CASE OF THE MONTH

DR. AJIT PRATAP SINGH PANAYCH
SENIOR RESIDENT
DEPARTMENT OF PATHOLOGY
RGCIRC

CASE REPORT:

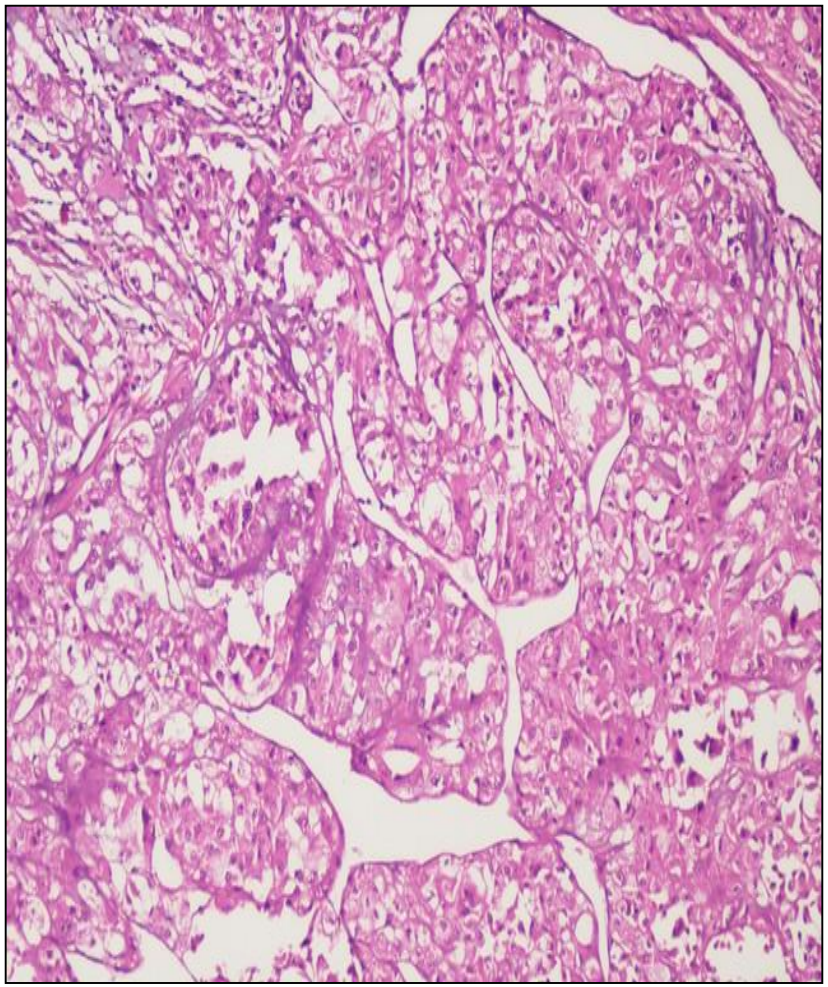
- 70 years old/ male
- C/O nodular swelling over lower chest wall for one month, underwent excision
- Subsequent PET CT of whole body showed an FDG avid circumferential mural thickening with mass formation involving the caecum and adjacent ascending colon with soft tissue thickening
- Underwent biopsy from the colonic mass followed by hemicolectomy

