Procedure standard di analitica

Table I. Proposed format for reporting genomic alterations detected through a comprehensive genomic panel. The results are categorized into SNV/INDEL, CNV, gene fusions, and genomic signatures, following a structured approach. Each alteration is reported with relevant details, including the affected gene, variant effect, variant classification and ESCAT level. Genomic signatures such as MSI and HRD/GIS are included with their corresponding values, reference thresholds, and ESCAT clas-silication where applicable.

SNV, INDEL variants

Example	Gene	Effect	Exon	Transcript ID	Nucleotid Variant	Protein Variant	Variant class	VAF	ESCAT
1	**EGFR**	Missense	21	NM_005228.5	c.2573T > G	p.(Leu858Arg)	Oncogenic	73%	IA
2	**EGFR**	Insertion in frame	20	NM_005228.5	c.2303_2311dup	c.p.(Ser768_Asp770dup)	Oncogenic	22%	IA
3	**BRCA1**	Splice acceptor variant	N/A	NM 007294.4	c.5075-2A > C	p.?	Oncogenic	52%	IA
4	**TERT**	Promoter variant	N/A	NM_198253.3	c.-124C>T	N/A	Oncogenic	14%	IV

Copy Number Variations

Example	Gene	Effect	Transcript ID	Region	Event	Variant class	Gone Copy Number	ESCAT
1	**MET**	Amplification	NM_001127500.3	7q31.2	gain	Oncogenic	20	IIB
2	**PTEN**	Deletion	NM_000314.8	1Oq23.31	loss	Oncogenic	0	IIIA

Fusions

Example	Gene	Effect	Transcript ID	Specific fusion	Phasity	Variant class	Read counts	ESCAT
1	**ALK**	Fusion	EML4: NM_019063; ALK: NM_004304.3	**EMLA:: ALK** (EML4 exon 13: ALK exon 20)	in frame	Oncogenic	800/188.000	IA
2	**NRG1**	Fusion	CD74: NM_004355.4; NRG1: NM_013956.5	**CD74:: NRG1** (CD74 exon 6::NRG1 exon 6)	in frame	Oncogenic	1890/166.000	IIB

Genomic Signature

Biomarker (example)	Score	Results	Threshold	ESCAT
MSI	**4**	MSS	18	N/A
HRD/GIS	**45/23**	Positive/High	42/16	IA

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