



All

Images

Videos

翻译成中文

关闭取词

11,700 Results

Any time ▾

## Mechanisms of CFTR Functional Variants That Impair ...

[journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1004376](https://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1004376) ▾

CFTR sev and CFTR m-v variants are associated with **recurrent acute pancreatitis** and chronic **pancreatitis** –. Recently, we reported that the variant CFTR R75Q, which was previously classified as benign, is associated with familial and sporadic chronic **pancreatitis**, either with another CFTR variant (recessive) or with the serine protease ...

Published in: PLOS Genetics · 2014

Authors: Jessica Larusch · Jinsei Jung · Ignacio J General · Michele D Lewis · Hyun Woo Park

Affiliation: University of Pittsburgh · Yonsei University · Mayo Clinic · University of Michigan · Brig...

About: HEK 293 cells · Male infertility · **Cystic fibrosis transmembrane conductance regul...**

## Type of CFTR Mutation Determines Risk of Pancreatitis in ...

[https://www.gastrojournal.org/article/S0016-5085\(10\)01455-1/fulltext](https://www.gastrojournal.org/article/S0016-5085(10)01455-1/fulltext)

Overall, the **diagnosis of pancreatitis** was first documented at a mean (SD) age of 24.2 (11.2) years. More specifically, the mean (SD) age at first documented episode of **pancreatitis** among patients with acute, recurrent acute, and **chronic pancreatitis** was 24.4 (9.9), 25.2 (11.4), and 20.7 (12.5) years, respectively ( $P = .5$ ). The onset of pancreatitis preceded the diagnosis of CF in 15 patients (24%).

Cited by: 168

Author: Chee Y. Ooi, Ruslan Dorfman, Marco Cip...

Publish Year: 2011

## Pancreatitis in Cystic Fibrosis and CFTR-Related Disorder ...

<https://www.intechopen.com/.../pancreatitis-in-cystic-fibrosis-and-cftr-related-disorder> ▾

Using this terminology, **patients** with **recurrent-acute** or chronic **pancreatitis** due to CFTR dysfunction, may receive a diagnosis of CF **pancreatitis** or CFTR-related **pancreatitis**. Individuals with a CFTR-related disorder are recognised to be at risk for developing CF disease (Farrell et al., 2008).

Cited by: 3

Author: Michael J. Coffey, Chee Y. Ooi

Publish Year: 2012

## Mechanisms of CFTR Functional ... - PubMed Central (PMC)

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4102440>

Jul 17, 2014 · CFTR sev and CFTR m-v variants are associated with **recurrent acute pancreatitis** and chronic **pancreatitis** –. Recently, we reported that the variant CFTR R75Q, which was previously classified as benign, is associated with familial and sporadic chronic **pancreatitis**, either with another CFTR variant (recessive) or with the serine protease ...

Cited by: 104

Author: Jessica LaRusch, Jinsei Jung, Ignacio J. ...

Publish Year: 2014

**Name of Journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 49215

**Manuscript Type:** CASE REPORT

**Cystic fibrosis transmembrane conductance regulator functional evaluations in a G542X+/- IVS8Tn:T7/9 patient with acute recurrent pancreatitis**

Caldrer S *et al.* CFTR functional assays in pancreatitis

Sara Caldrer, Gabriella Bergamini, Angela Sandri, Silvia Vercellone, Luca Rodella, Angelo Cerofolini, Francesco Tomba, Filippo Catalano, Luca Frulloni, Mario Buffelli, Gloria Tridello, Hugo de Jonge, Baroukh Maurice Assael, Claudio Sorio, Paola Melotti