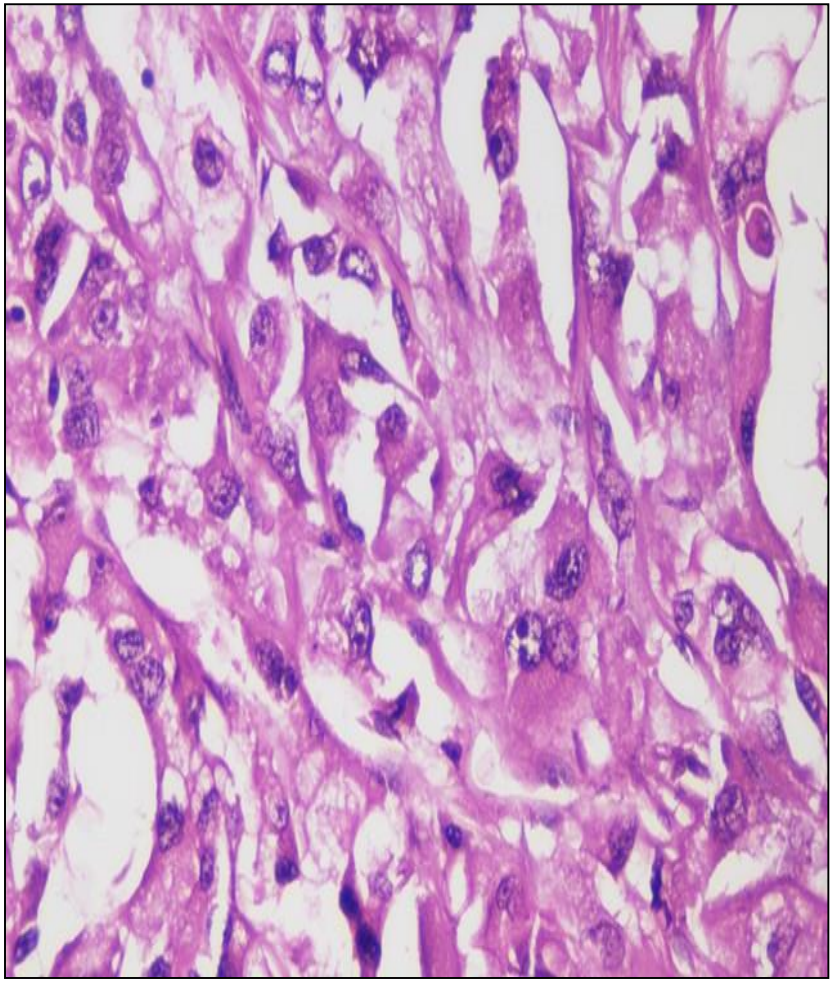
CHEST WALL MASS

MICROSCOPY:

- Tumor was composed of pleomorphic epithelioid cells arranged in sheets, islands & alveolar pattern with ill defined boundaries located in deep dermis

- Many giant cells were seen along with areas of necrosis





Histomorphology of the chest wall swelling :

Pleomorphic epithelioid cells arranged in sheets, islands & alveolar pattern with ill defined boundaries.

