Targeted RNASeq Panel – Test Information

Indications for ordering

This test detects translocations/fusions, exon skipping, internal tandem duplications, and sequence variants in select oncogenes (see **Gene list**) that may have diagnostic, prognostic, or therapeutic implications for patients with certain tumour types (see below). This test may be ordered for patients with the following tumour types:

- Sarcomas
- Cholangiocarcinomas
- Brain tumours
- Rare solid tumours
- Certain hematological malignancies

Test Methodology

The *Targeted RNASeq panel* is a targeted next generation sequencing capture panel for analysis of a targeted list of genes, relevant to certain tumour types. This test only analyzes RNA and can detect multiple types of alterations, including (i) translocations/fusions, (ii) internal tandem duplications, (iii) single nucleotide variants (SNVs) and small insertion/deletions (indels), (iv) alterations that cause exon skipping.

Test Interpretation

Only variants with a recognized or predictive diagnostic, prognostic or therapeutic clinical significance are reported (Tier I and II; PMID: 27993330). Additional results are available upon request. This test cannot definitively determine whether a variant is present in the germline or restricted to the tumour.

Specimens accepted

- **Tumour cellularity ≥ 20%** is required. This information is mandatory to assess the validity of the test.

Preferentially:

- **Frozen tissue** minimally 30 mg of tissue on dry ice
- **Fresh tissue** on RNA later minimally 30 mg of tissue

If no fresh/frozen tissue is available:

- **FFPE blocks** are NOT accepted
- **Scrolls** or **unstained slides** from Formalin-fixed paraffin-embedded (FFPE) specimens (cytology or histology) are accepted
  - Scrolls: **Ten** scrolls at **10um**
  - Unstained slides: 10 unstained labelled slides of 10 µm thickness, unbaked and uncharged, with 1 H&E slide (tumour area should be outlined with permanent marker if macrodissection is needed).
- The full procedure is detailed in the document "*Sample Preparation and Shipping Guidelines*".

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 result does not fully rule out the presence of an alteration and may be due to the limitations of this assay (i.e. insufficient % of tumour cell content or poor fixation).

