

## Case 9

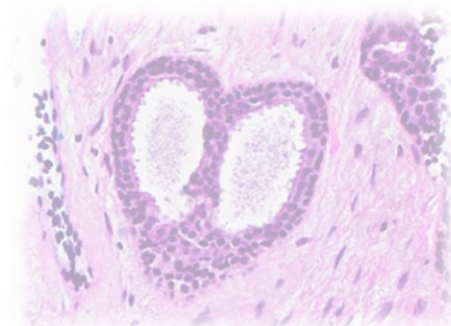
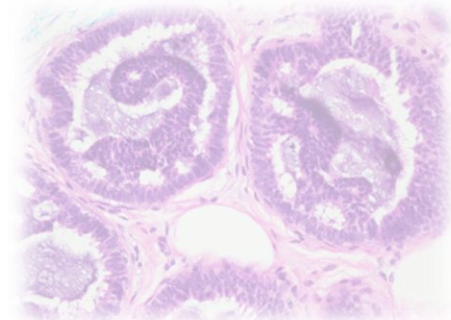
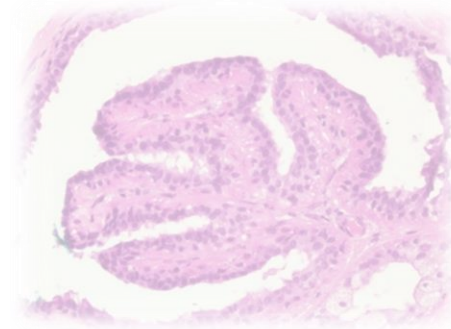
67 year old Thai female.

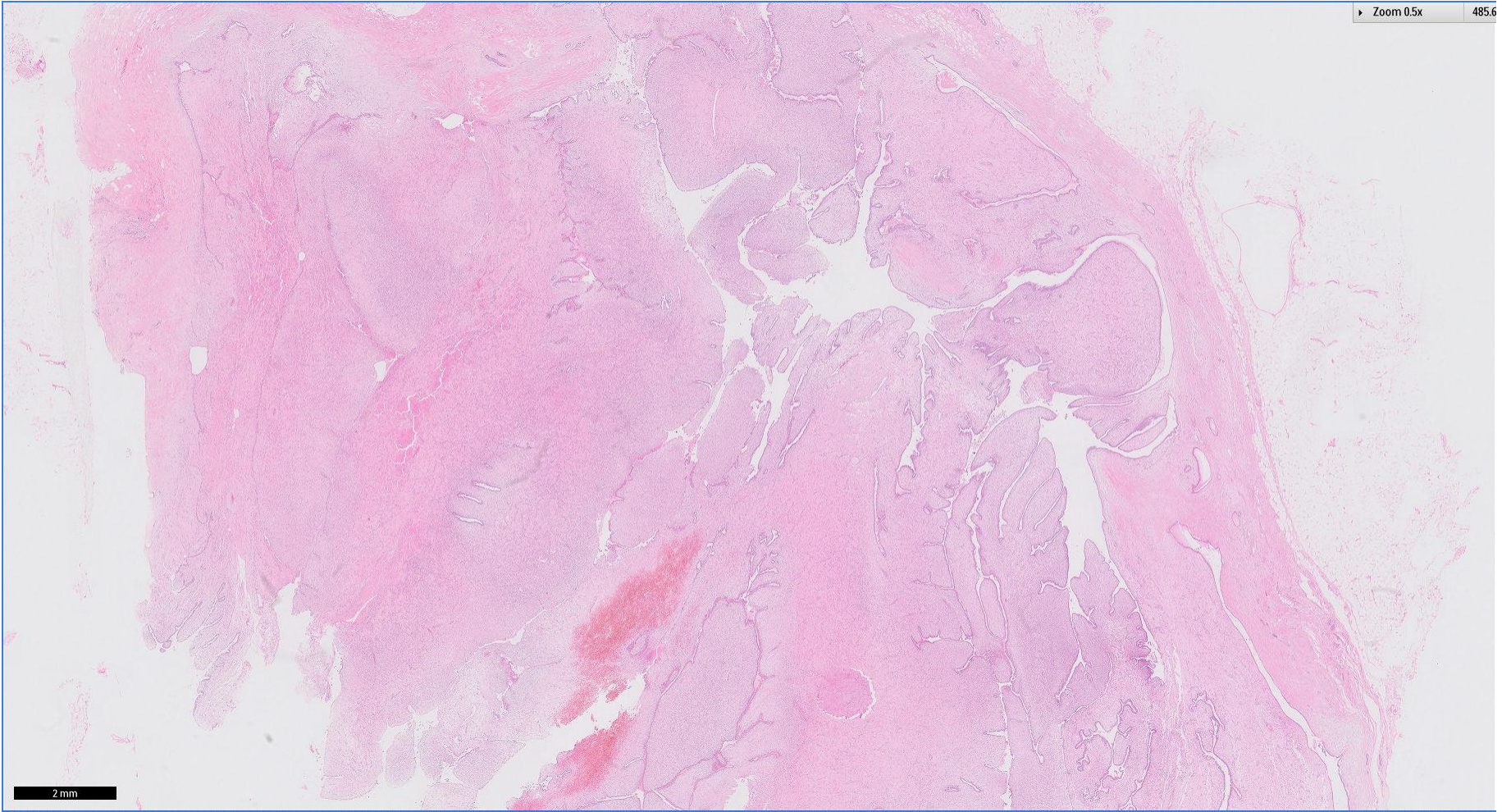
Right breast palpable mass.