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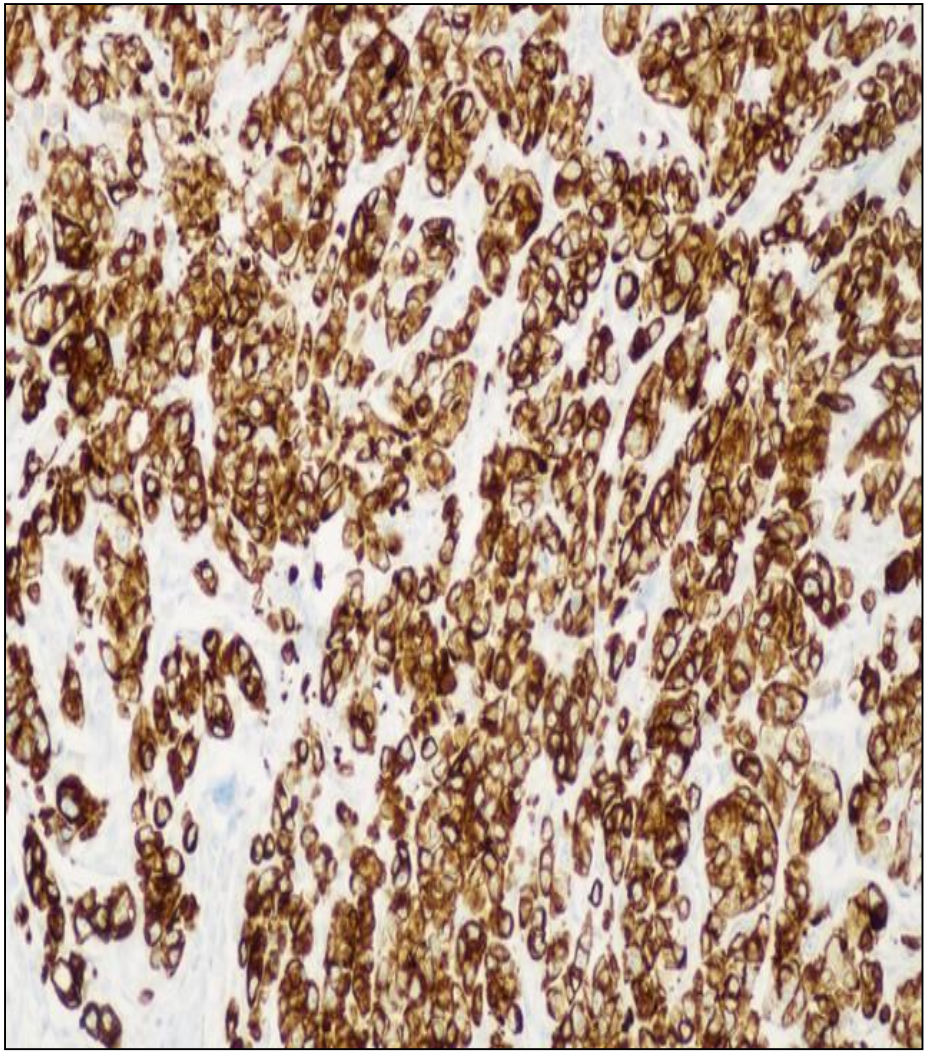
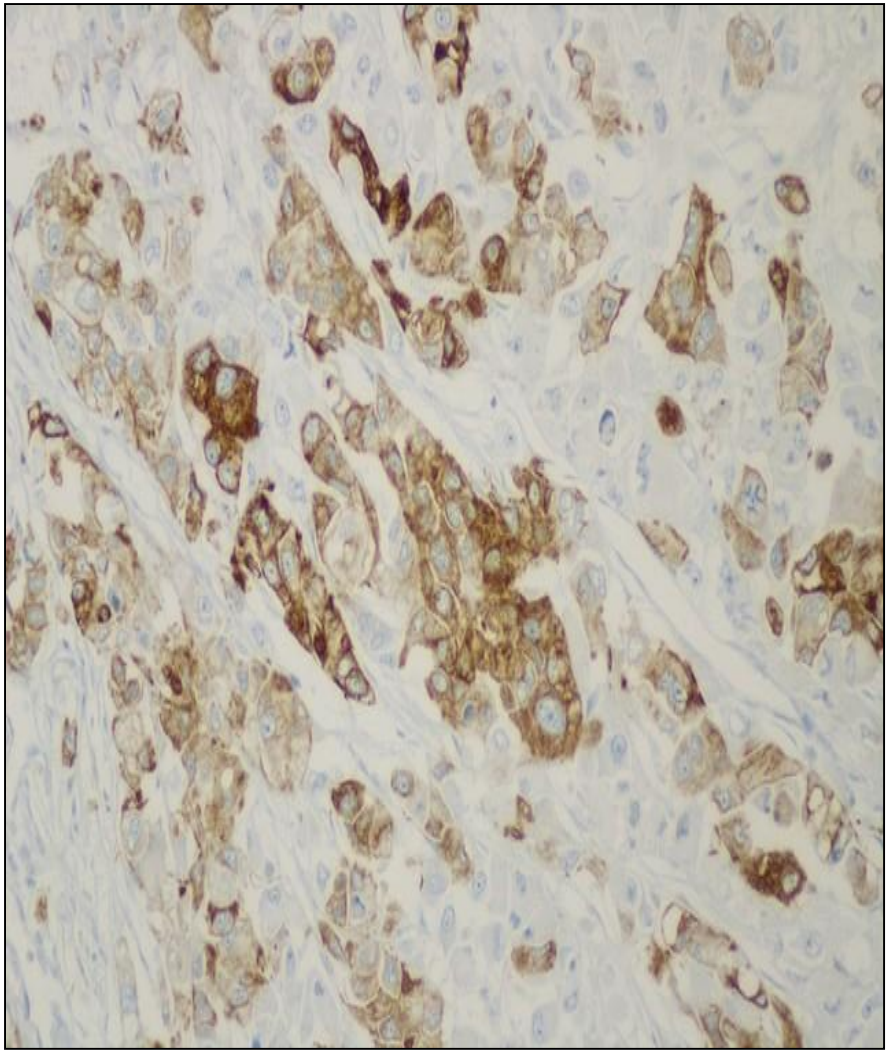
On IHC,

