

Providing precise melanoma diagnosis and therapy-decision support with MelArray

Oncobit's mission is to enable personalized cancer care by finding the best cancer treatment for each patient. Oncobit designs and develops precise diagnostic and monitoring tests and provides data analysis, interpretation, and therapy decision support.

Commercially available gene panels cover genes that are frequently mutated in different types of cancer. Yet the assessment of mutations in melanoma-specific genes and of additional parameters is required to determine if patients qualify for clinical trials, off-label and/or compassionate therapies. MelArray was designed to include the entire genomic information relevant for melanoma, allowing a precise diagnosis and the most optimal treatment selection.

Technical details: Dx captures > 4000 exons, 28 introns, and intergenic probes of heterozygous single nucleotide polymorphisms (SNPs), allowing the assessment of:

- mutations in 58 melanoma-relevant genes (see below)
- copy number variants (CNV) in a genome-wide manner and chromosomal aneuploidies
- total tumor mutational burden (TMB)
- HLA types

Sequenced and demultiplexed libraries are subjected to mutation calling. The resulting output is used to determine the mutation signature and TMB of a sample and to generate an annotated list of (potentially) pathogenic mutations. Intergenic probes are utilized to assess chromosomal aneuploidies as well as smaller deletions and duplications, known as CNVs. In parallel, HLA types will be assessed.

Specimen type: FFPE block or slides

Turnaround time: 10 working days

Key Points

- * only melanoma-specific gene panel
- * comprehensive examination of all melanoma-relevant genes and other parameters
- * only test combining the assessment of genes, genome wide CNVs, TMB and HLA type

Assessed genes:

ATM	AKT1	AKT2	AKT3	ALK	BAP1
BRAF	BRCA1	BRCA2	CBL	CCND1	CCND2
CCND3	CDK4	CDK6	CDKN2A	CTNNB1	EGFR
ERBB2	EZH2	FGFR1	FGFR2	FGFR3	GNA11
GNAI2	GNAQ	GNAS	HRAS	IDH1	JAK1
JAK2	KDR	KIT	KRAS	MAP2K1	MAP2K2
MAPK1	MAPK3	MET	MITF	NF1	NRAS
NTRK1	PDGFRA	PIK3CA	PPP6C	PTEN	PTPN11
RAC1	RAF1	RASA1	RASA2	RASA3	RB1
RET	ROS1	SF3B1	TP53		

Example of CNV reporting:

