EasyNGS HCPanel

HoopBio

**Panel of 40 genes created for and by experts in clinical diagnosis.
Allows the analysis of the most relevant DNA regions involved in a increased predisposition to develop hereditary cancer syndromes.**

Determination of hereditary cancer by massive sequencing

GENES STUDIED IN THE PANEL

HEREDITARY CANCER				Lynch syndrome	Familial Paraganglioma Polyposis	Pheochromocytoma Paraganglioma	Other Genes to analyze according comparative criteria
Breast + Ovary		Breast	Ovary				
ATM	MLH1	ATM	BCRA1	MLH1	APC	FH	PTENPTE
BRCA 1	MSH2	BRCA 1	BCRA2	MSH2	MUTYH	MAX	STK11
BRCA 2	MSH6	BRCA 2	BRIP1	MSH6	BMPR1A	MET	TP53
CHEK2	RAD50	CHEK2	MLH1	PMS2	SMAD4	NF1	NF1
NBN	RAD51C	NBN	MSH2	EPCAM	POLE	RET	CDH1
PALB2	RAD51D	PALB2	MSH6	POLE	POLD1	SDHA	
BRIP1	EPCAM	RAD50	EPCAM	POLD1		SDHB	
			RAD51C			SDHC	
			RAD51D			SDHD	
						TMEM127	
						SDHAF2	

Cowden syndrome	Von Hippel-Lindau syndrome	Hereditary Retinoblastoma	Peutz-Jeghers syndrome	Multiple Endocrine Neoplasia Type 1	Multiple Endocrine Neoplasia Type 2	Hereditary diffuse gastric cancer	Familial Melanoma syndrome
PTEN	VHL	RB1	STK11	MEN1	RET	CDH1	CDKN2A

Aproximately 5-10% of all cancers have an inherited character, hence the high interest in studying them.

Technical informations

- Starting sample: blood
- 1M readings per sample
- 126bp medium amplicon size
- Covers the exonic region of 40 genes
- + 10bp of intronic region
- Great uniformity of readings
- Starting quantity: 10ng

Study of:

- Mutations
- Insertions
- Deletions
- CNVs (It allows to develop a specific laboratory baseline for CNV detection)

www.hoopbio.com

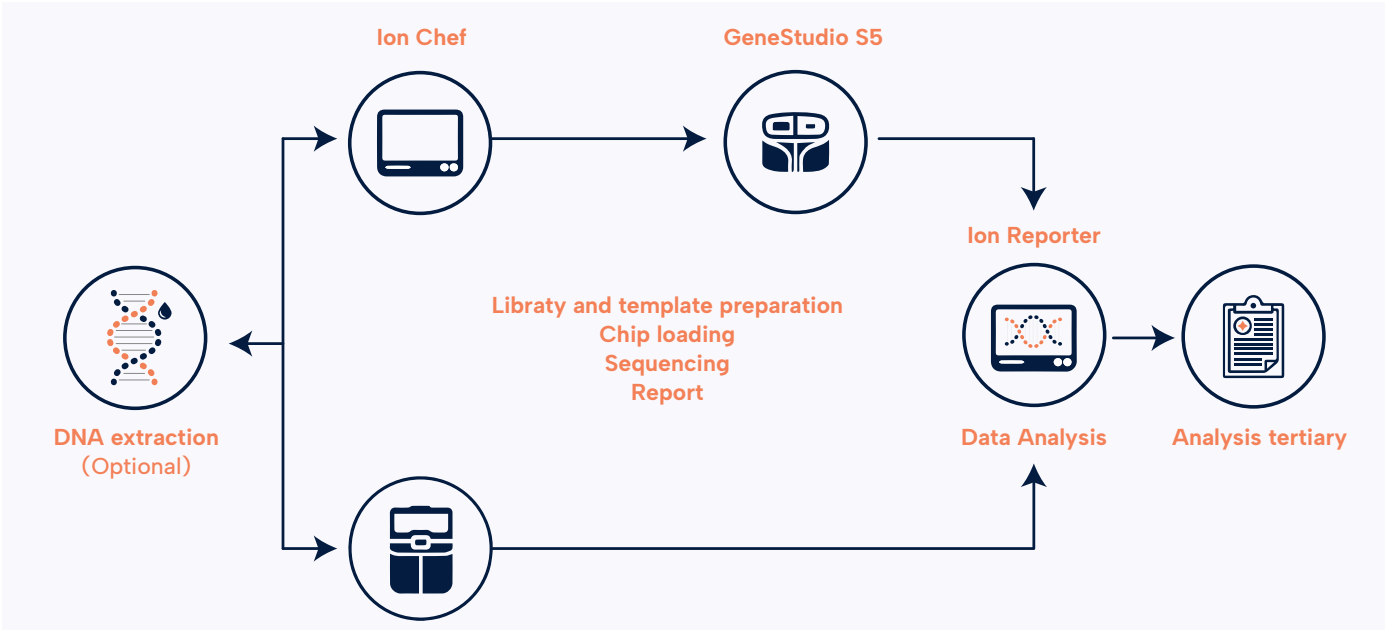
CE IVD **ISO 13485**

Coverage Overview

The perfect combination of EasyNGS HC + Ion Torrent

- **From DNA to variants call:**
- 3 days with S5+Ion Chef
- 1 day with Genexus
- **Automated library prepartion**
- Consistant results
- Error limitation

- **Workfow validated with tertiary Software**
- **Panel and workfow verified by end users**
- **Flexibility with 5 diferent chips** (from 2M to 130M reads)



- Uniformity of amplicon coverage: 97.75%
- Amplicons with at least 1 read: 99.90%
- Amplicons with at least 20 reads: 99.38%
- Amplicons with at least 100 reads: 95.07%
- Amplicons with at least 500 reads: 5.95%
- Amplicons with no strand bias: 98.72%
- Amplicons reading end-to-end: 97.79%
- Amplicon base composition bias: 0.566

- Percent base reads on target: 93.86%
- Average base coverage depth: 387.7
- Uniformity of base coverage: 97.61%
- Target base coverage at 1x: 99.94%
- Target base coverage at 20x: 99.56%
- Target base coverage at 100x: 95.95%

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