Radiology showed an 8.7 x 6.7cm well-defined macrolobulated mass with calcifications at the right breast UOQ, classified as BIRADS 4.

Section provided of the mass.

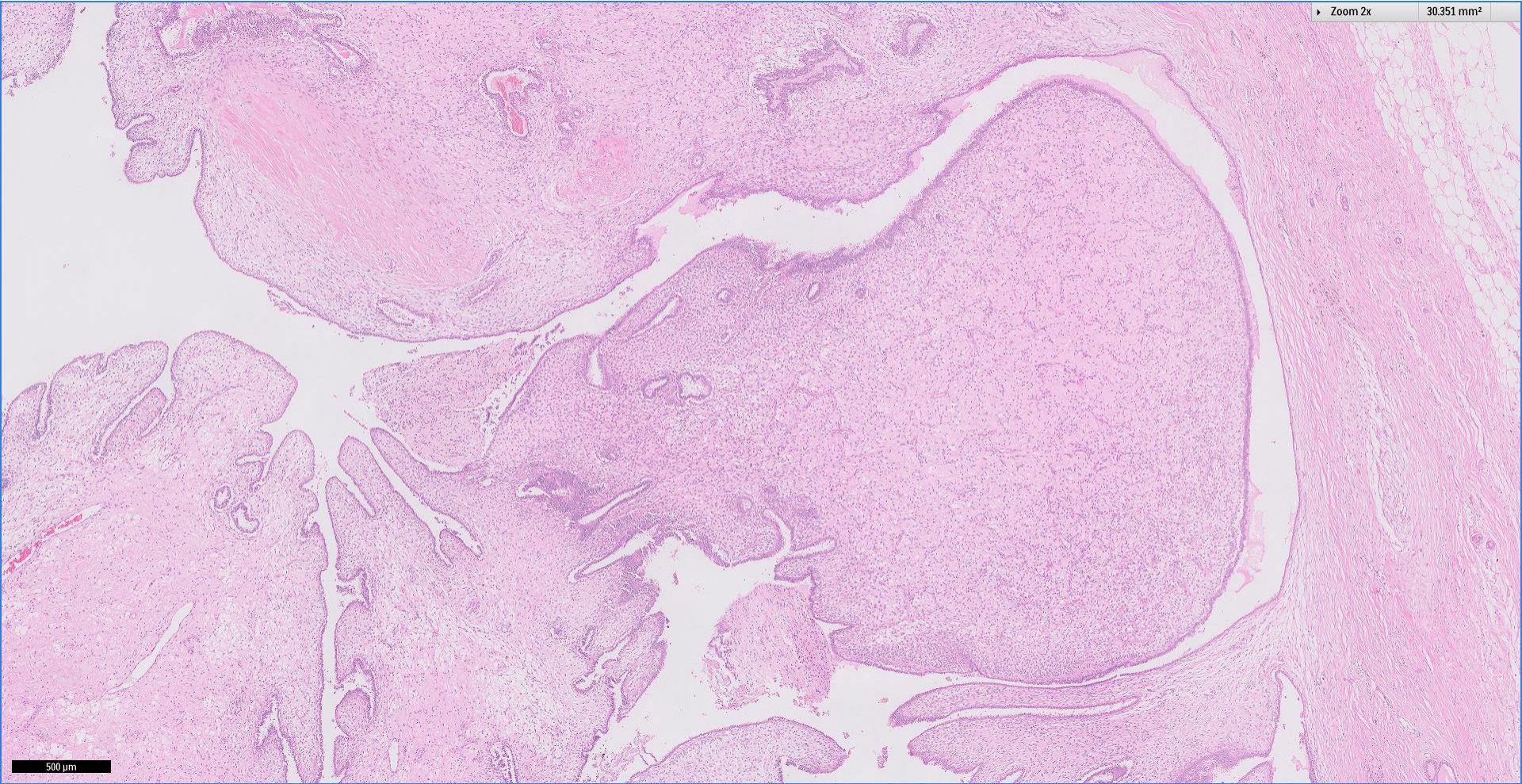
*Contributed by Dr Kittisak Wongchansom,  
Bangkok Thailand*





