4bases

Precision Matters

NGS solutions for

Oxford NANOPORE Technologies

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Table of Contents

4bases
Precision Matters

1. The Company

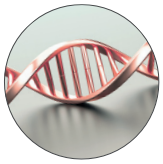
1.1 Who we are

4bases is a Swiss, ISO 13485 certified company specialized in NGS diagnostics.

Our Mission

development and production of comprehensive solutions
for genetic profiling, precision oncology
and microbiology analyses

What We Offer



- platform indepedent NGS kits
- data analysis software
- diagnostic protocols

Our Values

- precision
- quality
- innovation
- patient focus

1.2 Technology

PRO kits

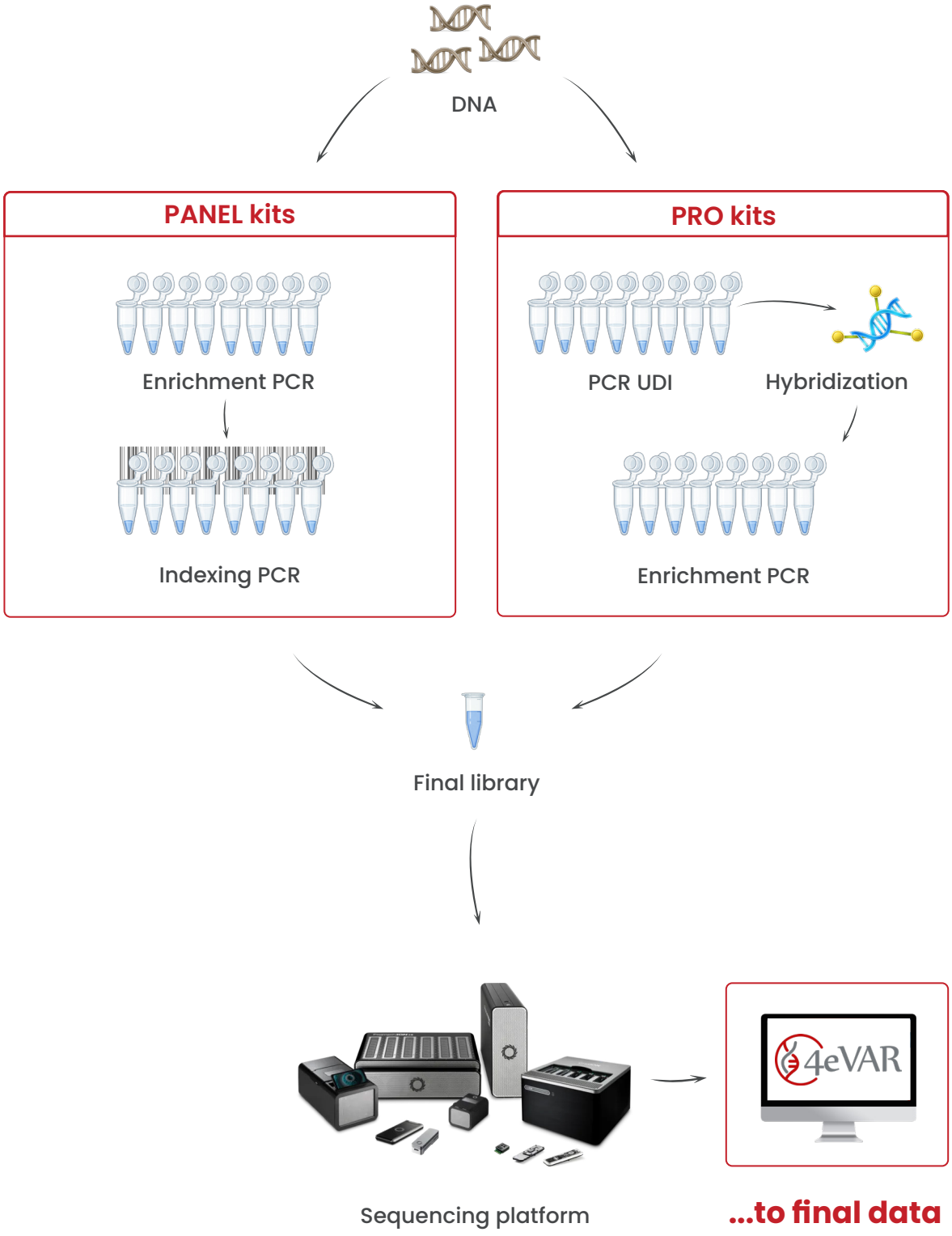
The PRO family kits are 4bases capture-based solutions, with one of the shortest protocols available on the market. dsDNA probes allows for stability over time and higher coverage uniformity, giving high specificity and sensitivity in detecting CNVs and SNVs. DNA purification beads, streptavidin beads, and UDIs are included in each 4bases pro kit. The whole protocol is automatable and compatible with the solutions available on the market.

