intron of the CFTR Gene. Human Heredity, 1999, 49, 15-20. | 0.4 | 13 |
| 39 | Cystic fibrosis in a southern Brazilian population: characteristics of 90% of the alleles. Clinical Genetics, 2007, 72, 218-223. | 1.0 | 13 |
| 40 | Identification of a frameshift mutation (1609delCA) in exon 10 of the CFTR gene in seven Spanish cystic fibrosis patients. Human Mutation, 1992, 1, 75-76. | 1.1 | 12 |
| 41 | PRENATAL DIAGNOSIS OF CYSTIC FIBROSIS IN A HIGHLY HETEROGENEOUS POPULATION. , 1996, 16, 215-222. | | 12 |
| 42 | 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 |
| 43 | CFTR Rearrangements in Spanish Cystic Fibrosis Patients: First New Duplication (35kb) Characterised in the Mediterranean Countries. Annals of Human Genetics, 2010, 74, 463-469. | 0.3 | 10 |
| 44 | Mutational spectrum of cystic fibrosis patients from CÃ³rdoba province and its zone of influence: Implications of molecular diagnosis in Argentina. Molecular Genetics and Metabolism, 2006, 87, 370-375. | 0.5 | 8 |
| 45 | Identification of a 31-bp insertion (3860ins31) in exon 20 of the cysticfibrosis (CFTR) gene. Human Molecular Genetics, 1993, 2, 1317-1318. | 1.4 | 7 |
| 46 | Suitability of oligonucleotide-mediated cystic fibrosis gene repair in airway epithelial cells. Journal of Gene Medicine, 2003, 5, 625-639. | 1.4 | 5 |
| 47 | Multimutational Analysis of Eleven Cystic Fibrosis Mutations Common in the Mediterranean Areas. Clinical Chemistry, 2004, 50, 2155-2157. | 1.5 | 5 |
| 48 | Molecular and clinical analyses of cystic fibrosis in the South of Spain. Clinical Genetics, 1994, 46, 287-290. | 1.0 | 5 |
| 49 | CFTR mutations in cystic fibrosis patients from Murcia region (southeastern Spain): implications for genetic testing. Clinical Genetics, 2009, 76, 577-579. | 1.0 | 5 |
| 50 | GUSB and ATP2B4 are suitable reference genes for CFTR gene expression data normalization in nasal epithelium cells. Journal of Cystic Fibrosis, 2012, 11, 398-404. | 0.3 | 4 |
| 51 | The p.Arg258Gly Mutation in Intracellular Loop 2 of CFTR is Associated with CFTR-Related Disorders. Genetic Testing and Molecular Biomarkers, 2009, 13, 765-768. | 0.3 | 3 |
| 52 | 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 |
| 53 | Low incidence of PRSS1 and SPINK1 mutations in patients with chronic pancreatitis in Spain. Gastroenterology, 2003, 124, A584. | 0.6 | 0 |
| 54 | Optimizing strategies for CFTR molecular testing. Journal of Cystic Fibrosis, 2011, 10, 143-144. | 0.3 | 0 |