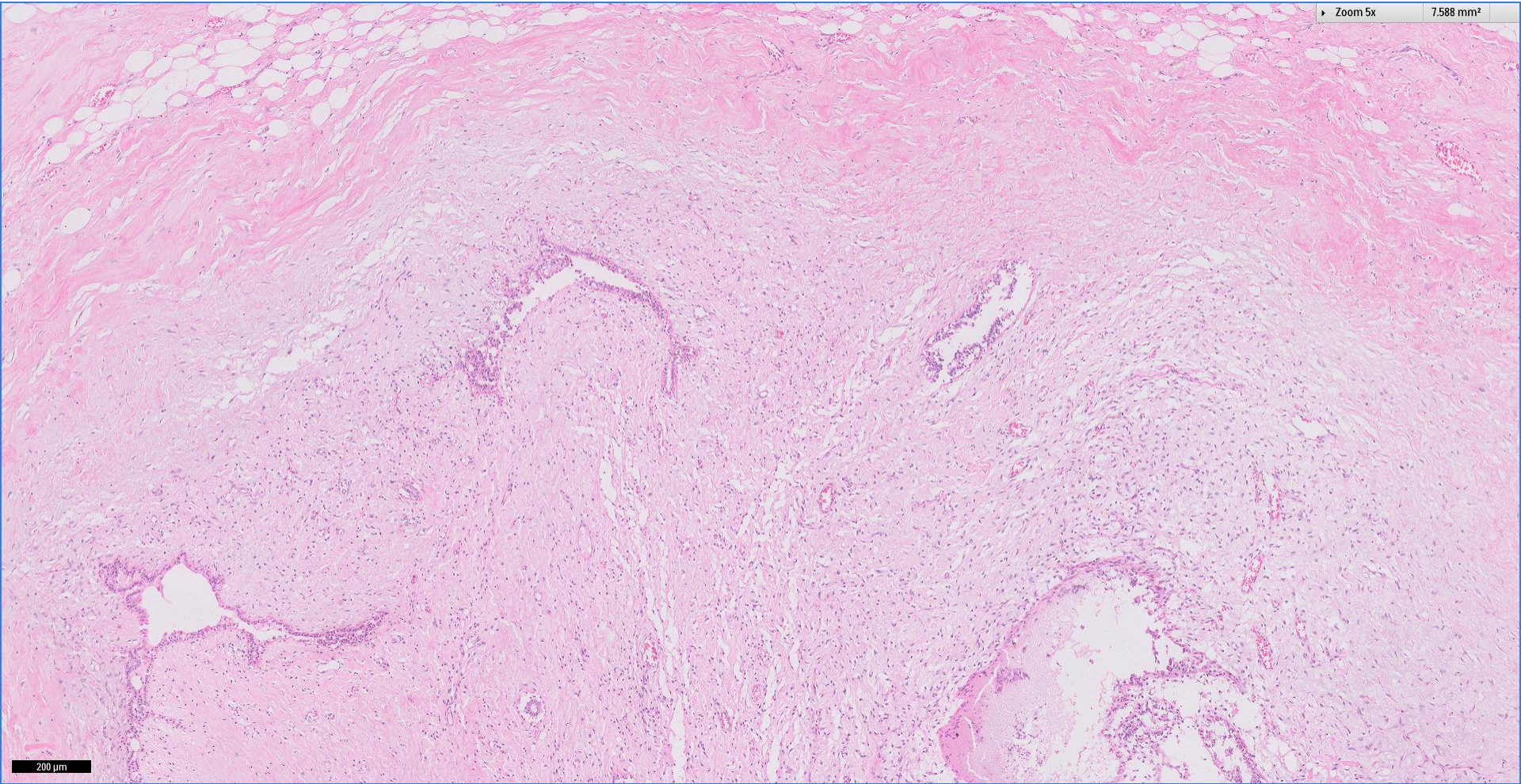
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