

MACQUARIE UNIVERSITY CANCER BIOBANK

MQCB00217A REPORT

10 July 2025

CLINICAL SUMMARY

Sex: Male
Age: 59
Pre-operative radiotherapy:

HISTOPATHOLOGY

Specimen type: Brain
Tumour site: Right Occipital
Tumour type: Melanoma
Tumour stage: Stage IV
Specimen tumour content: 80%

NEXT GENERATION GENOME/TRANSCRIPTOME DATA

Provider: Australian Genome Research Facility - 150bp paired end
Data yield: Germline DNA: 25,797,869 paired end, 7.79 Gbp
Tumour DNA: 75,314,667 paired end, 22.75 Gbp
Tumour RNA: 46,741,161 paired end, 14.12Gbp

Top pathogenic somatic mutations			
Gene	Alteration	Allele Frequency	Function
NRAS	c.181C>A Q61K	42%	Gain
TP53	p.733G>A G245S	90%	Loss
B2M	c.227delT p.F76fs*27	97%	Loss

FLOW CYTOMETRY TUMOUR DISSOCIATE ANALYSIS

