



# MACQUARIE UNIVERSITY CANCER BIOBANK

MQCB00072A REPORT

07 OCTOBER 2022

## CLINICAL SUMMARY

**Sex:** Female  
**Age:** 41  
**Pre-operative radiotherapy:** Nil neoadjuvant

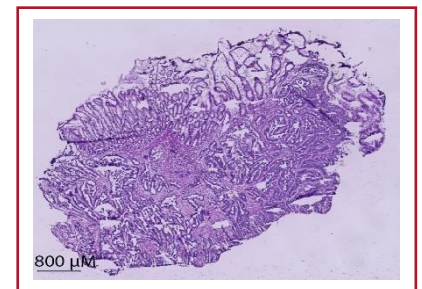
## HISTOPATHOLOGY

**Specimen type:** Colon  
**Tumour site:** Anterior Resection Sigmoid  
**Tumour type:** Adenocarcinoma  
**Tumour stage:** pT3, N1a Stage IIIB, CDX2 Positive.  
 Normal MLH1, PMS2, MSH2 and MSH6  
 MMR proficient, high microsatellite instability unlikely



## NEXT GENERATION GENOME/TRANSCRIPTOME DATA

**Provider:** Australian Genome Research Facility - 150bp paired end  
**Data yield:** Germline DNA: 500,219,740 paired end, 151.07 Gbp  
 Tumour DNA: 1,100,753,145 paired end, 332.42 Gbp  
 Tumour RNA: 66,451,693 paired end, 20.07 Gbp



60 – 75% Tumour Content

Top pathogenic somatic mutations			
Gene	Alteration	Allele Frequency	Function
ATM	c.1009C>T, p.R337C	41%	Loss
APC	c.4141_4142delCC, p.P1381fs*4	66%	Loss
TP53	c.375G>A, p.T125T (splicing variant)	36%	Loss

*No Tumour Dissociates were banked from this sample*