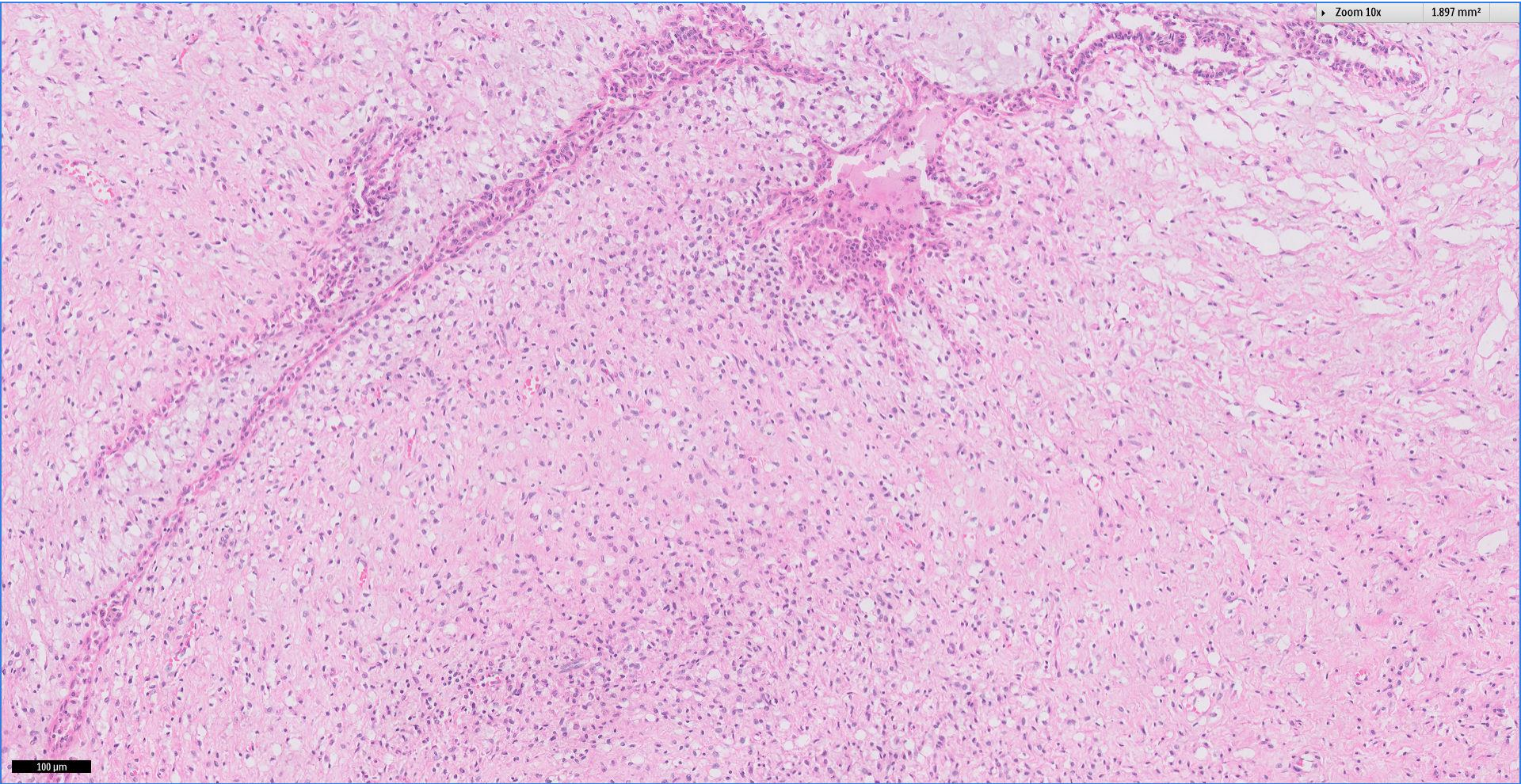




