5,670 Results Any time

Clinical and genetic characteristics of cystic fibrosis in ...

https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0968-2

Dec 17, 2018 · Ho YL, Wu WJ, Wong KS, Huang CB, Niu CK, Shyur SD, Huang LH, Wu PC. **Novel mutation** (I1023R) in two Taiwanese siblings of **cystic fibrosis**. J Pediatr Respir. 2007;5(3):152–8. Google Scholar; Cheng Y, Ning G, Song B, Guo YK, Li XS. A **Chinese** girl with **cystic fibrosis**: a case report identified by sweat and genetic tests. Chin **Med** J. 2012;125(4):719.

Author: Xiaobei Guo, Xiaobei Guo, Keqiang Li... **Publish Year:** 2018

Epidemiology and genetics of cystic fibrosis in Asia: In ...

onlinelibrary.wiley.com/doi/10.1111/resp.12656/full

The spectrum of the **cystic fibrosis transmembrane conductance regulator** (CFTR) variants in this population is quite heterogeneous. In total, 166 variants have been reported on approximately **3700 Asian CF** chromosomes. The frequency of **F508del** among Asians is low compared with Caucasians.

Published in: Respirology · 2015

Authors: Meenu Singh · Cristina Rebordosa · Juliane Bernholz · Neeraj Sharma

Affiliation: Novartis · Johns Hopkins University School of Medicine

About: Epidemiology · **Cystic fibrosis** transmembrane conductance regulator · **Cystic fibrosis**

p.G970D is the most frequent CFTR mutation in Chinese ...

nature.com › human genome variation

Introduction. One **mutation**, p.**F508del**, has been **found** to occur in ~70% of the **CFTR** alleles of Caucasian CF **patients**. 7 However, its frequency varies considerably among human populations. Apart from p.**F508del**, ~20 **mutations** occur with a frequency >0.1%, together accounting for ~15–20% of the CF alleles of Caucasians.

Cited by: 7 **Author:** Xinlun Tian, Yaping Liu, Jun Yang, Han W...

Publish Year: 2016

Liver Failure in a Chinese Cystic Fibrosis Child With ...

https://www.frontiersin.org/articles/10.3389/fped.2019.00036

Cystic fibrosis (CF) is a relatively rare disease in Asians with various clinical characteristics, including **CF-associated liver disease** (CFLD), which is a common early non-pulmonary **complication**.

Author: Haiyan Li, Li Lin, Xiaoguang Hu, Chan... **Publish Year:** 2019

Name of Journal: *World Journal of Clinical Cases*

Manuscript NO: 46809

Manuscript Type: CASE REPORT

c.753_754delAG, a novel *CFTR* mutation found in a Chinese patient with cystic fibrosis: A case report and review of literature

Yu-Qing Wang, Chuang-Li Hao, Wu-Jun Jiang, Yan-Hong Lu, Hui-Quan Sun, Chun-Yan Gao, Min Wu

Abstract

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c.753_754delAG, a novel CFTR mutation found in a Chinese patient with cystic fibr

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Clinical and genetic characteristics of cystic fibrosis in ...

https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0968-2

Dec 17, 2018 · An ethnicity-specific **CFTR** variant spectrum was also observed in **CF** patients of **Chinese** origin, with p.Gly970Asp as the most common **mutation** while p.Phe508del, the most common **pathogenic mutation in CF** patients of **Caucasian** origin, is rare, suggesting the necessity of a **Chinese-specific CFTR** variant screening panel. Besides, multiplex ligation-dependent probe amplification analysis should be routinely considered, especially for those with **unidentified mutations**.

Author: Xiaobei Guo, Xiaobei Guo, Keqiang Li... **Publish Year:** 2018

Detection of novel CFTR mutations in Taiwanese cystic ...

https://www.researchgate.net/publication/10649656_Detection_of_**novel_CFTR_mutations**_in...

This case report describes **a Chinese CF patient harboring a homozygous nonsense mutation** (**c.1657C>T, p**.R553X) who was failure to thrive and had intermittently diarrhea during ...

[PDF] Case Report Chinese data of the CFTR mutation: a report ...

ijcem.com/files/ijcem0065112.pdf

Abstract: Background and objective: **Cystic fibrosis** (CF) is a serious genetic disorder that is caused by **mutations in the cystic fibrosis transmembrane conductance regulator** (CFTR) gene. Little is known about the genetic information of the **CFTR mutation in the Chinese population**. The objective of this study is to report a new **CFTR mutation** in a

p.G970D is the most frequent CFTR mutation in Chinese ...

https://www.nature.com/articles/hgv201563

Jan 07, 2016 · **Cystic fibrosis** (**CF**) **in China** is most often caused by **a mutation** not **found** elsewhere in the world. **CF** is an inherited disease of the secretory glands that arises from **mutations** in a gene called **CFTR**.

Cited by: 10 **Author:** Xinlun Tian, Yaping Liu, Jun Yang, Han W...

Publish Year: 2016 **Author:** Xinlun Tian