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



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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 · 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](https://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](https://www.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, <sup>2</sup> a novel *CFTR* mutation found in a Chinese patient with  
cystic fibrosis: A case report and review of literature

Wang YQ *et al.* Novel *CFTR* mutation in a Chinese CF patient

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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## [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...](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](http://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