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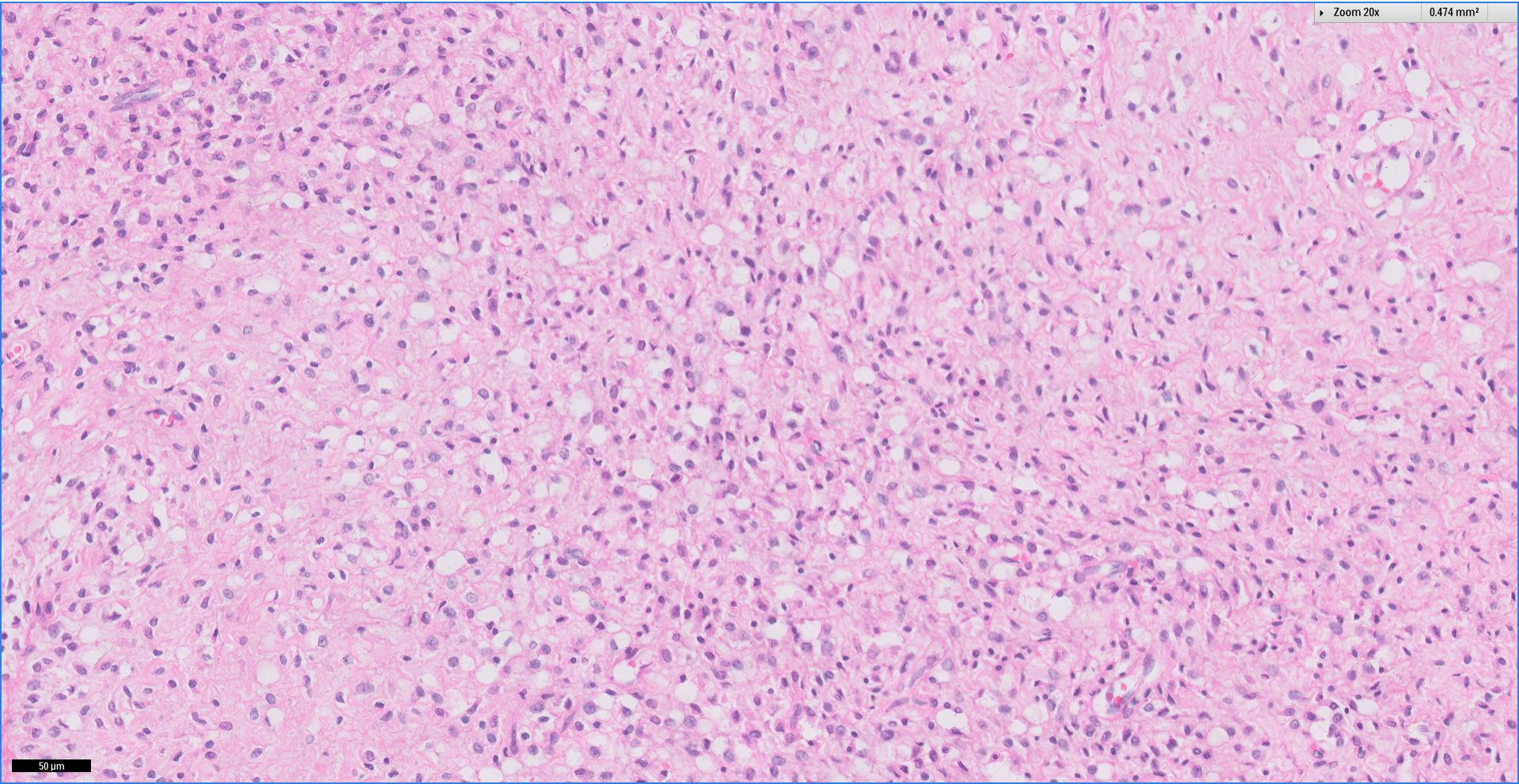


