

Resource Summary Report

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CFTR2

RRID:SCR_019078

Type: Tool

Proper Citation

CFTR2 (RRID:SCR_019078)

Resource Information

URL: <https://cftr2.org/>

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Description: International initiative led by team of researchers and clinicians and supported by the US Cystic Fibrosis Foundation that seeks to provide complete, advanced and expert reviewed functional and clinical information on CFTR mutations. Provides information for patients, researchers, and general public about specific variants. For each variant or variant combination included in database, website will provide information about whether variant or variant combination is CF-causing, and information about sweat chloride, lung function, pancreatic status, and Pseudomonas infection rate in patients in CFTR2 database with this variant or variant combination.

Synonyms: Clinical and Functional TRAnslation of CFTR, cftr2, Clinical and Functional TRAnslation 2

Resource Type: data or information resource, database, disease-related portal, portal, topical portal

Keywords: CF gene, cystic fibrosis gene, functional testing, CFTR2 variant, data, CFTR mutations, clinical information, , FASEB list

Related Condition: cystic fibrosis

Availability: Free, Freely available

Resource Name: CFTR2

Resource ID: SCR_019078

License URLs: <https://cftr2.org/sites/default/files/Legal-Terms-Conditions-CFTR2.pdf>, <https://cftr2.org/sites/default/files/Privacy-Policy-CFTR2.pdf>

Record Creation Time: 20220129T080343+0000

Ratings and Alerts

No rating or validation information has been found for CFTR2.

No alerts have been found for CFTR2.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 174 mentions in open access literature.

Listed below are recent publications. The full list is available at [Kravitz](#) .

Jhangiani AR, et al. (2026) Profiling the CFTR Variant Selectivity and Off-Target Interactions of VX-121. bioRxiv : the preprint server for biology.

Yu M, et al. (2026) Cystic fibrosis risk variants confer protection against inflammatory bowel disease. Cell genomics, 6(2), 101071.

Lin JH, et al. (2026) When splicing is not all or none: GT>GC 5' splice-site variants as a model for intermediate effects and challenges in variant classification. HGG advances, 7(3), 100602.

Freyberg M, et al. (2026) Large variations in total and allele-specific transcript expression in a disease mutation-independent manner. Scientific reports, 16(1).

Vasilyeva T, et al. (2026) A Diagnostic Dilemma: Concurrent Diagnosis of Cystic Fibrosis and Definitive Kabuki Syndrome Type 1. International journal of molecular sciences, 27(5).

El Makhzen N, et al. (2026) CFTR gene variant detection in moroccan individuals via nanopore long-read sequencing. Frontiers in genetics, 17, 1769093.

Chen D, et al. (2026) Homozygous 896delT (c.764del) in Somali-American Siblings With Cystic Fibrosis. Case reports in pulmonology, 2026, 1148927.

Yuan P, et al. (2026) Spectrum and Classification of CFTR and ADGRG2 Variants in Chinese Patients With Isolated CAVD: A Large Cohort Study and Risk Assessment of CFTR Variant Carriage in Couples. Human mutation, 2026, 5588277.

Lopatina M, et al. (2026) Cellular Models and Functional Assays for Assessing CFTR Function: A Comprehensive Review. International journal of molecular sciences, 27(12).

Ko W, et al. (2025) ACE-tRNAs are a platform technology for suppressing nonsense mutations that cause cystic fibrosis. Nucleic acids research, 53(13).

Tedman A, et al. (2025) General trends in the calnexin-dependent expression and pharmacological rescue of clinical CFTR variants. *eLife*, 14.

Lu Y, et al. (2025) Cystic fibrosis-causing variants in Chinese patients with congenital absence of the vas deferens: a cohort and meta-analysis. *Asian journal of andrology*, 27(5), 611.

Kireeva TN, et al. (2025) Cystic fibrosis therapy: from symptoms to the cause of the disease. *Vavilovskii zhurnal genetiki i selektsii*, 29(2), 279.

Burgel PR, et al. (2025) Elexacaftor-tezacaftor-ivacaftor in people with cystic fibrosis harbouring two CFTR Class I variants: real-world data from the French compassionate programme. *EClinicalMedicine*, 88, 103476.

Pinnaro CT, et al. (2025) Unbalanced long-chain fatty acid beta-oxidation in newborns with cystic fibrosis and congenital hypothyroidism. *Molecular genetics and metabolism reports*, 42, 101182.

Yin X, et al. (2025) Pathogenic germline variants in Chinese pancreatic adenocarcinoma patients. *Nature communications*, 16(1), 2214.

Chandane P, et al. (2025) Clinical and genetic profiles of paediatric patients with cystic fibrosis from Western India. *Lung India : official organ of Indian Chest Society*, 42(2), 103.

Demchenko A, et al. (2025) Human Induced Lung Organoids: A Promising Tool for Cystic Fibrosis Drug Screening. *International journal of molecular sciences*, 26(2).

Nov-Klaiman T, et al. (2025) More of the same? Israel's expanded carrier screening for cystic fibrosis. *European journal of human genetics : EJHG*, 33(12), 1555.

Blevings PJ, et al. (2025) Mutation characterisation of the cystic fibrosis transmembrane conductance regulator (CFTR) gene in people with cystic fibrosis in Northern Ireland. *The Ulster medical journal*, 94(2), 64.