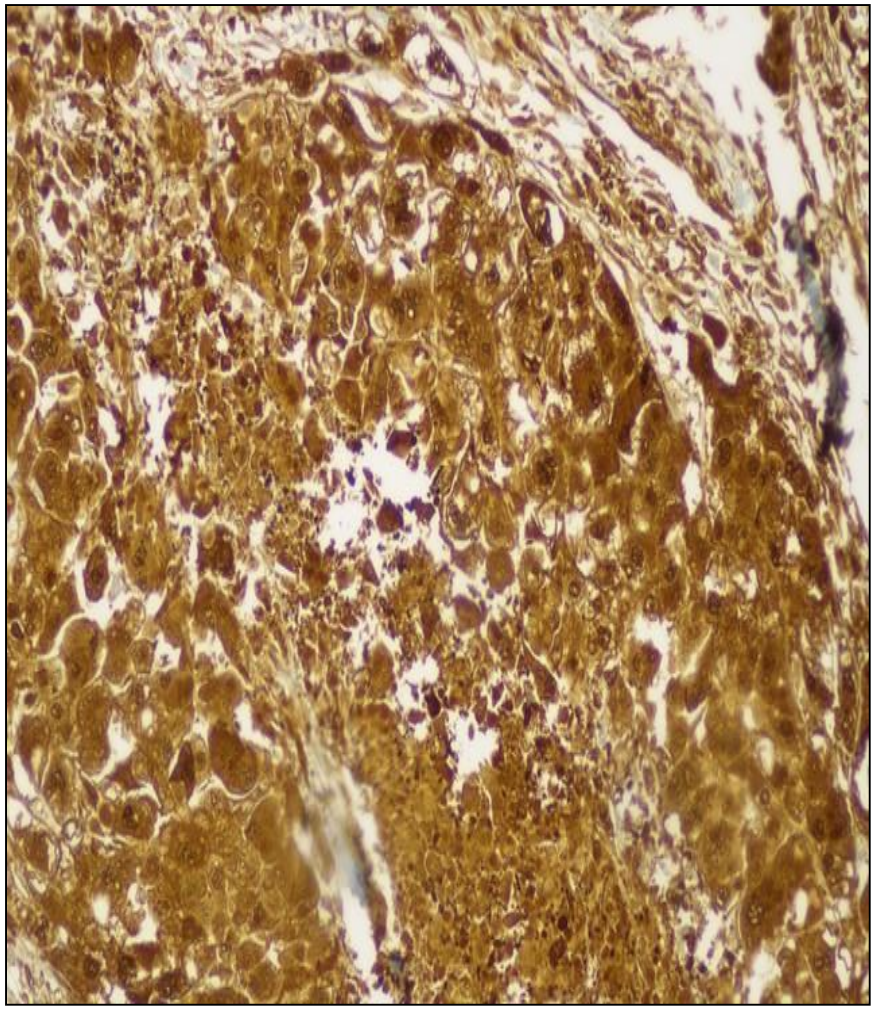
- The tumor cells expressed CK, CK7, S100 and SOX10
- The tumor cells were negative for EMA, CK20, SATB2, p40, p63, desmin, HMB45, PAX8, GFAP, Gata3, TTF1, WT1, 34BetaE12, CD34, CA125, synaptophysin, GCDFP & Calponin