100 µm



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50  $\mu$ m

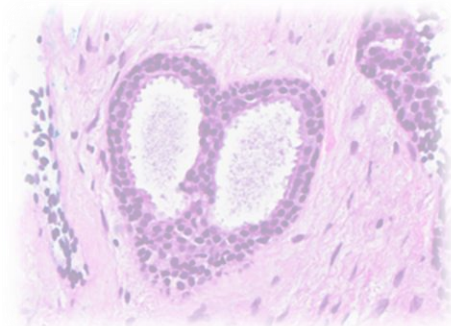
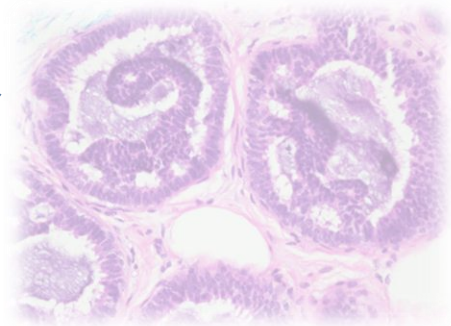
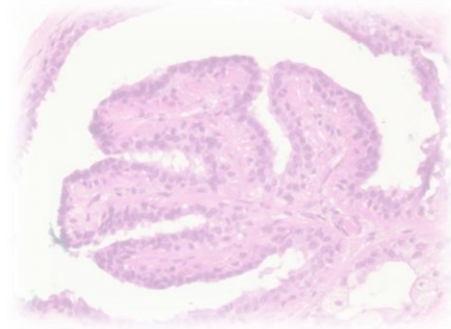


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## Additional findings



# Genomic results

**Sample ID:** 61213013/11

**16-gene FEB panel results:** *MED12*, *TERT* promoter, *RARA*, *SETD2* and *KMT2D* mutations

**Remark:** Consistent with the borderline grade

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Gene	Type of Mutation	Variant Allele Frequency
MED12	Missense	31.78
TERT	Promoter mutation	34.79
RARA	Missense	34.30
SETD2	Missense	39.09
KMT2D	Missense	52.42



