ClinEX pro

Extended Clinical Exome NGS kit



ClinEX PRO

4bases ClinEX pro kit is designed to detect genetic factors responsible of disease conditions not attributable to known pathologies, genetically heterogeneous diseases, and complex syndromic pictures. The kit targets more than 10,000 genes described in OMIM as related to known pathologies, plus ~130 hotspots, SNVs, deep intronic regions with a known hereditary to Mitochondrial genes (as described in MitoMap) are also optionally covered and can be included in the analysis as needed. The total target size of ClinEX pro is 19.5 Mb and is optimized for Oxford Nanopore Technologies' Next Generation Sequencing (NGS) platforms. The complete table of target genes can be accessible upon request.

TECHNOLOGY

ClinEX pro kit contains all reagents needed to target and capture more than 10,000 genes (table available upon request), allowing for library multiplexing and NGS sequencing using Oxford Nanopore Technologies platforms (Table 1).

WORKFLOW

The ClinEX pro kit is part of a capture-based family solution of 4bases molecular technologies. Its accuracy and specificity in detecting genetic variants (i.e. SNVs, CNVs), with one of the shortest capture-based protocols available on the market (36 hours protocol, <3 hours hands-on-time), are the key to its reliability. Thanks to 4bases proprietary analysis platform, 4eVAR, the clinical results are easily and efficiently accessible through a validated internal workflow.

SOFTWARE ANALYSIS

The data analysis on 4eVAR is designed based on the characteristics and technologies of the kit used, to increase accuracy of results, and to have the complete control on the entire process. A complete report including SNVs and CNVs with ACMG classification 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.

VALIDATION

The **ClinEX pro** kit has been validated both internally and in clinical laboratories, and the whole protocol was tested on DNA standards and on clinical samples.



Table 1: ClinEX pro samples per sequencing run on Oxford Nanopore Technologies' sequencers.



Flow Cell	Samples per Flow Cell (Coverage 50x)	Oxford Nanopore sequencer	Flow Cells per sequencer *
FLO-MIN114	2	MinION	1
		GridION	Up to 5
FLO-	FLO-		Up to 2
PRO114M	8	PromethION 24	Up to 24
		PromethION 48	Up to 48

*Multiple Flow Cells can be used during the same run session (GridION, PromethION), either with different libraries or with the same library to increase sequencing depth.

PLEASE NOTE: The volumes of the reagents in the kit are calculated to allow the subdivision into multiples of 8 samples per analysis session. Dividing the kit differently will decrease the total number of tests that can be performed. For different settings, please contact us at support@4bases.ch.

PLEASE NOTE: The number of multiplexable samples and coverage depend on starting gDNA degradation, presence of contaminants, external factors, library loading procedure.

ORDERING INFORMATION **

	Product	REF
LIBRARY AMPLIFICATION	ClinEX pro	DC3030-16X (16 test)
	ClinEX pro	DC3030-96X (96 test)
REAGENTS	ClinEX pro AUTOMATIC	C3030A-96X (96 test)
MULTIPLEXING	UDI Primers Set C	7003 (96 test)
REAGENTS	UDI Primers Set 16	7005 (16 test)
OXFORD NANOPORE ADAPTERS	NANO ADAPTER	M1050-6 (6 sequencing run)
		M1050-24 (24 sequencing run)

^{**}UDI are always included in CEIVD kit.

PLEASE NOTE: The reagents required for sequencing on Oxford Nanopore Technologies' platforms have to be purchased separately: contact 4bases for further information.

- For more detailed information regarding validated solutions for automatic library preparation, please contact us at info@4bases.ch
- For the target regions of *ClinEX pro kit* (.bed file), please contact us at **support@4bases.ch**