

NCCP Framework for Decision Making for Tests (Predictive for Systemic Anti-Cancer Therapy Treatment) in the Irish Molecular Pathology Service

Appendix 3

List of molecular tests predictive for drug treatment performed or out sourced in Ireland (Based on responses from hospitals to NCCP survey 2018) [Note: responses from two cancer centres awaited]

1p19q co-deletion	EWSR1-ERG	MGMT promoter methylation
9q, 10q,17p	EWSR1-Fli1; Erg or various other fusions	MGMT promoter methylation
ALK amplification	EWSR1-WT1 fusion	Micro Satellite Instability (MSI)
ALK amplification or rearrangements	FGFR1 amplification	MLPA uveal melanoma
ALK fusion	FGFR1 duplication	Monosomy 6
ALK-VARIOUS fusions	FGFR1 mutation	MYB rearrangement
Amplification	FN1	MYB-QKI fusion
BCOR fusions, various	FOXO1 rearrangements	MYC amplification
BCOR ITD fusion	FUS- CREB3L3 fusion	MYCN amplification
BRAF mutation	FUS-Erg; FEV fusions	MYCN status
BRAFV600 mutation	Gain 1q,9q,12q,15q,17q,20q	NF1 mutation
BRAFV600E mutation	Gains/Losses	NR4A3-VARIOUS fusions
BRCA1 mutation	GLI1 amplification	NRAS
BRCA2 mutation	H3.1 K27M	NRAS mutation
C11orf95-RELA	H3.3 G34R/V	NUT fusions
C19MC amplification 19q13.42	H3.3 K27M	Other CNV
CDKN2A deletion	HER EXON 20 mutation	PAX3/7-FOXO1 fusion
Chromosome 1p, 11q and 17q status	HER2	PDGFRA amplification

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Version 1

Publication date: 09/07/18

Due for update: 09/07/21

CIC-fusions	HER2	PDGRFA
Col1A1-PDGFB fusion	HER2 ERBB2	PLAG1-VARIOUS fusions
CTNNB1 mutation	HPV 16	PTEN deletion
CTNNB1 mutation (BCatenin)	HPV 18	RAS
DDIT3	IDH1	RET FISH in lung cancer
Deletion 1p,6q,10,14q,18q,9p	IDH2	RET mutation
DNA methylation profiling	isochromosome 17	ROS1
DNA methylation profiling (method 1 of 2)	KIAA1549 EXON 15-BRAF EXON 9 Fusion	ROS1 fusion
EBV	KIAA1549 EXON 16-BRAF EXON 11 Fusion	Segmental aberrations
EGFR	KIAA1549 EXON 16-BRAF EXON 9 Fusion	Segmental aberrations and chr + or -
EGFR amplification	KIT mutation	SLC44A1/PRKCA fusion
EGFR exon 20 insertion	KRAS	SMAARCB1 mutation
EGFR mutation	KRAS mutation	SMARCA4 mutation
EGFR T790M	LOH 1p; 16q	SMARCE1 mutation
EML4-Aik	LOH 1p; 2p; 11q; 17q	SS18 rearrangements
ERBB2	Loss chr 3/Gain chr 8	SS18-SSX fusion
ERBB2	Loss/Gain chr 6	TFE3/B fusions
ETV6-NTRK4 fusion	MAML2	TP53 mutation
EWSR1-ATF1 fusion	MDM2 FISH	USP6-VARIOUS fusions
EWSR1-CREB/ATF2 fusions	MET FISH in lung cancer	YWHAE-NUTM3 fusion
EWSR1-DDIT4 fusion	MGMT	