Turnaround time: 15 working days

Revised and approved by Dr. Gomez 2024-05-01

GENE LIST

Actionable Genes Fusions									
ABL1	ALK	ATF1	AXL	BCL2	BCL6	BCR	BRAF	BRCA1	BRCA2
CBFB	CD74	EGFR	EIF3E	EML4	ERBB2	ERG	ESR1	ETV4	EWSR1
FGFR1	FGFR2	FGFR3	FGFR4	FIP1L1	FLI1	KMT2A	MAST1	MET	MYC
MYH11	NFKB2	NOTCH1	NRG1	NTRK1	NTRK2	NTRK3	PAX8	PDGFRA	PDGFRB
PML	PPARG	PTPRK	QKI	RAF1	RARA	RET	ROS1	RSPO2	RSPO3
RUNX1	TERT	TMPRSS2							
Clinically Relevant Genes Fusions									
ABCC4	ABI1	ABL2	ACACA	ACE	ACER1	ACKR3	ACSL6	ADD3	AFF1
AFF3	AFF4	AGR3	AHI1	AHRR	AKAP12	AKT3	ANKRD28	AR	ARHGAP20
ARHGAP26	ARNT	ASPSCR1	ASTN2	ATIC	ATP1B4	ATXN1	AUTS2	BACH2	BAG4
BAIAP2L1	BAZ2A	BCAS3	BCAS4	BCL10	BCL11A	BCL11B	BCL2L1	BCL3	BCL9
BCOR	BDNF	BICC1	BIRC3	BIRC6	BRD1	BRD3	BRD4	BRWD3	BTBD18
BTG1	C11orf1	C11orf95	C2CD2L	C3orf27	CAMTA1	CAPRIN1	CARS	CASC5	CASP7
CBFA2T3	CBL	CCAR2	CCDC28A	CCDC6	CCDC88C	CCNB1IP1	CCNB3	CCND1	CCND2
CCND3	CDH11	CDK5RAP2	CDK6	CDX1	CDX2	CEBPA	CEBPB	CEBPD	CEBPE
CEP170B	CEP85L	CHD6	CHIC2	CHMP2B	CHST11	CIC	CIITA	CLP1	CLTC
CLTCL1	CMKLR1	CNBP	CNOT2	CNTRL	COG5	COL1A1	COL1A2	COL6A3	COX6C
CPSF6	CRADD	CREB1	CREB3L1	CREB3L2	CREBBP	CRLF2	CRTC1	CSF1	CSF1R
CTDSP2	CTNNB1	CUX1	DAB2IP	DACH1	DACH2	DDIT3	DDX10	DDX20	DEK
DMRT1	DNAJB1	DNASE2	DPM1	DUSP22	DUX4	EBF1	ECHDC1	EEFSEC	EGR1
EGR2	EGR3	EGR4	EIF4A2	ELF4	ELK4	ELL	ELN	EML1	EP300
EP400	EPC1	EPOR	EPS15	ERBB3	ERC1	ERCC1	ERLIN2	ETS1	ETV1
ETV5	ETV6	EZR	FAM19A2	FCGR2B	FCRL4	FEN1	FEV	FGF8	FGFR1OP
FGFR1OP2	FGR	FHIT	FLNA	FLT3	FLT3LG	FNBP1	FOSB	FOSL1	FOXO1
FOXO4	FOXP1	FRK	FRYL	FUS	GAS5	GAS7	GATA1	GATAD2A	GIT2
GLI1	GORASP2	GOSR1	GOT1	GPR107	GPR128	GPR34	GRHPR	GRID1	GTF2I
H2AFX	HAS2	HEY1	HHEX	HIP1	HIPK1	HIST1H4I	HLF	HMGA2	HNF1A
HOXA10	HOXA11	HOXA13	HOXA9	HOXC11	HOXC13	HOXD11	HOXD13	HSP90AA1	ID4
IKZF1	IL2	IL21R	IL3	INPP5D	INSR	IQCG	IRF2BP2	IRF4	IRS4
ITK	JAK1	JAK2	JAZF1	KANK1	KAT6A	KAT6B	KDM5A	KIAA1524	KIF5B
KPNB1	KSR1	LASP1	LCK	LCP1	LGR5	LHFP	LHX2	LHX4	LINC00598
LINC00982	LMBRD1	LMO1	LMO2	LNP1	LPP	LPXN	LRMP	LRRC37B	LTBP1
LYL1	LYN	MACROD1	MAFB	MALT1	MAML2	MAPRE1	MAST2	MBNL1	MBTD1
MAF	MDS2	MEAF6	MECOM	MEF2D	MGEA5	MKL1	MKL2	MLF1	MLLT1
MLLT10	MLLT11	MLLT3	MLLT4	MLLT6	MN1	MNX1	MSI2	MSMB	MSN
MTCP1	MTHFD1L	MUC1	MUSK	MUTYH	MYB	MYBL1	MYH9	MYO18A	MYO1F
NAB2	NAPA	NBEAP1	NBR1	NCOA1	NCOA2	NCOA3	NCOA4	NDE1	NF1
NFATC2	NFIB	NGF	NGFR	NIN	NIPBL	NKX2-5	NONO	NOTCH2	NPM1
NR4A3	NR6A1	NSD1	NT5C2	NTF3	NTF4	NUMA1	NUMBL	NUP107	NUP214
NUP98	NUTM1	NUTM2A	NUTM2B	OFD1	OLIG2	OLR1	OMD	P2RY8	PAPPA
PATZ1	PAX3	PAX5	PAX7	PBX1	PCM1	PDE4DIP	PDGFB	PER1	PHF1
PHF23	PHIP	PICALM	PIK3CA	PIM1	PKM	PKN1	PLAG1	POM121	POU2AF1
POU5F1	PPAP2B	PPARGC1A	PPFIBP1	PPP2R1B	PRCC	PRDM16	PRKACA	PRKAR1A	PRKCA
PRKCB	PRKG2	PRRX2	PSIP1	PSMD2	PTPRR	PVT1	RABEP1	RAD51B	RANBP2
RAP1GDS1	RBM15	RBM6	RCOR1	RCSD1	RELA	RHOH	RNF213	RPL22	RPN1
RREB1	RRM1	RTEL1	RUNX1T1	SARNP	SEC31A	SEPT2	SEPT5	SEPT6	SEPT9
SERPINE1	SERPINF1	SET	SETBP1	SFPQ	SH3D19	SH3GL1	SIK3	SLC34A2	SLC45A3
SLCO1B3	SMAP1	SMARCA5	SMARCB1	SNHG5	SORBS2	SORT1	SP3	SPDYE4	SPECC1
SPTBN1	SQSTM1	SRF	SRSF3	SS18	SS18L1	SSBP2	SSX1	SSX2	SSX4
ST6GAL1	STAT5B	STAT6	STRN	SUGP2	SUZ12	SYK	TACC1	TACC2	TACC3
TAF15	TAF6L	TAL1	TAL2	TAOK1	TBX15	TCF12	TCF3	TCL1A	TCTA
TEAD1	TEAD2	TEAD3	TEAD4	TEC	TENM1	TET1	TFE3	TFEB	TFG
TFPT	TFRC	TGFBR3	THADA	THRAP3	TIRAP	TLX1	TLX3	TMEM66	TNFRSF17
TOP1	TOP2B	TP53BP1	TPM3	TPM4	TP63	TRHDE	TRIM24	TRIP11	TRPS1
USP16	USP42	USP6	VGLL3	WASF2	WDR18	WDR70	WHSC1	WHSC1L1	WSB1
WT1	WWTR1	XIAP	YAP1	YTHDF2	YWHAE	ZBTB16	ZC3H7A	ZC3H7B	ZFP64
ZFPM2	ZFYVE19	ZMIZ1	ZMYM2	ZMYND11	ZNF207	ZNF384	ZNF444	ZNF521	ZNF585B
ZNF687	ZNF84								
Genes for variant calling (selected hotspots)									
ACVR1	ALK	BRAF	BCOR	CTNNB1	EGFR	FGFR1	FGFR2	FGFR3	H3-3A
H3-C2	H3-C3	HRAS	IDH1	IDH2	KIT	KRAS	NRAS	PDGFRA	PIK3CA
TP53									
Genes for Internal Tandem Repeats									
BCOR	BRAF	CEBPA	EGFR	FGFR1	FLT3	KIT	WT1		

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