



MACQUARIE UNIVERSITY CANCER BIOBANK

MQCB00206A REPORT

30 JUNE 2025

CLINICAL SUMMARY

Sex: Female
Age: 68
Pre-operative radiotherapy: Unknown

HISTOPATHOLOGY

Specimen type: Spleen
Tumour site: Spleen
Tumour type: Melanoma
Tumour stage: Metastatic melanoma

NEXT GENERATION GENOME/TRANSCRIPTOME DATA

Provider: Australian Genome Research Facility - 150bp paired end
Data yield: Germline DNA: 34,765,001 paired end, 10.50 Gbp
Tumour DNA: 60,639,485 paired end, 18.31 Gbp
Tumour RNA: 46,399,163 paired end, 14.01 Gbp

95% Tumour Content

Top pathogenic somatic mutations			
Gene	Alteration	Allele Frequency	Function
KRAS	c.185_186insAAAGTACAGTGCAATGAG p.E62_E63insKYSAMR	63%	Loss
KRAS	c.188_189insCCA p.E63delinsDQ	78%	Loss
TERT	c.-124 C>T	32%	Gain
MEN1	c.783+2 T>G	44%	

No Tumour Dissociates were banked from this sample