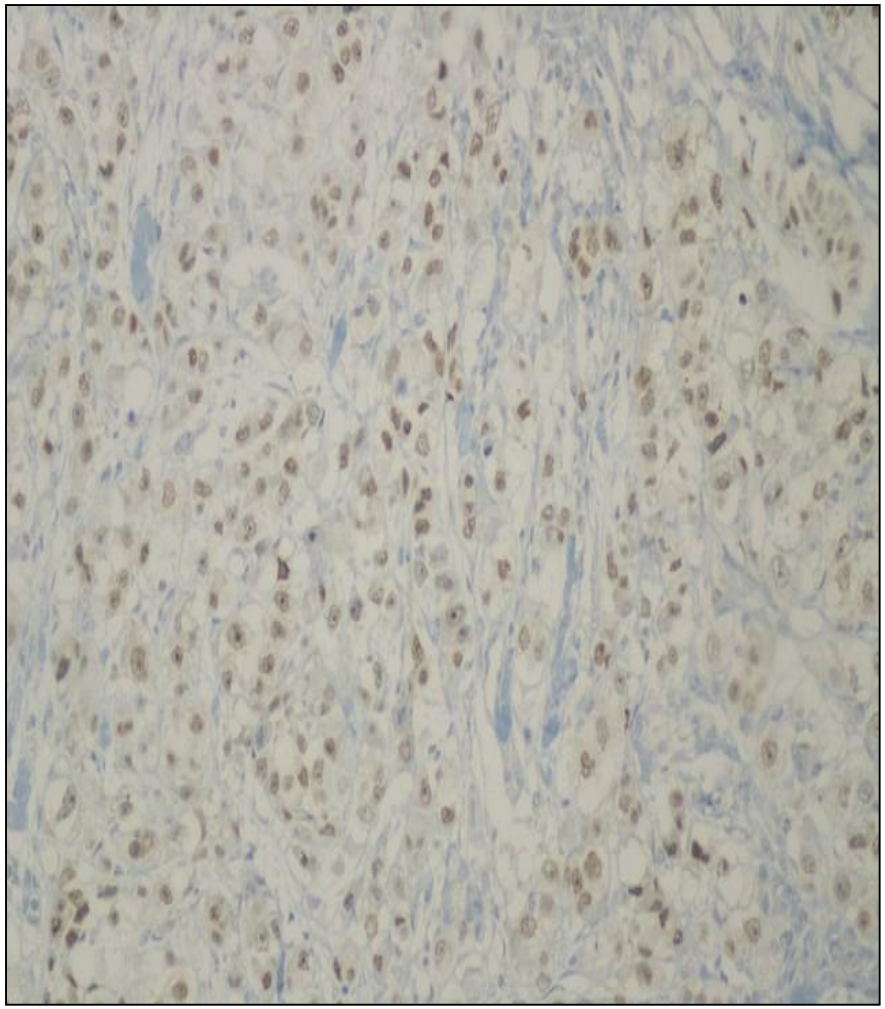
Positive CK expression



Positive CK7 expression



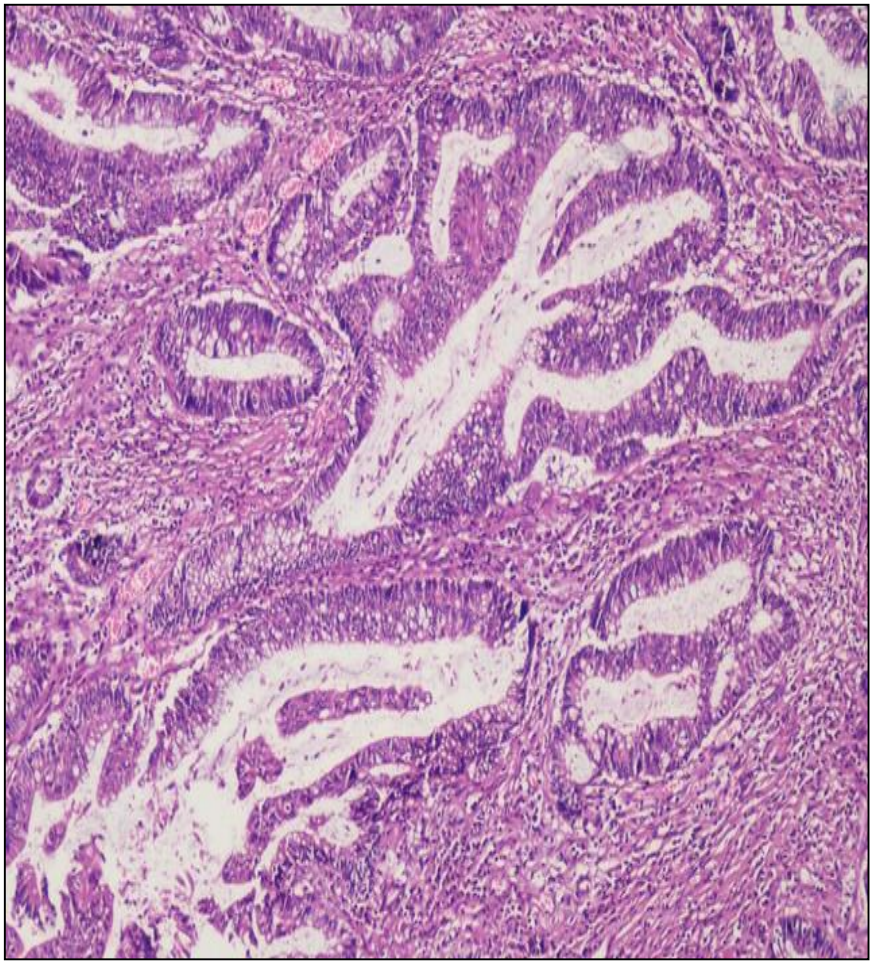
Positive S-100 expression

