

# All-in-one NGS solution for cystic fibrosis diagnostics

Worldwide, 162 428 people are estimated to be living with cystic fibrosis. 105 352 cases of cystic fibrosis are diagnosed. 57 076 patients are undiagnosed.<sup>[1]</sup>

Cystic fibrosis (CF) is a multisystem disease with autosomal recessive inheritence, affecting the cystic fibrosis transmembrane conductance regulator (CFTR) gene. CF mainly affects the lungs and pancreas, but also the upper airways, liver, intestine, and reproductive organs. Currently, more than 2000 germline sequence variants of the CFTR gene have already been identified, though not all result in CF. <sup>[2] [3] [4]</sup>

According to the Cystic fibrosis foundation, a child born in 2019 is predicted to live 48 years or more, whereas in 1995–1999 patients were predicted to live 32 years. <sup>[5]</sup> Early recognition of cystic fibrosis and the associated initiation of treatment is an important factor in the prognosis of this disease <sup>[3]</sup>. The fastGEN technology provides a method of sensitive, specific and time-efficient examination of the mutational status of clinical markers using NGS technology. fastGEN



Guo J., Garratt A. and Hill A. Worldwide rates of diagnosis and effective treatment for cystic fibrosis. Journal of Cystic Fybrosis 2022; 21: 456-462.
Bareil C. and Bergougnoux A. CFTR gene variants, epidemiology and molecular pathology. Archives de Pédiatrie 2020; 27: eS8-eS12.

[3] Naehrig S., Chao Ch.M. and Naehrlich L. Cystic Fibrosis. Deutsches Ärzteblatt International 2017: 114: 564-574.

[4] Sosnay P. R., Raraigh K. S. and Gibson R. L. Molecular Genetics of Cystic Fibrosis Transmembrane Conductance Regulator: Genotype and Phenotype. Pediatric Clinics of North America 2016; 63: 585-598.
[5] Cystic Fibrosis Foundation. (2002) Cystic Fibrosis Foundation. United States. [Web Archive] Retrieved from the Library of Congress, https://www.loc.gov/item/lcwaN0000114/.

## 4 reasons why fastGEN worths attention

#### Unique

Simple, ultra-sensitive, specific and effective technology

#### Reliable

Significantly lower probability of mistakes, uniform coverage for different amplicons and different samples



Excellent speed (<30 min hands-on time)

#### Robust

Robustness achieved by using short amplicons obtained by a single polymerase chain reaction with special tagged hybrid primers

## You are only a few drops from the results

#### **Technical parameters**

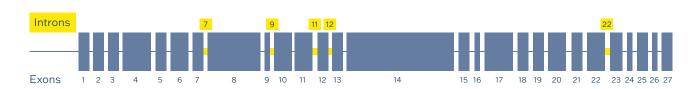
DNA Input The minimum input amount of DNA is just 10 ng. Suitable samples DNA from whole blood DNA from buccal swab DNA from Guthrie cards

#### Kit components

Ready-to-use Master Mixes with sample indexes Seq primers read1 and read2 Index seq primer

#### **Covered regions**

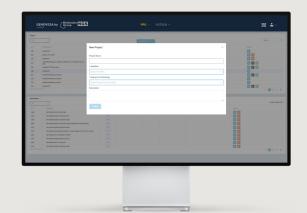
All the exomes including the parts of introns (see the diagram)



Cat. No.	Product Name	Index Set	Size	<b>Regulatory Status</b>
RDNGS0005	fastGEN CFTR 24-kit	A, B, C	24 samples	RUO
RDNGS0006A	fastGEN CFTR 8-kit	А	8 samples	RUO
RDNGS0006B	fastGEN CFTR 8-kit	В	8 samples	RUO
RDNGS0006C	fastGEN CFTR 8-kit	С	8 samples	RUO

## fastGEN modul in GENOVESA software for your service

- Advanced quality control of sequencing data
- Automatic notification of low-coverage regions for simple filtering of relevant variants
- Monthly update of annotation databases
- Customization
- Patient data and variants stored in an internal database
- One-click report



Take the opportunity to test the demo kit in your lab and contact our specialists today.

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DISTRIBUTOR:



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