Resource Summary Report

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Cystic Fibrosis Mutation Database

RRID:SCR_000685
Type: Tool

Proper Citation

Cystic Fibrosis Mutation Database (RRID:SCR_000685)

Resource Information

URL: http://www.genet.sickkids.on.ca/cftr/

Proper Citation: Cystic Fibrosis Mutation Database (RRID:SCR_000685)

Description: Collection of mutations in CFTR gene for international cystic fibrosis genetics research community. Provides up to date information about individual mutations in CFTR gene. All known CFTR mutations and sequence variants have been converted to standard nomenclature recommended by Human Genome Variation Society. On line process for submission of new mutations has been added.While they continue to ensure quality of data, they urge international community to give them feedback and suggestions. Clinical information in this database relates only to details of discovery of specific mutations. As part of 2010 upgrade, CFTR1 joined new project called CFTR2 - Clinical and Functional TRanslation of CFTR. Links to CFTR2 for many mutations in CFTR1 will provide up-to-date summaries of genotype-phenotype information from patient registries around the world.

Abbreviations: CFTR1, CFMDB

Resource Type: service resource, storage service resource, data or information resource, data repository, database

Keywords: Gene, genetic, amino acid, clinical, cystic fibrosis, mutation, phenotype, genotype-phenotype, genotype, dna sequence, mouse, sequence, genetic variation, polymorphism, translation, function, sequence variation, metadata standard, cftr2, FASEB list

Related Condition: Cystic fibrosis

Funding:

Availability: Free, Freely available

Resource Name: Cystic Fibrosis Mutation Database

Resource ID: SCR_000685

Alternate IDs: nif-0000-21105

Record Creation Time: 20220129T080203+0000

Record Last Update: 20250426T055423+0000

Ratings and Alerts

No rating or validation information has been found for Cystic Fibrosis Mutation Database.

No alerts have been found for Cystic Fibrosis Mutation Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 42 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

Martin ER, et al. (2020) In vivo crystals reveal critical features of the interaction between cystic fibrosis transmembrane conductance regulator (CFTR) and the PDZ2 domain of Na+/H+ exchange cofactor NHERF1. The Journal of biological chemistry, 295(14), 4464.

Laselva O, et al. (2018) Molecular Mechanism of Action of Trimethylangelicin Derivatives as CFTR Modulators. Frontiers in pharmacology, 9, 719.

Carlile GW, et al. (2018) A novel triple combination of pharmacological chaperones improves F508del-CFTR correction. Scientific reports, 8(1), 11404.

Hinzpeter A, et al. (2017) The importance of functional tests to assess the effect of a new CFTR variant when genotype-phenotype correlation is not possible. Clinical case reports, 5(5), 658.

Villalona S, et al. (2017) R248G cystic fibrosis transmembrane conductance regulator mutation in three siblings presenting with recurrent acute pancreatitis and reproductive issues: a case series. Journal of medical case reports, 11(1), 42.

Molinski SV, et al. (2017) Orkambi® and amplifier co-therapy improves function from a rare CFTR mutation in gene-edited cells and patient tissue. EMBO molecular medicine, 9(9),

1224.

Yeh HI, et al. (2017) A common mechanism for CFTR potentiators. The Journal of general physiology, 149(12), 1105.

Suzuki S, et al. (2016) TALENs Facilitate Single-step Seamless SDF Correction of F508del CFTR in Airway Epithelial Submucosal Gland Cell-derived CF-iPSCs. Molecular therapy. Nucleic acids, 5(1), e273.

Mohseni M, et al. (2016) Novel CFTR Mutations in Two Iranian Families with Severe Cystic Fibrosis. Iranian biomedical journal, 20(4), 201.

Tian X, et al. (2016) p.G970D is the most frequent CFTR mutation in Chinese patients with cystic fibrosis. Human genome variation, 3, 15063.

Farhat R, et al. (2015) N1303K (c.3909C>G) mutation and splicing: implication of its c.[744-33GATT(6); 869+11C>T] complex allele in CFTR exon 7 aberrant splicing. BioMed research international, 2015, 138103.

Bepari KK, et al. (2015) Allele frequency for Cystic fibrosis in Indians vis-a/-vis global populations. Bioinformation, 11(7), 348.

Griesenbach U, et al. (2015) Recent advances in understanding and managing cystic fibrosis transmembrane conductance regulator dysfunction. F1000prime reports, 7, 64.

Neocleous V, et al. (2014) Apparent Homozygosity of p.Phe508del in CFTR due to a Large Gene Deletion of Exons 4-11. Case reports in genetics, 2014, 613863.

Puzik A, et al. (2014) Lethal course of meconium ileus in preterm twins revealing a novel cystic fibrosis mutation (p.Cys524Tyr). BMC pediatrics, 14, 13.

Arora K, et al. (2014) Stabilizing rescued surface-localized ?f508 CFTR by potentiation of its interaction with Na(+)/H(+) exchanger regulatory factor 1. Biochemistry, 53(25), 4169.

Rauniyar N, et al. (2014) Quantitative proteomic profiling reveals differentially regulated proteins in cystic fibrosis cells. Journal of proteome research, 13(11), 4668.

Zi?tkiewicz E, et al. (2014) CFTR mutations spectrum and the efficiency of molecular diagnostics in Polish cystic fibrosis patients. PloS one, 9(2), e89094.

Stokes ME, et al. (2014) Towards personalized agriculture: what chemical genomics can bring to plant biotechnology. Frontiers in plant science, 5, 344.

Dal'Maso VB, et al. (2013) Diagnostic contribution of molecular analysis of the cystic fibrosis transmembrane conductance regulator gene in patients suspected of having mild or atypical cystic fibrosis. Jornal brasileiro de pneumologia : publicacao oficial da Sociedade Brasileira de Pneumologia e Tisilogia, 39(2), 181.