



MACQUARIE UNIVERSITY CANCER BIOBANK

MQCB00205A REPORT

30 JUNE 2025

CLINICAL SUMMARY

Sex: Female
Age: 74
Pre-operative radiotherapy: Unknown

HISTOPATHOLOGY

Specimen type: Brain
Tumour site: Parietal Lobe
Tumour type: Melanoma
Tumour stage: Metastatic Melanoma

NEXT GENERATION GENOME/TRANSCRIPTOME DATA

Provider: Australian Genome Research Facility - 150bp paired end
Data yield: Germline DNA: 25,703,930 paired end, 7.76 Gbp
Tumour DNA: 90,601,655 paired end, 27.36 Gbp
Tumour RNA: 46,837,024 paired end, 14.14 Gbp

70% Tumour Content

Top pathogenic somatic mutations			
Gene	Alteration	Allele Frequency	Function
BRAF	V600E	51%	Gain
TP53	R248W	77%	Loss/Gain
ATM	c.7280_7281dupTT p.R2428fs*13	24%	Loss
ATM	c.7277_7278delTC p.L2426fs*2	24%	Loss
TERT	c.-124C>T	34%	Gain
CDKN2A	c.341C>T p.P114L	87%	Loss

No Tumour Dissociates were banked from this sample