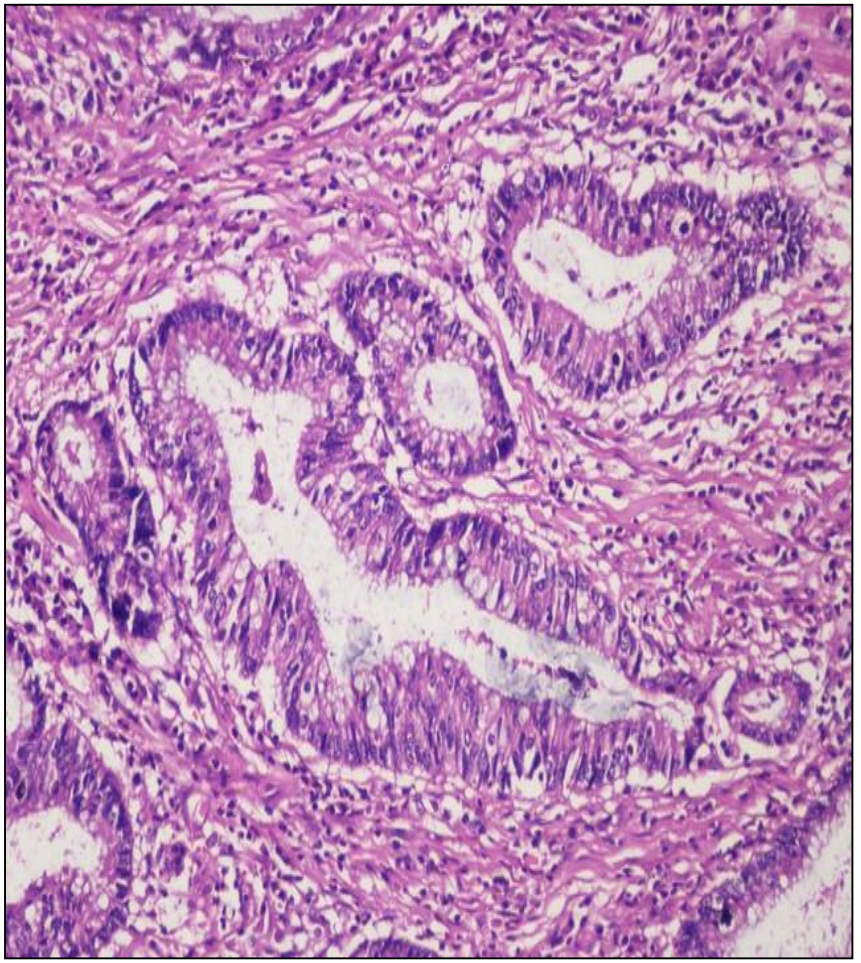


Positive SOX-10 expression

- Histomorphological features and IHC were assessed
- A diagnosis of ***myoepithelial carcinoma of soft tissue*** was rendered

COLONIC MASS



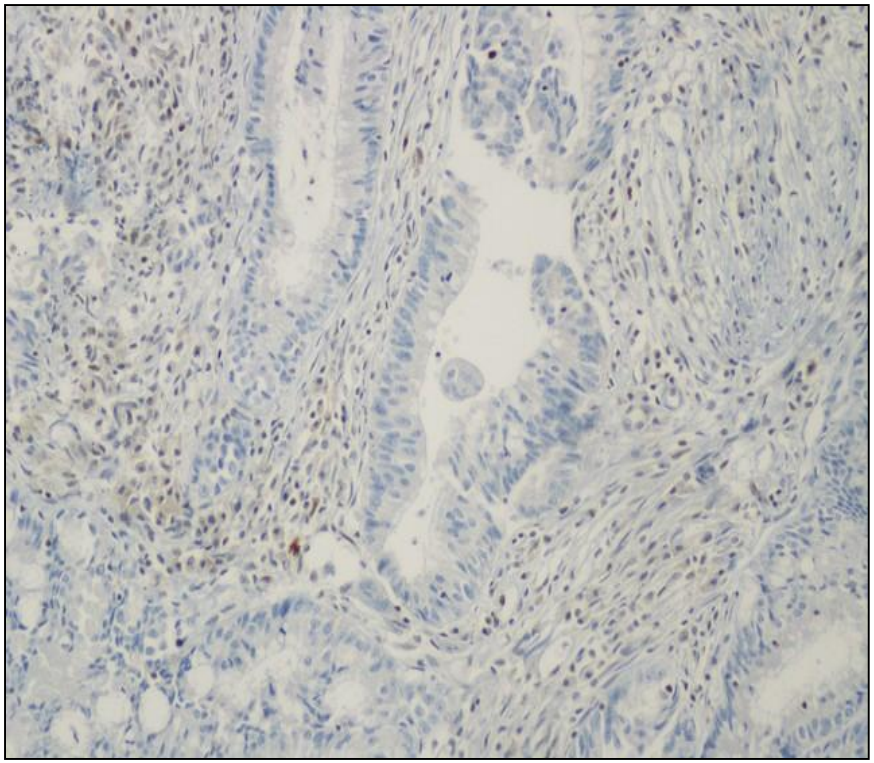


Microscopic examination of the colonic mass revealed a moderately differentiated adenocarcinoma

