## Case 20

## Adult female with a breast lump.

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## IAD

International Academy of Pathology








# Immunohistochemistry ~ 

Cytokeratins negative SMA, desmin focally positive
$75 \%$ of 200 nuclei scored showed one copy of RB1 gene and one copy of the control probe at 13q34, suggesting monosomy 13.
Note: RB1 gene is labelled with Spectrum Orange while the control probe is labelled with Spectrum Green. (red dots are the RB1 gene and the green dots are the control probe at 13q34)


Diagnosis: Myofibroblastoma

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## Myofibroblastoma

- Benign tumour of the mammary stroma composed of fibroblasts and myofibroblasts.
- Most common site is the breast, but may occur in extramammary sites (inguinal/groin area, vulva, perineum, scrotum).
- Clinical presentation as a slow-growing, painless, nontender mass.
- Males and females equally affected.
- Hormonal aetiology ~
- Expression of hormone receptors
- Association with gynaecomastia, PASH


## Myofibroblastoma

- Derived from a precursor cell of the mammary stroma.
- Genetic monoallelic or biallelic deletions of 13q (13q14), sometimes in combination with $16 q$ monosomy.
- 13q deletion results in the loss of RB1 expression on immunohistochemistry.
- Morphological patterns ~
- Lipomatous
- Myxoid
- Fibrous/collagenous
- Epithelioid/deciduoid
- Palisading/Schwannian-like


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## Myofibroblastoma

- Essential criteria~
- Well-circumscribed margins
- Mesenchymal tumour
- No significant cytologic atypia
- Low mitotic count
- Short interlacing fascicles
- Desirable criteria ~
- IHC positivity for CD34, ER/PR/AR, desmin
- FISH 13q14 deletion


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