

Hereditary variants profiling in cystic fibrosis



4bases CFTR panel is **an amplicon-based CE-IVD** solution for rapid and cost-effective identification of known and novel germline variations (CNVs, SNPs, indels, poly-T and TG repeats) in the **CFTR gene**.

It allows the identification of the most common, well-known causative mutations of the CFTR gene, new exonic and deep intronic variants, **through Next Generation Sequencing (NGS)**.

The kit is validated for the analysis of DNA extracted from blood (also from dried blood spot).

**4bases CFTR panel** allows for **precise**, **reliable**, and **effective results**, **speeding up** the activity of clinical reporting.

From DNA to sequencing in less than 1,5 days with less than 2 hours hands-on time.



From gDNA



**Enrichment PCR** 



**Indexing PCR** 



Final library



Sequencing



Final Data

#### Straightforward protocol

Tailor-made protocol for operator's time schedule, presence of many stopping points, reduced hands-on time

### Sequencing platform flexibility

Common overall protocol for different sequencing applications (Illumina, MGI, Oxford Nanopore Technologies, Ion Torrent, Element Biosciences)

#### Platform 4eVAR

Safe and secure data analysis via 4bases proprietary solution, with a continuous experienced customer support





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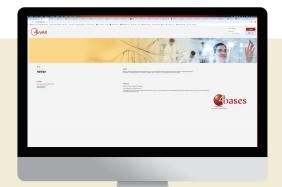
# **Specifications**

Sample type	DNA extracted from blood samples (also dried blood spot)
Input	Minimum of 10 ng DNA per sample
Panel size	35 kb
Gene content	CFTR coding regions (including of 25 bp proximal to the 3' and 5' end) and intronic variants
Variant called	CNVs, SNPs, indels, poly-T and TG repeats
Instrument Type	Element Biosciences, Illumina, Ion Torrent, MGI, Oxford Nanopore Technologies
Data Analysis	4eVAR

## Cod. kit

Product	Cod.	Tests number
CFTR panel	H1060-16	16
CFTR panel	H1060-96	96

# 4eVAR: Comprehensive output files for analysis and reporting



- Quality metrics
- Genotype
- Alignment
- Coverage
- CNVs status
- PolyGT-PolyT status
- Full variant table
- Customizable report

# **Data Analysis**

Product	Cod.	Tests number
4eVAR	A6060-H1060-16	16
4eVAR	A6060-H1060-96	96

# If you wish to learn more:





