

MACQUARIE UNIVERSITY CANCER BIOBANK

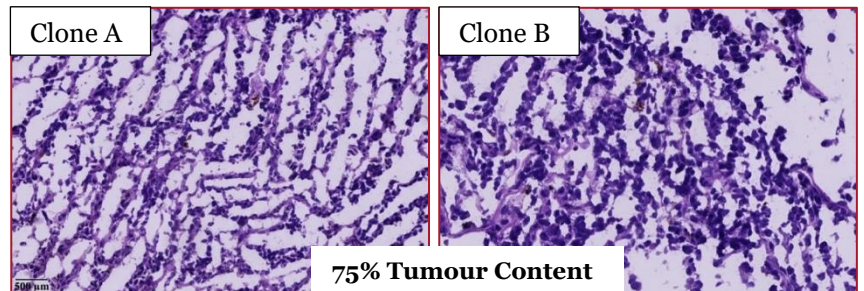
MQCB00196A CLONE A AND CLONE B REPORT

14 NOVEMBER 2022

CLINICAL SUMMARY

Right superior fundus mass lesion

HISTOPATHOLOGY



Specimen type:	Enucleation, Right eye
Tumour Size:	11mm (basal diameter) x 8mm (thickness)
Tumour type:	Choroidal Melanoma, Grade 2
Tumour stage:	pT2a (AJCC, 8 th edition)
Cell type:	Spindled, heavily pigmented (region/clone A) Predominantly epithelioid, amelanotic (region/clone B)
Molecular Studies:	BAP1 retained, chromosome 3 disomy, chromosome 6p gain, chromosome 6q disomy, chromosome 8q disomy, segmental loss of chromosome 17p, loss of chromosome Y
Immunohistochemistry:	CD68+ high (A); moderate (B) CD163+ moderate (A), moderate to high (B)

NEXT GENERATION GENOME/TRANSCRIPTOME DATA

Provider:	Australian Genome Research Facility - 150bp paired end
Data yield:	Germline DNA: 519,446,425 paired end, 156.87 Gbp
	Tumour DNA: Clone A: 792,884,149 paired end, 239.44 Gbp
	Clone B: 1,202,016,740 paired end, 360.01 Gbp

Top pathogenic somatic mutations			
Gene	Alteration	Allele Frequency	Function
GNAQ	c.626A>C, p.Q209P	Clone A: 55%; Clone B: 48%	Gain

No Tumour Dissociates were banked from this sample