Biomarker and Focus Panel –Test Information

Indications for ordering

This test allows the detection of genomic abnormalities with a diagnosis, prognosis or predictive role in diverse solid tumours (e.g. lung, lower GI, melanoma).

Select **DNA** in Next Generation Sequencing section of the requisition form for hotspot variation testing (single nucleotide variants (SNVs) and small insertion / deletions (indels).

Select **RNA** in Next Generation Sequencing section of the requisition form for gene fusion testing.

Test Methodology

The Focus Panel is a targeted next generation sequencing assay for analysis of hotspot regions in 52 genes with known relevance to solid tumours. The panel enables the simultaneous analysis of genes associated with several most common cancers (e.g. lung, colon). The DNA and RNA analysis can detect multiple alterations including: (i) single nucleotide variants (SNVs), (ii) small insertion / deletions (indels) and (iii) gene fusions in RNA samples. See below for gene list.

Gene List

DNA (hotspot variations)				
AKT1	EGFR	FGFR4	JAK3	MYCN
ALK	ERBB2	GNA11	KIT	NRAS
AR	ERBB3	GNAQ	KRAS	PDGFRA
BRAF	ERBB4	HRAS	MAP2K1	PIK3CA
CCND1	ESR1	IDH1	MAP2K2	RAF1
CDK4	FGFR1	IDH2	MET	RET
CDK6	FGFR2	JAK1	MTOR	ROS1
CTNNB1	FGFR3	JAK2	MYC	SMO
DDR2				

RNA (fusions)				
ABL1	EGFR	ETV5	NTRK1	PPARG
ALK	ERBB2	FGFR1	NTRK2	RAF1
AKT3	ERG	FGFR2	NTRK3	RET
AXL	ETV1	FGFR3	PDGFRA	ROS1
BRAF	ETV4	MET		

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Test Interpretation

Only variants with a recognized clinical significance, diagnostic, prognostic or predictive are reported (Tier I and II; PMID: 27993330). Additional results are available upon request.
Note: This test does not detect large deletions. For more information on the hotspot regions tested, please contact the laboratory.

Type of specimens accepted

- Tumour cell content ≥ 20% is required. This information is **mandatory** to assess the validity of the test.
- Specimen (cytology or histology), frozen or formalin-fixed paraffin-embedded (FFPE) are accepted.

Limitations

Results must be interpreted in the context of clinical, radiological and histological findings. If results obtained do not match other clinical or laboratory findings, or if you have novel relevant information, please contact the laboratory as soon as possible for updated interpretation. Results may be compromised if the recommended procedures (tissue fixation and preparation) have not been followed. A negative (wild type) result does not fully rule out the presence of an alteration but may be linked with limits of detection of this assay (i.e. insufficient % of tumour cell content or poor fixation). Rare polymorphisms may be present that could lead to false-negative or false-positive results. Turnaround time: 10 working days

Abbreviations for indications:

AC	Adenocarcinoma
SCC	Squamous cell carcinoma
HNSCC	Head and Neck Squamous cell carcinoma
TNBC	Triple negative breast cancer
Cervical SCC	Cervical squamous cell carcinoma (gynecopathology)
Eso/GEJ AC/SCC	Esophageal or gastroesophageal junction adenocarcinoma or squamous cell carcinoma (does not include gastric)
Eso/GEJ/Gas AC	Esophageal or gastroesophageal junction or gastric adenocarcinoma (does not include squamous cell carcinoma)