PANEL kits

The PANEL family kits are 4bases amplicon-based solutions, with a straightforward protocol. Even with low input DNA, specific genes or genetic regions are targeted and amplified with ease.

1.3 Workflow

From DNA sampling ...



...to final data

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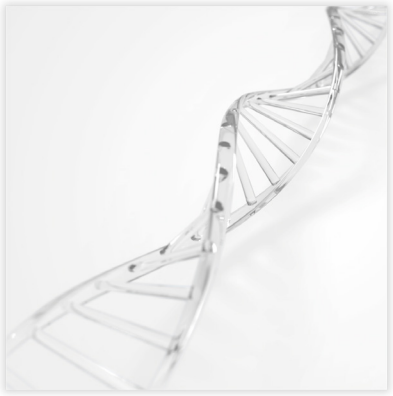
2. Applications

Since 2013, 4bases produces advanced diagnostic solutions for clinical applications in precision medicine.

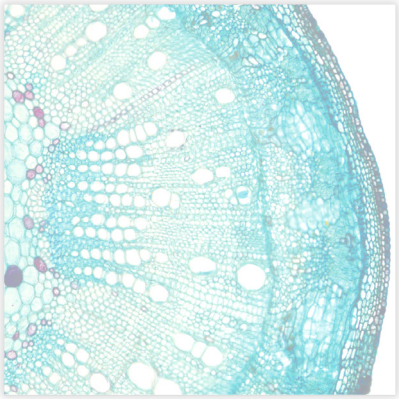


Genetics

Comprehensive panels for profiling across multiple genetic diseases

Oncology

Oncology applications enabling precision cancer care



Microbiology

Kits for bacterial, fungal and viral genome sequencing

DATA ANALYSIS

4eVAR

ONE CODEX

Proprietary and dedicated software applications

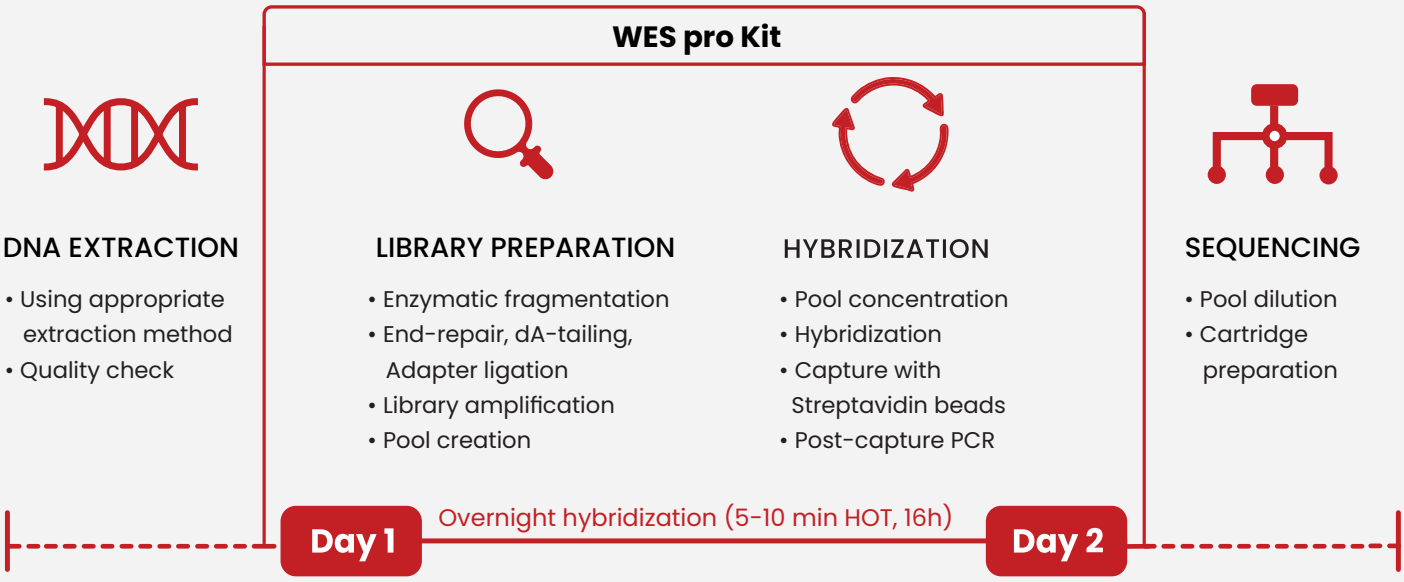
3. Genetic solutions

3.1 WES pro

WES pro kit is designed to target the coding regions of more than 22,000 genes of the human genome. WES Pro is an IVDR-certified class A product — ensuring the highest standards and quality for the exome-enriched library preparation in a laboratory workflow.

- Panel size: About 40 Mb
- Mitochondrial genes (as described in MitoMap) included
- CE-marked
- **Results analysis using the proprietary 4eVAR software***

From DNA to Final data in less than 2,5 days with less than 3 hours hands-on time.



Product	Cod.	Tests number
WES pro	Series DC4130	16-96

