

Specialist Pathology Laboratory
The Royal Marsden NHS Foundation Trust
Downs Road, Sutton, SM2 5PT

rmh-tr.cytogenetics@nhs.net
rmh-tr.moleculardiagnosics@nhs.net



Patient Name:	WEBSERVER VARIANT, Testing	Authorising Clinician Andrew JONES, Consultant
Patient DOB:	14/10/1994	
Patient Gender:	Female	
NHS Number:	Missing	
RMH MRN:	510804277	
External Patient ID:		External requesting Location:

Multi-target NGS panel small variant (FH, SDHA, SDHB, SDHC, SDHD, VHL, ELOC (TCEB-1), (Final result)

Test Directory Code
M18.2

Interpretation

Please note the tumour assessment has been performed by the referral histopathology laboratory. The request form accompanying this sample indicates tumour nuclei >70% and high cellularity.

Likely pathogenic/pathogenic variants in ATR, BAP1 and VHL were detected. A TERT promoter variant C228T was also detected.

Please correlate with other clinical findings.

No variants detected in following genes: FH, SDHA, SDHB, SDHC, SDHD, TCEB1, TSC1, TSC2, MET, BRAF, TFE3.

VARIANTS

Tier 1 and Tier 2 Clinically Relevant Variants

Gene	DNA Change	Protein Change	Molecular Consequence	VAF	Supporting Reads	Transcript
ATR	c.2320dup	p.(Ile774AsnfsTer3)	Duplication	9.9%	75	NM_001184.4
BAP1	c.685_703del	p.(Asn229ProfsTer14)	Deletion	19.3%	99	NM_004656.4
TERT	c.-124C>T		Substitution	14.5%	72	NM_198253.3
VHL	c.349T>A	p.(Trp117Arg)	Substitution	22.6%	157	NM_000551.4

No oncogenic variants detected: FH, SDHA, SDHB, SDHC, SDHD, TCEB1, TSC1, TSC2, MET, BRAF, TFE3.

This custom capture panel is property of the Royal Marsden NHS Foundation Trust and has undergone clinical validation for the genomic regions listed in the Solid DNA NGS Panel Specification, available at <https://www.royalmarsden.nhs.uk/services/genomics/nhs-healthcare-providers>. Full details of panel coverage, assay performance characteristics, and test limitations are also available at this location. For further information, please contact rmh-tr.moleculardiagnosics@nhs.net.

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Reported by

George, Mr. Andrew (Clinical Scientist)

Authorised by

Brittany Mills (Clinical Scientist)

26SP-159M0049: Resulted by Clinical Genomics. Verified: 26/9/2026 12:26.

External Specimen ID:

See attached media documents:

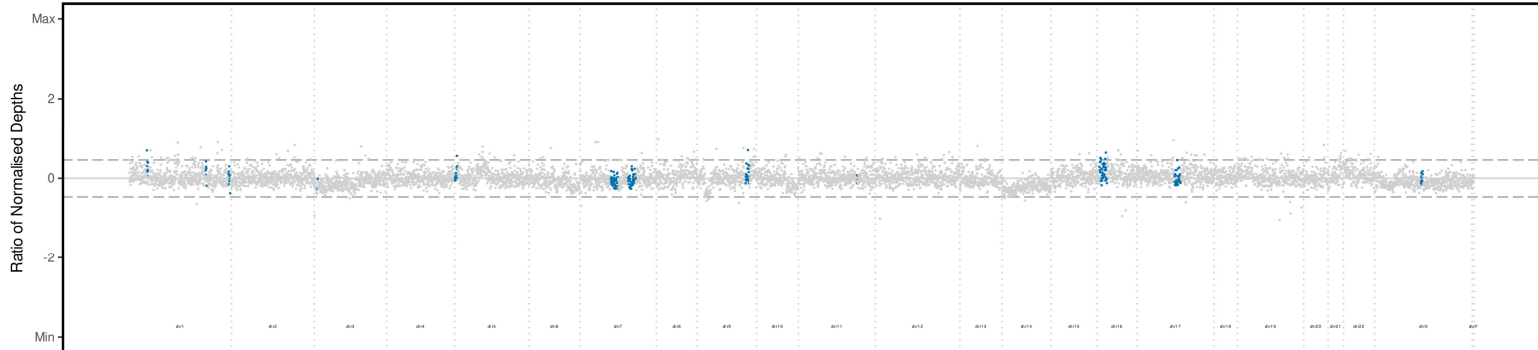
Document 26/9/2026 12:23 Rmh_Interface_Webserver_Icr_V2

Resulting Labs

Clinical Genomics	RMH CLINICAL GENOMICS, A UKAS accredited medical laboratory No.8097, Downs Rd, Sutton, Surrey, SM2 5PT	020 8915 6565
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Specimen Details

	Type	Source	Collected	Received
26SP-159M0049	FFPE Tissue		5/6/2026 18:06	8/6/2026 10:29



The copy number plot above presents copy number variation in sequential chromosomal order. Genes within the requested virtual panel are highlighted in blue.