MACQUARIE UNIVERSITY CANCER BIOBANK

MQCB00077A REPORT

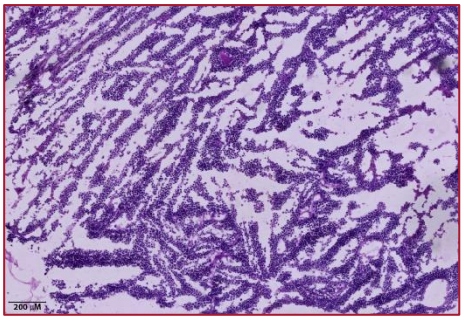
14 OCTOBER 2022

CLINICAL SUMMARY

Sex: Male
Age: 79
Pre-operative radiotherapy: No

HISTOPATHOLOGY

Specimen type: Brain
Tumour site: Right Parietal
Tumour type: Metastatic Melanoma, SOX10 and Melan-A positive
Tumour stage: Metastatic
No Mutations in BRAF, KIT or NRAS genes



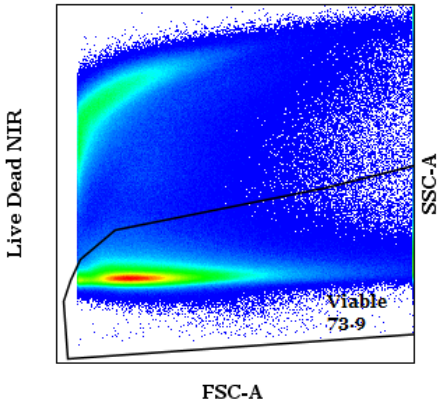
75 – 80% Tumour Content

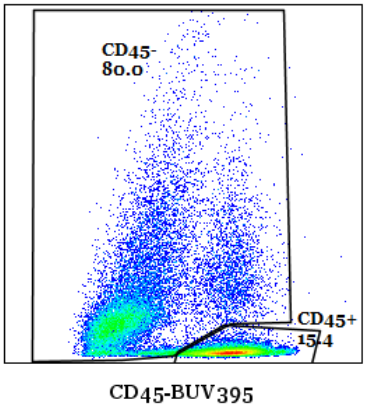
NEXT GENERATION GENOME/TRANSCRIPTOME DATA

Provider: Australian Genome Research Facility - 150bp paired end
Data yield: Germline DNA: 538,499,871 paired end, 162.63 Gbp
Tumour DNA: 944,358,377 paired end, 285.21 Gbp
Tumour RNA: 55,979,531 paired end, 16.91 Gbp

Top pathogenic somatic mutations			
Gene	**Alteration**	**Allele Frequency**	**Function**
TERT	c.-146C>T	47%	Gain
KMT2A	c.10951C>T p.Q3651*	49%	Loss
POLE	c.3358C>T p.Q1120*	49%	Loss
STAG2	c.2139C>A p.Y713*	22%	Loss

FLOW CYTOMETRY TUMOUR DISSOCIATE ANALYSIS





Cell Type	(%) of subset
Viable cells	73.9
Viable>CD45-	80.0
Viable>CD45+	15.4
Viable>CD45+>B cells	1.7
Viable>CD45+>CD4+/Foxp3-	11.0
Viable>CD45+>Tregs+	2.6
Viable>CD45+>CD8+	36.7
Viable>CD45+>Monocytes/Macrophages	13.1