RASPO pro

For the screening of Neurofibromatosis and Rasopathies

RASPO pro

RASPO pro is a kit for the identification of mutations in genes related to Neurofibromatosis and Rasopathies.

Rasopathies and Neurofibromatosis are characterized by high heterogeneity: the dysregulation of the RAS-MAPK pathway can be determined not only by germline mutations of the RAS gene, but also by mutations in genes that regulate RAS activity (PTPN11, SOS1, SHOC2, NF1 e SPRED1), downstream signal transduction (RAF1, BRAF, MEK1 e MEK2) o the activity of receptors (CBL).

RASPO pro is a kit for the analysis of 29 genes through a molecular protocol based on Next Generation Sequencing (NGS) technology. The kit is validated for the germline analysis (SNPs, indels, CNVs) of DNA samples extracted from blood or tissues. RASPO pro kit contains all the reagents needed for the preparation and capture of specific probes designed for sequencing on Thermofisher platforms.

WORKFLOW

Library preparation follows a straightforward, capture-based protocol that can be completed in as little as 36 hours, with < 3 hours hands-on time. Resulting libraries can be normalized, pooled, and then loaded on to a flow cell for sequencing. Prepared libraries are sequenced on any compatible Thermofisher sequencers.

VALIDATION

To demonstrate assay capabilities, clinical samples were run in a clinical setting. DNA quality and quantity of the libraries prepared were verified using Qubit and Agilent Bioanalyzer.
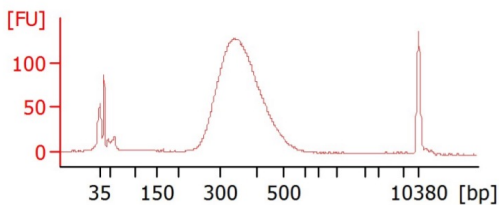

Figure 1. RASPO pro library Bioanalyzer profile.

Table 1: **RASPO pro** genes list.

A2ML-1	MRAS	SPRED1	SYNGAP1
BRAF-1	NRAS	CBL	MAPK1
HRAS	PPP1CB	RAF-1	MAPK3
KRAS	PTPN11	SHOC-2	NF1
RRAS	RASA1	RIT-1	NF2
LZTR1	RASA2	SOS1	
MAP2K-1	KIT1	SOS2	
MAP2K-2	RIT-1	SMARCB1	

SAMPLE PER RUN

Instrument	Samples per run*
	Germline
Ion 316™ Chip/Ion 510™ Chip	16
Ion 318™ Chip/Ion 520™ Chip	32
Ion 530™ Chip	112
Ion PI™ Chip/Ion 540™ Chip	352
Ion 550™ Chip	992

*the estimated maximum number of samples per chip assumes a reading depth of 300x for the germline. The optimal number of samples can be empirically estimated on the local setup.
The volume present in the kit is calculated to allow the subdivision into multiples of 8 analysis sessions. Dividing the kit in different ways decrease the total number of tests that can be performed.

ORDERING INFORMATION

Product	REF
RASPO pro	RC3010Y-16 (16 test)
RASPO pro	RC3010Y-96 (96 test)
Adapter	
Y ADAPTER	R9001-16 (16 test)
Y ADAPTER	R9001-96 (96 test)