*For secondary analysis, 4bases provides its proprietary 4eVAR software, which laboratories may choose to integrate as part of their diagnostic workflow. In this case, the workflow must be validated by the laboratory within the overall procedure.The software enables the generation of a complete report including SNVs, CNVs and mitochondrial genes (optional).

3.2 WholEX pro

WholEX pro kit is designed to target the coding regions of more than 22,000 genes of the human genome.

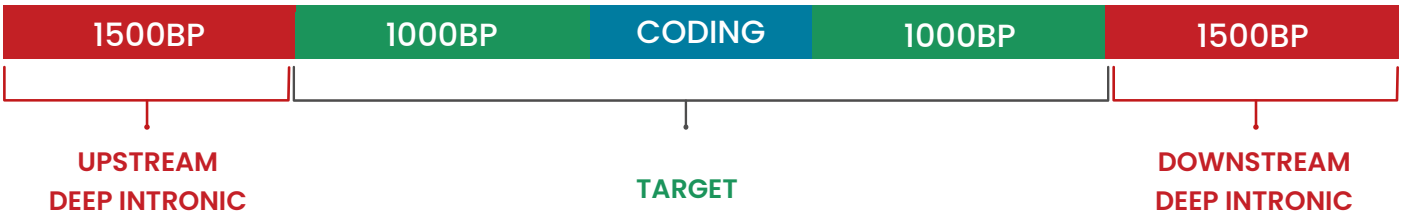
- Panel size: About 40 Mb
- Mitochondrial genes (as described in MitoMap) included
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs, CNVs and mitochondrial genes (optional).

Product	Cod.	Tests number
WholEX pro	Series RDC3130	16-96

3.3 WholEX pro LONG

WholEX pro Long kit is designed to target more than 22,000 genes of the human genome.

- Read: > 5 Kb
- Mitochondrial genes (as described in MitoMap) included
- Complete report with SNVs, CNVs and mitochondrial genes (optional)
- For research use
- **Results analysis via the dedicated proprietary 4evar software**



CODING: High-coverage analysis of coding regions of >99% protein coding genes.
TARGET: CODING +/-1000 bp: Includes coding regions plus 1000 bp flanks to capture promoters and terminators.
DEEP INTRONIC: Extends 1500 bp beyond the target to explore deep intronic areas, useful for identifying deletion breakpoints despite lower coverage.

Product	Cod.	Tests number
Wholex pro Long	Series RDC3130L	16-96

3.4 ClinEX pro

ClinEX pro kit is designed to detect genetic factors responsible of disease conditions attributable to known pathologies.

