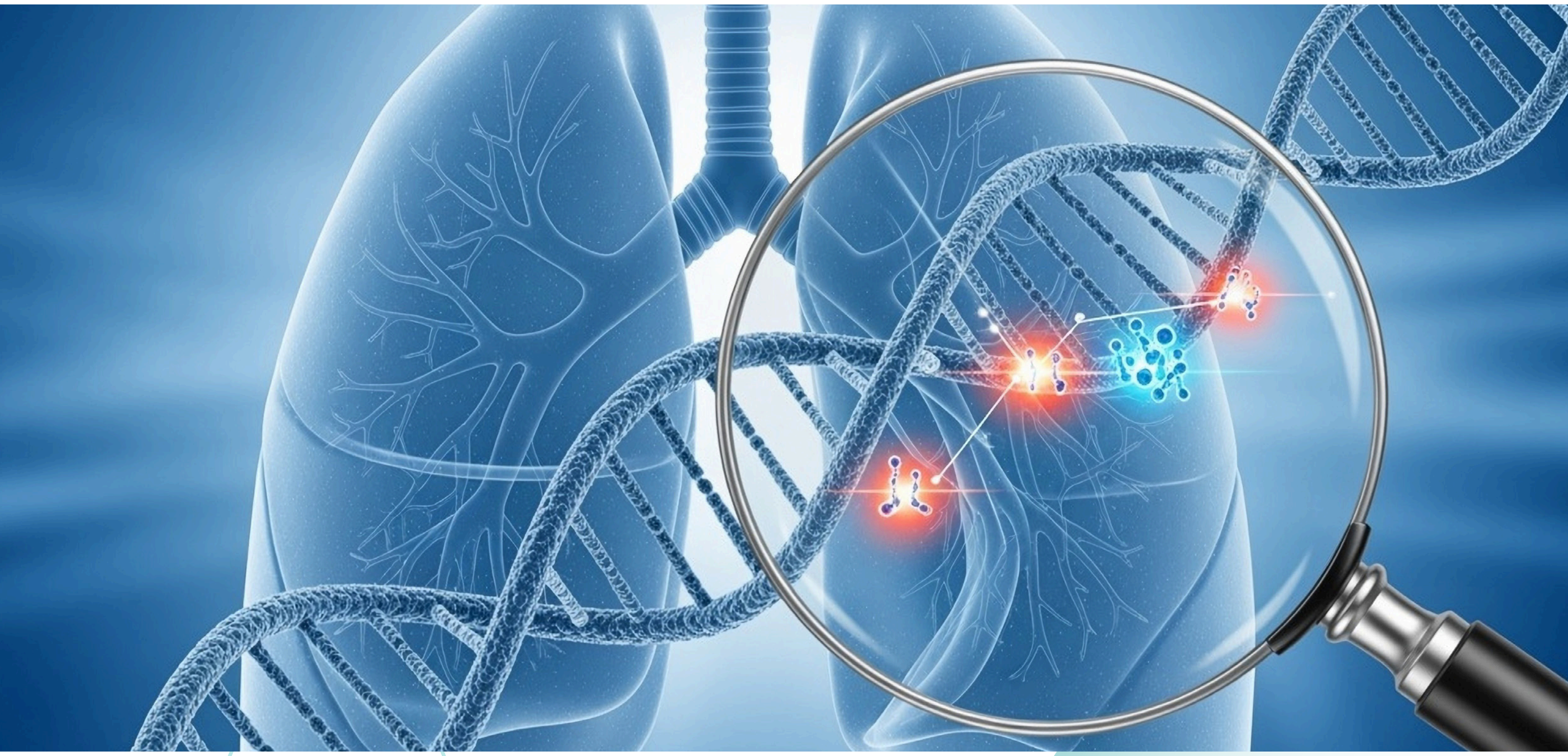
GENEQUALITY® CFTR

GENEQUALITY® CFTR is an *in vitro* diagnostic medical device for the preparation of amplification-based libraries for the qualitative detection of mutations, indels and CNVs in exons, their flanking regions, and clinically relevant intronic regions of the CFTR gene, intended for in vitro diagnostic assays based on NGS.



Low DNA Input
Just **10 ng** per reaction

Sequencing Ready
Compatible with **Illumina** platforms

Complete CFTR Coverage
Exons, **Exon–Intron boundaries**, **Promoter regions**, clinically relevant **Intronic variants**, **poly-T** and **TG repeats**, direct **CNV analysis**.

