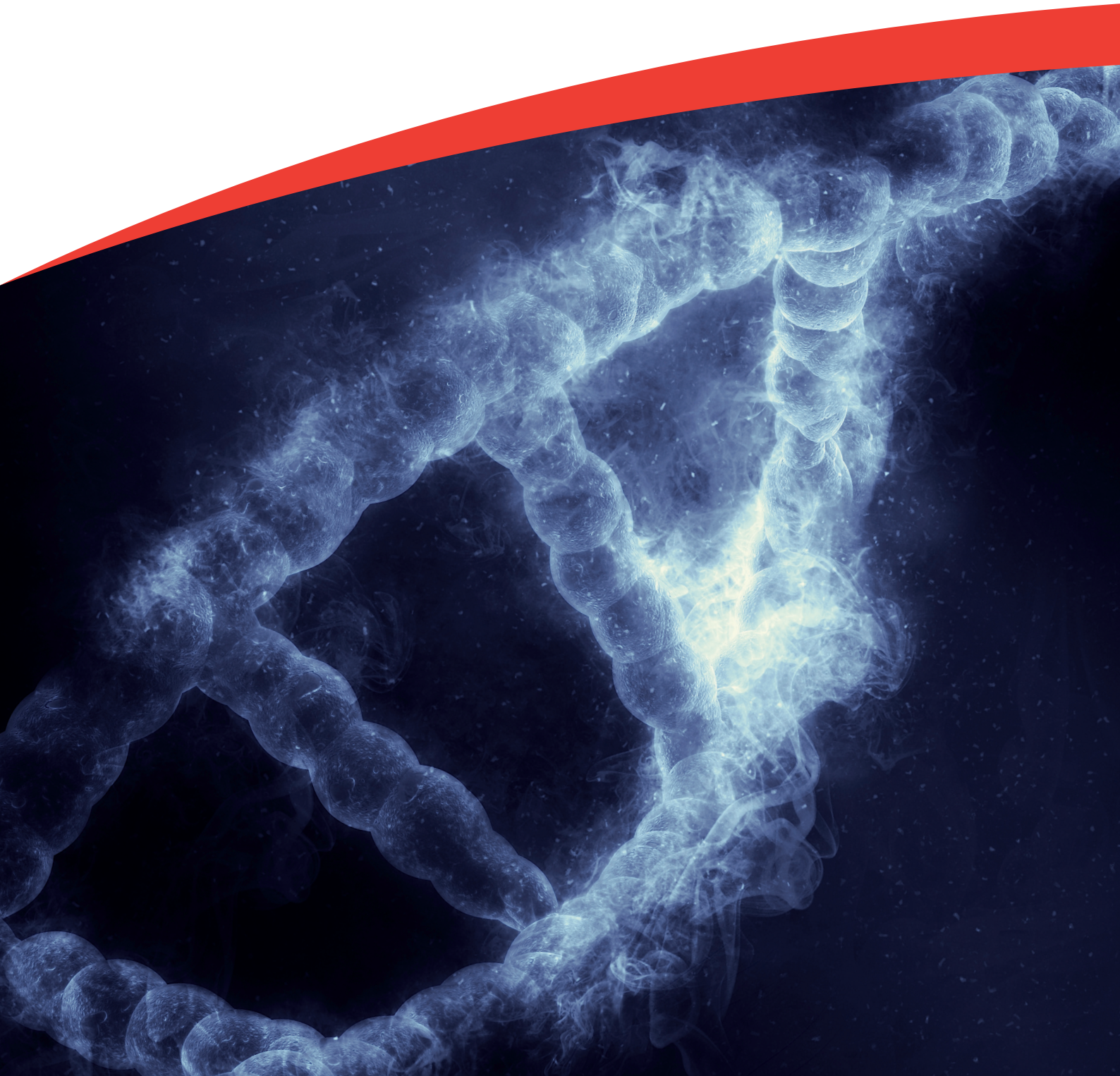
ALMAC

Partnering to Advance Human Health

Performance specification of the Almac TP53 clinical trial assay



Assay background

The Almac *TP53* clinical trial assay (CTA) is a qualitative Next Generation Sequencing (NGS) assay intended to detect genomic aberrations. It is validated for use with Formalin-Fixed Paraffin-Embedded (FFPE) solid tumour and lymph node samples, from patients with pan-cancer diseases, as well as fresh-frozen (FF), bone marrow aspirate (BMA) and peripheral blood (PB) samples from patients with haematological malignancies. The assay detects single nucleotide variants (SNVs) and small indels from DNA, enabling samples to be accurately classified as *Mutation Detected* or *Mutation Not Detected*.

Analytical validation

The assay has been analytically validated in accordance with Clinical Laboratory Improvement Amendment (CLIA) and New York State Department of Health Clinical Laboratory Evaluation Program (CLEP) requirements and is suitable for interventional use within clinical trials, such as for determining molecular eligibility. The assay is designed and developed in accordance with ISO 13485:2016 (Medical Devices - Quality Management Systems – Requirements for Regulatory Purposes) and ISO 14971:2019 (Medical Devices – Application of risk management to medical devices) and comprises a validated bioinformatics analysis pipeline and integrated quality control (QC) software which conform to IEC 62304:2006+A1:2015 (Medical Device Software – Software life-cycle processes) and ISO 14971 standards.

Performance specification

Performance parameter	FFPE	BMA/PB
Accuracy: Overall Percentage Agreement	99.99% (99.99% Lower 95% CI, 100.00% Upper 95% CI)	100.00% (99.99% Lower 95% CI, 100.00% Upper 95% CI)