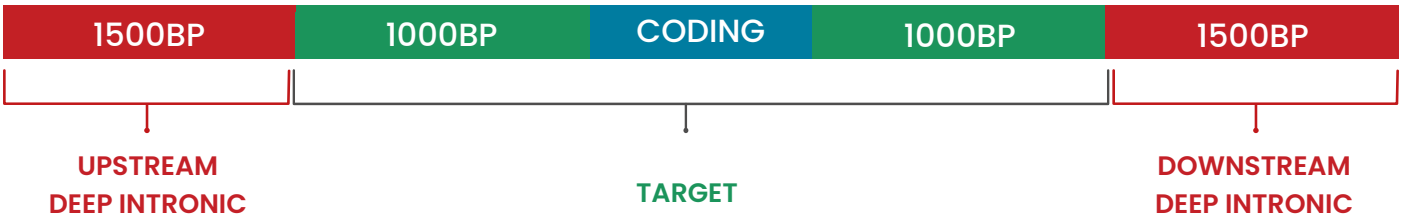
- Panel size: About 21 Mb
- More than 10,000 genes described in OMIM as related to known pathologies, plus ~130 hotspots, SNPs
- Mitochondrial genes (as described in MitoMap) included
- CE-marked
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs, CNVs and mitochondrial genes (optional).

Product	Cod.	Tests number
ClinEX pro	Series DC3030	16-96

3.5 ClinEX pro LONG

ClinEX pro Long kit is designed to detect genetic factors responsible of disease conditions attributable to known pathologies.

- Target size about 21 Mb
- Read: > 5 Kb
- More than 10,000 genes described in OMIM as related to known pathologies, plus ~130 hotspots, SNPs, deep intronic regions
- Mitochondrial genes (as described in MitoMap) included
- Complete report with SNVs, CNVs and mitochondrial genes (optional)
- For research use
- **Results analysis via the dedicated proprietary 4evar software**



CODING: High-coverage analysis of coding regions of disease-associated genes.
TARGET: CODING +/-1000 bp: Includes coding regions plus 1000 bp flanks to capture promoters and terminators.
DEEP INTRONIC: Extends 1500 bp beyond the target to explore deep intronic areas, useful for identifying deletion breakpoints despite lower coverage.

Product	Cod.	Tests number
ClinEx pro LONG	Series RDC3030L	16-96

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3.6 Human WGS

Human WGS Kit enables comprehensive whole-genome sequencing, delivering high-quality detection of genetic variants to support a broad range of applications.

- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
Human WGS	Series RC3290	16-96

3.7 Methylation pro

Methylation PRO kit is designed for the evaluation of methylation status in PCR-free DNA libraries.

- Complete report with methylation status, haplotypes and DMR
- For research use
- **Results analysis via the dedicated proprietary 4evar software**

Product	Cod.	Tests number
Methylation pro	Series RDC3230Y	16-96

3.8 CARDIO pro

CARDIO pro kit enables genetic characterization of familial cardiovascular diseases, allowing the analysis of over 200 genes associated with hereditary cardiomyopathies, rare cardiac disorders, and hereditary aneurysmal diseases, in line with the latest international guidelines.

- Panel size: About 750 Kb
- Mitochondrial genes (as described in MitoMap) included
- Validated for germline samples (DNA extracted from tissues, blood or body fluids)
- CE-marked
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs, CNVs and mitochondrial genes (optional)

Product	Cod.	Tests number
CARDIO pro	Series DC3140	16-96

3.9 CFTR panel

- CFTR panel is a kit for the identification of mutations in CFTR gene.

- Panel size: 35 Kb
- Coverage of the entire coding region and ± 25 bp beyond the flanking regions
- 1st and 2nd level analysis
- Coverage of all variants present in the CFTR2 database
- CE-marked
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including CNVs, SNVs and Poly-T and poly-TG detection.

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

3.10 THALASSEMIA panel

THALASSEMIA panel is a kit designed for the identification of alpha and beta Thalassemia related variants.

- Panel size: 9 Kb
- Molecular profiling of variants causing alpha and beta Thalassemia
- CE-marked
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
THALASSEMIA panel	Series H1090	16-96

3.11 Neoscreen pro

Neoscreen pro is a kit for the screening of clinically significant genes, including inherited disease-related genes, in newborns.

