

MACQUARIE UNIVERSITY CANCER BIOBANK

MQCB00221A REPORT

16 July 2025

CLINICAL SUMMARY

Sex: Female
Age: 36

HISTOPATHOLOGY

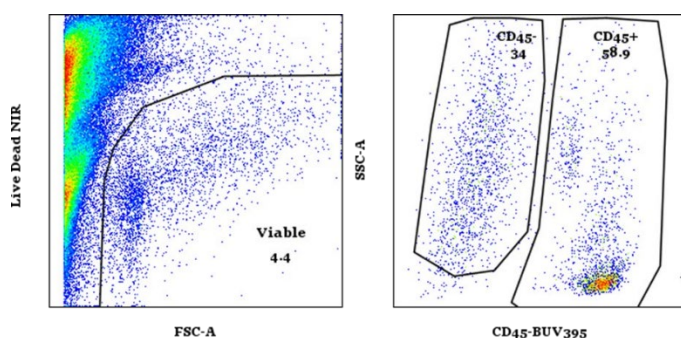
Specimen type: Brain
Tumour site: Right Posterior Frontal Lobe
Tumour type: Melanoma
Tumour stage: Stage IV
Specimen tumour content: 40%

NEXT GENERATION GENOME/TRANSCRIPTOME DATA

Provider: Australian Genome Research Facility - 150bp paired end
Data yield: Germline DNA: 27,497,528 paired end, 8.30 Gbp
Tumour DNA: 82,601,048 paired end, 24.95 Gbp
Tumour RNA: 50,051,074 paired end, 15.12 Gbp

Top pathogenic somatic mutations			
Gene	Alteration	Allele Frequency	Function
BRAF	c.1799T>A p.V600E	40%	Gain
NF1	c.2059C>G p.L687V	50%	Loss
TERT	c.-146C>T	33%	Gain
ARID2	c.3754G>T p.E1252*	68%	Loss
PTEN	c.183_193dupTAAAAACCATT p.Y65fs*38	56%	Loss

FLOW CYTOMETRY TUMOUR DISSOCIATE ANALYSIS



Cell Type	(%) of subset
Viable Cells	4.4
Viable Cells>CD45-	3.4
Viable Cells>CD45+	58.9
Viable Cells>CD45+>B Cells	0.8
Viable Cells>CD45+>CD4+	20.9
Viable Cells>CD45+>CD8+	35.8