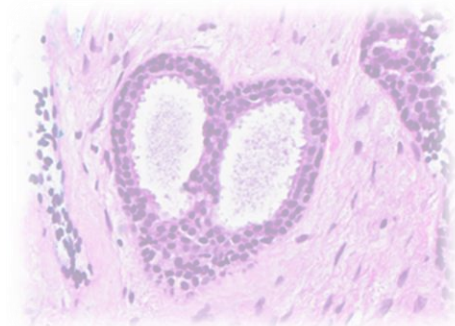
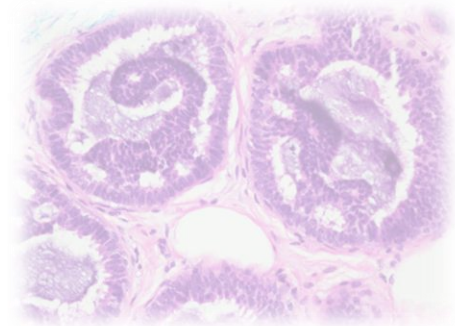
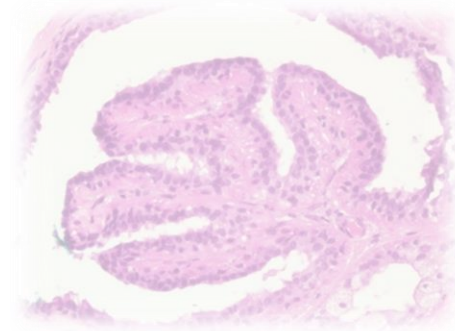
# Comparison of 16 gene panel results between borderline and malignant phyllodes tumours

Gene symbol	Borderline (n = 117)	Malignant (n = 54)	p-value
MED12	58 (50%)	20 (37%)	0.1398
TERT	71 (61%)	25 (46%)	0.0975
KMT2D	19 (16%)	10 (19%)	0.8268
RARA	22 (19%)	4 (7%)	0.0668
FLNA	26 (22%)	10 (19%)	0.6881
SETD2	18 (15%)	6 (11%)	0.6362
TP53	10 (9%)	9 (17%)	0.1245
RB1	8 (7%)	6 (11%)	0.375
NF1	6 (5%)	6 (11%)	0.1982
PTEN	1 (1%)	6 (11%)	0.0043 *
PIK3CA	5 (4%)	3 (6%)	0.7085
EGFR	8 (7%)	2 (4%)	0.5074
BCOR	6 (5%)	3 (6%)	>0.9999
ERBB4	2 (2%)	2 (4%)	0.5915
MAP3K1	5 (4%)	0 (0%)	0.1808
IGF1R	3 (3%)	1 (2%)	>0.9999

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# Diagnosis, case 9

- Right breast mass:  
Borderline phyllodes tumour, 8cm.



# Questions

- *Is there round cell liposarcoma in this phyllodes tumour?*

*No*

- *Is the diagnosis of liposarcoma indicative of a malignant phyllodes tumour?*

*No*



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# Liposarcoma in phyllodes tumours

- Although liposarcoma was traditionally regarded as a malignant heterologous component, there is evidence to suggest that metastatic risk is low when well-differentiated liposarcoma occurs as the sole heterologous element in a phyllodes tumour.
- These abnormal adipocytes within phyllodes tumours lack *MDM2* or *CDK4* amplifications, in contrast to extramammary well-differentiated liposarcoma.
- Therefore, it is recommended that a diagnosis of malignant phyllodes tumour is not made based purely on the finding of well-differentiated liposarcoma, but also on the basis of other stromal features.
- Rare pleomorphic liposarcomas in phyllodes tumours have shown more-adverse outcomes.
- Although myxoid liposarcoma has been described in phyllodes tumours, the lack of associated characteristic molecular aberrations calls into question its true existence within phyllodes tumours.

WHO 2019

# Correlation of phyllodes tumour grades with genomic aberrations, *J Pathol.* 2019 Aug 8.

- A significantly higher number of genetic aberrations was observed **with increasing grade of PTs**, in particular with regard to ***TERT*** promoter (32% vs 61% vs 46%,  $p < 0.0001$ ), ***FLNA*** (13% vs 22% vs 19%,  $p = 0.0289$ ), ***TP53*** (3% vs 9% vs 17%,  $p = 0.0003$ ) and ***RB1*** (3% vs 7% vs 11%,  $p = 0.0297$ ) for benign, borderline and malignant PTs respectively.
- ***MED12*** mutations on the other hand significantly decreased as the PTs progressed (62% vs 50% vs 37%,  $p = 0.0006$ ).
- A comparison between **borderline and malignant PTs** did not show significant differences, apart from ***PTEN*** (1% vs 11%,  $p = 0.0043$ ).

*Thank You*