

MACQUARIE UNIVERSITY CANCER BIOBANK

MQCB00207A REPORT

30 JUNE 2025

CLINICAL SUMMARY

Sex: Male
Age: 69
Pre-operative radiotherapy: Unknown

HISTOPATHOLOGY

Specimen type: Knee Joint
Tumour site: Right Popliteal Fossa
Tumour type: Melanoma
Tumour stage: Metastatic Melanoma

NEXT GENERATION GENOME/TRANSCRIPTOME DATA

Provider: Australian Genome Research Facility - 150bp paired end
Data yield: Germline DNA: 28,449,474 paired end, 8.59 Gbp
 Tumour DNA: 70,839,008 paired end, 21.39 Gbp
 Tumour RNA: 37,360,243 paired end, 11.28 Gbp

75% Tumour Content

Top pathogenic somatic mutations			
Gene	Alteration	Allele Frequency	Function
IDH1	c.394C>T p.R132C	26%	Gain
NF1	c.5431G>T p.E1811*	63%	Loss
NRAS	c.183A>T p.Q61H	23%	Gain
TP53	c.535C>T p.H179Y	44%	Loss
APC	c.3367C>T p.Q1123*	47%	Loss
RNF43	c.433C>T p.R145*	61%	Loss
TERT	c.-146C>T	45%	Gain
TEX15	c.9474G>A p.W3158*	48%	Loss

No Tumour Dissociates were banked from this sample