### Match Overview

1	<b>Crossref</b> 29 words A D Mandić, E Bennek, J Verdier, K Zhang et al. "c-Jun N-terminal kinase 2 promotes enterocyte survival and goblet c ...	1%
2	<b>Crossref</b> 24 words Chee Y. Ooi, Tanja Gonska, Peter R. Durie, Steven D. Fre... man. "Genetic Testing in Pancreatitis", Gastroenterology, 2	1%
3	<b>Internet</b> 22 words crawled on 21-Jan-2014 <a href="http://mukoviscidoz.org">mukoviscidoz.org</a>	1%
4	<b>Internet</b> 21 words crawled on 06-Apr-2018 <a href="http://erj.ersjournals.com">erj.ersjournals.com</a>	1%
5	<b>Internet</b> 13 words crawled on 07-Oct-2013 <a href="http://www.jmedicalcasereports.com">www.jmedicalcasereports.com</a>	<1%
6	<b>Internet</b> 12 words crawled on 15-Jul-2016 <a href="http://abcmutations.hegelab.org">abcmutations.hegelab.org</a>	<1%
7	<b>Crossref</b> 11 words J. F. Dekkers, G. Berkers, E. Kruisselbrink, A. Vonk et al. "C haracterizing responses to CFTR-modulating drugs usin ...	<1%
8	<b>Crossref</b> 10 words "Poster Session Abstracts", Pediatric Pulmonology, 09/2010	<1%





5,220 Results

Any time ▼

## Relation between Mutations of the Cystic Fibrosis Gene and ...

<https://www.nejm.org/doi/full/10.1056/NEJM199809033391002>

Sep 03, 1998 · The most common inherited disease of the exocrine pancreas is cystic fibrosis. 5-8 In cystic fibrosis, mutations of the cystic fibrosis transmembrane conductance regulator (CF TR) gene lead ...

Cited by: 1010

Author: Jonathan A. Cohn, Kenneth J. Friedman, ...

Publish Year: 1998

## Cystic fibrosis transmembrane conductance regulator (CFTR ...

[https://www.researchgate.net/publication/225094360\\_Cystic\\_fibrosis\\_transmembrane...](https://www.researchgate.net/publication/225094360_Cystic_fibrosis_transmembrane...)

Diabetes is a common and important complication of cystic fibrosis, an autosomal recessive genetic disease due to mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene.

## CFTR cystic fibrosis transmembrane conductance regulator ...

<https://www.ncbi.nlm.nih.gov/gene/1080>

Cystic fibrosis transmembrane conductance regulator interacts with multiple immunoglobulin domains of filamin A; Did not find common mutations in the CFTR gene in patients with pancreatic divisum. Nasal potential difference was measured in patients with diffuse bronchiectasis, a normal sweat test, classified by CFTR mutation.

## Molecular consequences of cystic fibrosis transmembrane ...

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1773762>

Identification of the cystic fibrosis gene 1– 3 led to the elucidation of its protein product, the cystic fibrosis transmembrane conductance regulator (CFTR), a cAMP regulated chloride channel. 4 Mutations in the CFTR gene result in absent or defective CFTR chloride channel function at the apical surface of epithelial cells.

Cited by: 182

Author: Ahmed N, Corey M, Forstner G, Zielenski...

Publish Year: 2003

## Mutations of the cystic fibrosis gene, but not cationic ...

<https://www.sciencedirect.com/science/article/pii/S0002927000010200>

CFTR = cystic fibrosis transmembrane conductance regulator; CF = cystic fibrosis. Family study of CFTR In the compound heterozygous patient S K we investigated the parents and the sister of this





国内版

国际版

Cystic fibrosis transmembrane conductance regulator function



All

Images

Videos

关闭取词

5,230 Results

Any time ▾

## Relation between Mutations of the Cystic Fibrosis Gene and ...

<https://www.nejm.org/doi/full/10.1056/NEJM199809033391002>

Sep 03, 1998 · The most common inherited disease of the exocrine pancreas is cystic fibrosis. 5-8 In cystic fibrosis, mutations of the cystic fibrosis transmembrane conductance regulator (CF TR) gene lead ...

Cited by: 1010

Author: Jonathan A. Cohn, Kenneth J. Friedman, ...

Publish Year: 1998

## Mechanisms of CFTR Functional Variants That Impair ...

<https://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1004376> ▾

The cystic fibrosis transmembrane conductance regulator (CFTR, GenBank Accession: AH006034.1) is an ATP-binding cassette (ABC) transporter-type protein localized to the apical plasma membrane of epithelial cells. It differs from other ABC transporters in that it acts as a regulated anion channel rather than a transporter. When the channel is ...