Next-Generation Sequencing

EU-IVDR coming soon

One-Tube Library Prep
Minimal contamination, **<45 min** hands-on, full protocol **<3 hours**

Easy Analysis
Optional **Seqpilot** software

Streamlined Workflow
Single-tube pooling & cleanup

Validated Sample Types
Whole blood, **amniotic fluid**, **buccal swab**, **DBS**

ANALITICA
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GENEQUALITY® CFTR

The CFTR gene encodes a membrane protein that regulates chloride and bicarbonate transport across epithelial surfaces.

Mutations cause thick, sticky secretions that block airways and ducts, promoting chronic infections.

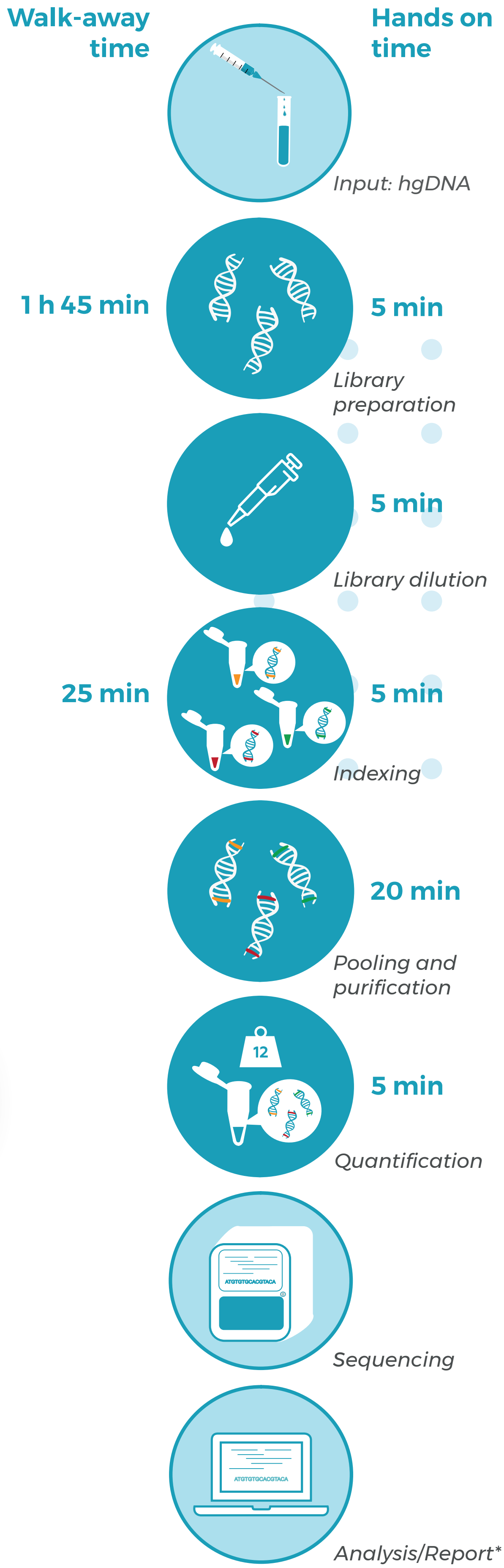
The easy-to-use **GENEQUALITY® CFTR** is a library preparation kit for NGS, specifically designed for the analysis of the CFTR gene through targeted amplification.

The workflow involves two PCR steps:

First PCR: highly multiplexed amplification of target regions in a single tube per sample.

Second PCR: addition of Unique Dual Indexes (UDI) for sample identification, along with Illumina-compatible adapters.

After amplification, samples are pooled, purified, quantified, and loaded onto high-throughput Illumina sequencing platforms.



PRODUCT CHARACTERISTICS:

- Input DNA: **10 ng** per reaction (2 ng/µL).
- Sequencing compatibility: **Illumina platforms**, paired-end 2×150 bp.
- Analysis software: **Seqpilot,** by JSI Medical Systems (*optional supply)
- **Comprehensive CFTR Gene Sequencing**: Reliable detection of exons, exon–intron boundaries, promoter regions, and clinically relevant intronic variants, including poly-T and TG repeats. Direct CNV analysis.
- **One-Tube NGS Library Preparation:** Single-tube NGS protocol minimizes contamination risk and reduces **hands-on time to under 45 minutes**.
- **Complete protocol in less than 3 hours**.
- **Streamlined Workflow & Library Cleanup:** Sample pooling in a single tube simplifies cleanup and optimizes the entire process.

SPECIMENS:

Validated on:

- **Whole blood**
- **Amniotic fluid**
- **Buccal swab**
- **Dried Blood Spots (DBS)**

ORDERING INFORMATION:

PRODUCT	CODE	FORMAT
GENEQUALITY® CFTR	04-NFC-24	24 tests

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