- Panel size: 940 Kb
- Validated for germline samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
Neoscreen pro	Series RDC3280	16-96

3.12 IVF pro

4bases IVF Pro kit has been designed for carrier analysis and newborn screening of autosomal recessive inherited diseases, rare conditions, X-linked diseases, and hereditary cancers.

- Panel size: About 425 Kb
- Validated for germline and samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
IVF pro	Series RDC3100	16-96

3.13 PID pro

PID pro kit is specific for the identification of mutations in genes related to Primary Immunodeficiency Disorders (PIDs).

- Panel size: 1,1 Mb
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
PID pro	Series RDC3090	16-96

3.14 FH panel

FH panel kit pro kit allows for the detection of well-know and novel germline variants associated with familial hypercholesterolemia.

- Panel size: 32 Kb
- Validated for germline samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
FH panel	Series RH1111	16-96

3.15 Iron chip pro

Iron chip pro is a kit for research for the identification of mutations in genes related to metabolism of iron.

- Panel size: 45 Kb
- Validated for germline samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Cod. Kit Product	Cod.	Tests number
Iron chip pro	Series RDC3300	16-96

3.16 RETINA pro

RETINA pro allows for the identification of the variants involved in the Retinopathies.

- Panel size: About 310 Kb
- Validated for germline samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
RETINA pro	Series RDC3180	16-96

3.17 NEPHRO pro

NEPHRO pro is developed for the evaluation of nephrotic syndromes, cystic kidney diseases, tubular and glomerular dysfunctions, enabling the detection of a wide range of clinically relevant genetic variants.

- Panel size: About 255 Kb
- Validated for germline samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs and CNVs.

Cod. Kit Product	Cod.	Tests number
NEPHRO pro	Series RDC3170	16-96

3.18 RIDS pro

RIDS pro kit is designed to investigate genetic variations involved in immune pathways and support personalized medicine.

- Panel size: About 175 Kb
- Validated for germline samples
- For research use
- **Results analysis via the dedicated proprietary 4evar software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
RIDS pro	Series RDC3000	16-96

4. Oncology solutions

4.1 BRaCA panel

BRaCA panel kit allows for the identification of mutations in BRCA1 and BRCA2 genes.

- Panel size: 30 Kb
- BRCA1 and BRCA2 genes coding regions
- Validated for germline samples
- CE-marked
- **Results analysis via the dedicated proprietary 4evar software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
BRaCA panel	Series H1070	16-96

4.2 BRaCA screen

BRaCA screen kit allows for the identification of mutations in BRCA1, BRCA2 and TP53 genes.

- Panel size: 42 Kb
- Validated for somatic samples
- For research use
- **Results analysis via the dedicated proprietary 4evar software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
BRACA screen	Series RS2000	16-96

4.3 HEVA pro

HEVA pro kit is specific for the identification of mutations in genes involved in Breast and Ovary cancer, familial adenomatous polyposis (FAP), and hereditary nonpolyposis colorectal cancer (HNPCC).

- Panel size: About 150 Kb
- Validated for germline and somatic samples
- CE-marked
- **Results analysis via the dedicated proprietary 4evar software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
HEVA pro	Series DC3040	16-96

4.4 HRD pro

HRD pro kit allows for the identification of mutations in genes related to the Homologous recombination and repair (HRR) pathways.

- Panel size: About 80 Kb
- Validated for germline and somatic samples
- For research use
- **Results analysis via the dedicated proprietary 4evar software.** The software enables the generation of a complete report including HRD score, SNVs and CNVs.

Product	Cod.	Tests number
HRD pro	Series RDC3080	16-96

4.5 PANCANCER pro

PANCANCER pro kit allows for molecular profiling of variants involved in onset and progression of solid tumors.

- Panel size: About 227 Kb
- Validated for germline and somatic samples
- CE-marked
- **Results analysis via the dedicated proprietary 4evar software**. The software enables the generation of a complete report including SNVs and CNVs.

Product	Cod.	Tests number
PANCANCER pro	Series DC3200	16-96

4.6 HEMATO pro

HEMATO pro kit is designed for the analysis of complex genomic variants associated with Lymphoid and Myeloid Diseases.

- Panel size: About 395 Kb
- Validated for germline and somatic samples
- CE-marked
- **Results analysis via the dedicated proprietary 4evar software**. The software enables the generation of a complete report including SNVs, ITDs and CNVs.