Accuracy: Sensitivity	98.28% (95.05% Lower 95% CI, 99.41% Upper 95% CI)	100.00% (91.80% Lower 95% CI, 100.00% Upper 95% CI)
Accuracy: Specificity	100.00% (99.99 Lower 95% CI, 100.00% Upper 95% CI)	100.00% (99.99 Lower 95% CI, 100.00% Upper 95% CI)
Limit of Detection	SNVs: 3.134% VAF Insertions: 3.600% VAF Deletions: 3.107% VAF	
Precision: Repeatability	99.99% (99.99% Lower 95% CI, 100.00% Upper 95% CI)	100.00% (99.99% Lower 95% CI, 100.00% Upper 95% CI)
Precision: Reproducibility	99.99% (99.99% Lower 95% CI, 100.00% Upper 95% CI)	100.00% (99.99% Lower 95% CI, 100.00% Upper 95% CI)
Reportable range	Chr17 7572862 to 7573068 Chr17 7573880 to 7574088 Chr17 7576498 to 7576686 Chr17 7576821 to 7577181 Chr17 7577424 to 7577651 Chr17 7578143 to 7578655 Chr17 7579245 to 7579603 Chr17 7579605 to 7579633 Chr17 7579635 to 7579642 Chr17 7579661 to 7579750 Chr17 7579836 to 7580028 Chr17 7590645 to 7590913	

Assay coverage

Isoform ID	Transcript ID	CDS ID	% CDS Covered
Isoform a	NM_000546.5	CCDS11118	100
Isoform a	NM_001126112.2	CCDS11118	100
Isoform c	NM_001126113.2	CCDS45605	100
Isoform b	NM_001126114.2	CCDS45606	100
Isoform d	NM_001126115.1	CCDS73966	100
Isoform e	NM_001126116.1	CCDS73968	100
Isoform f	NM_001126117.1	CCDS73967	100
Isoform g	NM_001126118.1	CCDS73969	100
Isoform h	NM_001276695.2	CCDS73970	100
Isoform i	NM_001276696.2	CCDS73971	100
Isoform j	NM_001276697.2	CCDS73963	100
Isoform k	NM_001276698.2	CCDS73965	100
Isoform l	NM_001276699.2	CCDS73964	100
Isoform g	NM_001276760.2	CCDS73969	100
Isoform g	NM_001276761.2	CCDS73969	100

The full analytical validation report is available on request from Almac Diagnostic Services

Visit the website