Cited by: 110

Author: Jessica LaRusch, Jinsei Jung, Ignacio J. ...

Publish Year: 2014

## Diagnosis of Cystic Fibrosis: Consensus Guidelines from ...

<https://www.sciencedirect.com/science/article/pii/S0022347616310484>

Cystic fibrosis (CF) is the most common life-threatening autosomal recessive disease in the US, affecting approximately 1 in 4000 newborns in the US, 1, 2, 3 and occurring at higher frequencies in some European countries. 4, 5 CF is a multisystem disorder caused by mutations in the gene for the CF transmembrane conductance regulator (CFTR), which encodes an ion channel protein, 6 with more ...

Cited by: 216

Author: Philip M. Farrell, Terry B. White, Clement...

Publish Year: 2017

## Novel personalized therapies for cystic fibrosis: treating ...

[onlinelibrary.wiley.com/doi/10.1111/joim.12314/full](https://onlinelibrary.wiley.com/doi/10.1111/joim.12314/full)

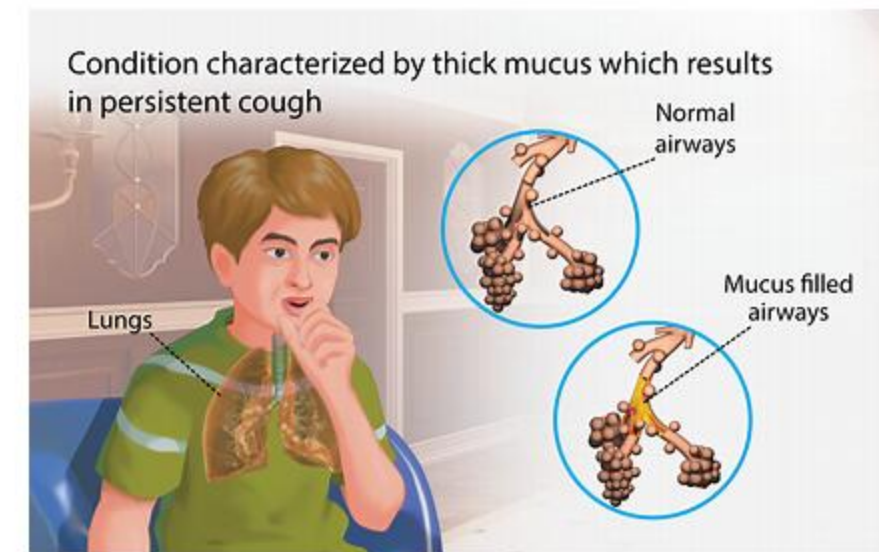
Jan 26, 2015 · The cystic fibrosis (CF) pathogenesis cascade in the lung. The mechanism of CF dysfunction starts with the primary CFTR gene defect and ultimate leads to severe lung deficiency. CFTR, cystic fibrosis transmembrane conductance regulator; ASL, airway surface liquid; ENaC, epithelial Na<sup>+</sup> channel.

Published in: Journal of Internal Medicine · 2015

Authors: Margarida D Amaral

## Cystic Fibrosis

Medical Condition



A genetic disorder, in which the lungs and the digestive system get clogged with mucus.

- Very rare (Fewer than 20,000 cases per year in US)
- Often requires lab test or imaging
- Treatments can help manage condition, no known cure
- Can be lifelong

Caused due to a genetic defect. Symptoms include difficulty in breathing, coughing and poor growth. Though incurable, medications can help in managing symptoms.

### Symptoms

CF affects cells that make mucus, sweat and digestive fluids. These fluids are thick in CF patients and cause the blockage of ducts throughout the body leading to a range of symptoms. The lungs and digestive system are the main areas that are affected. Symptoms may appear soon after birth or in early childhood. In rare cases, they are not manifested until adulthood.

- Persistent cough with phlegm
- Postnasal drip