Product	Cod.	Tests number
HEMATO pro	Series DC3150	16-96

4.7 BENKit panel

BENKit panel kit is designed to enable comprehensive analysis for profiling hotspots relevant to therapy selection in metastatic colorectal cancer and non-small cell lung carcinoma.

- Panel size: 2 Kb
- Validated for somatic samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including hotspot variants.

Product	Cod.	Tests number
BENKit panel	Series RH1020	16-96

4.8 THYRO-ID panel

THYRO-ID panel kit is an advanced medical device developed for the precise molecular profiling of genetic variants associated with thyroid cancer development.

- Panel size: About 2 kb
- Validated for somatic samples
- For research use
- **Results analysis using the proprietary 4eVAR software**. The software enables the generation of a complete report including SNVs.

Product	Cod.	Tests number
THYRO-ID panel	Series RH1030	16-48

4.9 FUSION pro

FUSION Pro is a kit for research specifically designed for the detection of clinically relevant gene fusions and associated transcriptomic alterations in cancer.

- Panel size: About 370 Kb
- Validated for somatic samples
- Complete report with SNVs, and fusion transcripts
- For research use
- **Results analysis via the dedicated proprietary 4eVAR software**.

Product	Cod.	Tests number
FUSION pro	Series RDC3190	16-96

5. Microbiology solutions

5.1 MICROBIOME panel

MICROBIOME panel kit allows for the analysis of the hypervariable regions of the bacterial 16S rDNA.

- Panel size: 3 kb
- The kit is validated for analysis of DNA extracted from fecal, pulmonary, oral, vaginal samples and from surface and soil samples.
- For research use
- **Data analysis available on One Codex**

Product	Cod.	Tests number
Microbiome panel	Series RH1050	16-96

5.2 MICROBIOME PLUS panel LONG

MICROBIOME PLUS panel LONG is designed for the molecular profiling of different human microbiome communities.

- Panel size: 6 Kb
- Amplification of bacterial (16S), fungal and mold (18S, ITS1, 5.8S, ITS2) genes
- Long reads-based protocol
- **Data analysis available on One Codex**

Product	Cod.	Tests number
MICROBIOME PLUS panel LONG	Series H1130L	16-96

5.3 MICROBIOME WGS

MICROBIOME WGS (Whole Genome Sequencing) is a kit designed for the complete genome profiling of different microbiome communities through a shotgun protocol.

- The kit is validated for clinical and environmental samples
- Applications: early identification of emerging infections; detection of polymicrobial and fastidious pathogens; microbiota–immune system interaction studies; antimicrobial resistance (AMR) gene profiling.
- For research use
- **Data analysis available on One Codex**

Cod. Kit Product	Cod.	Tests number
MICROBIOME WGS	Series RC3270	16-96

5.4 COVID panel

COVID Panel is a kit designed for the analysis of the entire SARS-CoV-2 genome.

- Panel size: 33 Kb
- The kit is validated for analysis of DNA and cDNA samples
- For research use
- **Results analysis via the dedicated proprietary 4evar software**. The software enables the generation of Nextclade report.

Product	Cod.	Tests number
COVID panel	Series RH1110	16-96

6. Data Analysis

6.1 4eVAR

4eVAR is 4bases proprietary analysis platform, where clinical results are easily and efficiently accessible through a validated and secured internal workflow.

- The data analysis on 4eVAR is designed based on the characteristics and technologies of the kit used, to increase accuracy of results.
- A complete report including SNVs and CNVs for variant interpretation is given per each sample. Specific links for wider yet complete classification of detected variants are also provided, together with quality metrics per each sample in the run.

4eVAR platform

4eVAR tutorial

ONE CODEX

6.2 One Codex

Thanks to the integration of One Codex database and pipeline analyses with 4bases short and long reads sequencing solutions, clinical laboratories can perform routine microbiome analyses as well as short- and long-term monitoring in easy, straightforward and efficient solutions.

This collaboration merges the reliability and efficiency of 4bases CE-IVD microbiome kits with the unique accuracy and sensitivity of One Codex database to characterize microbial communities in clinical samples.

Microbiome Suite

4bases

Precision Matters

4bases SA

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