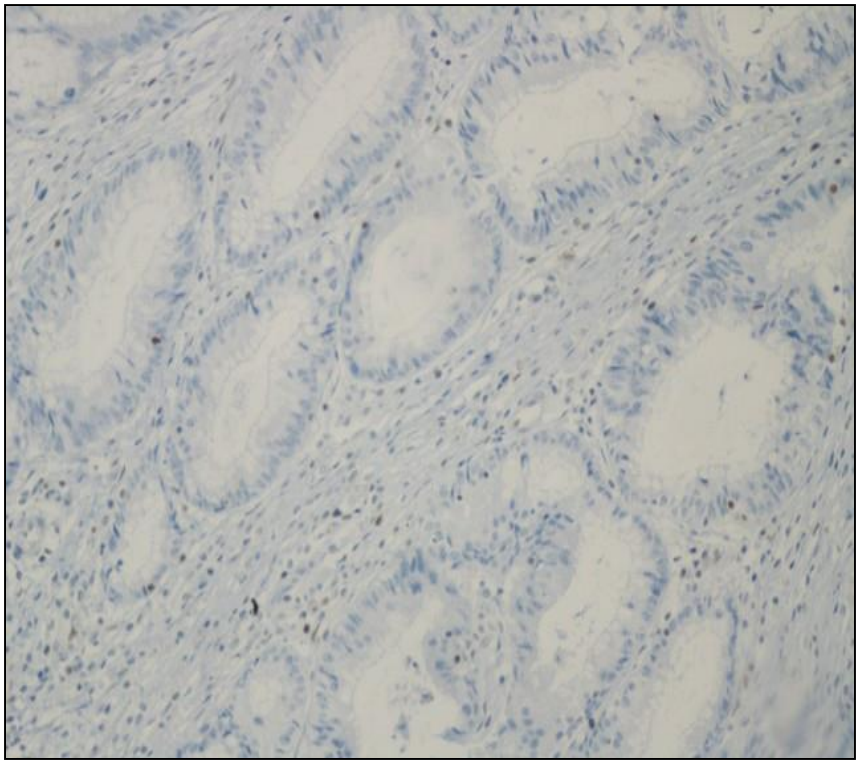
MMR PROTEIN IHC

MMR protein IHC was applied to the colonic adenocarcinoma

Loss of expression of MSH2 and MSH6 was observed



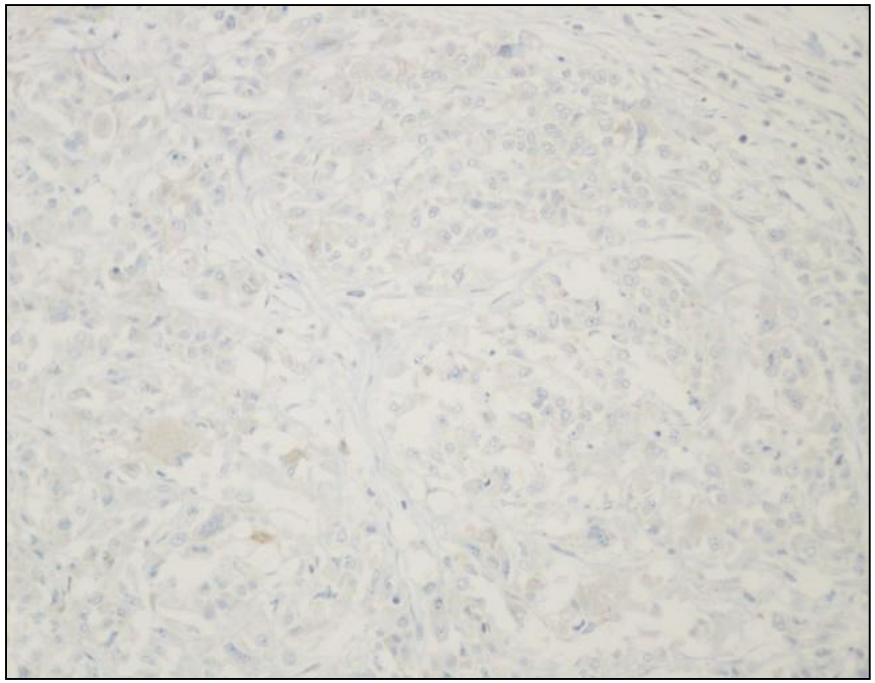
Loss of MSH2 in colonic adenocacrcinoma



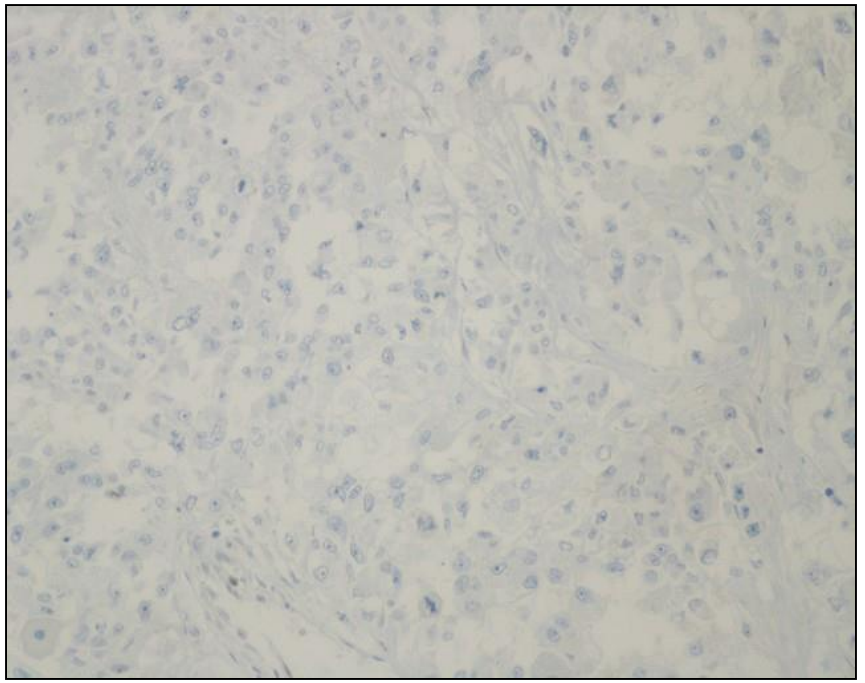
Loss of MSH6 in colonic adenocarcinoma

Subsequently, MMR protein IHC was applied to the chest wall lesion (myoepithelial carcinoma)

Similar loss of expression of MSH2 and MSH6 was observed



Loss of MSH2 in myoepithelial carcinoma



Loss of MSH6 in myoepithelial carcinoma

DISCUSSION:

- This case is of an elderly male who presented with a chest wall mass which was diagnosed as myoepithelial carcinoma of the soft tissue

- The diagnosis in the present case was further confounded by :
  - The concurrent presence of adenocarcinoma of the ascending colon
  - Both lesions showed similar lack of expression of MMR proteins

- Soft tissue tumors are generally not considered to occur in the context of such cases, but have been reported to rarely display loss of MMR protein expression

- It is particularly sarcomas that have been reported in four case reports, and these included:

  - Three cases of malignant fibrous histiocytoma (MFH) and
  - One case of pleomorphic rhabdomyosarcoma

- The two tumors that were genetically investigated (one MFH and the rhabdomyosarcoma) showed MSI and immunohistochemical loss of MSH2 expression

- So far, some soft tissue tumors have specifically been associated with inactivation of MSH2/MSH6, and a higher risk of extraintestinal tumors have been described in families with mutations affecting the MSH2 gene

- Since MSH2 and MSH6 functionally interact, a simultaneous loss of both proteins is expected as was seen in our case

- Suwa et al found MSI in three of 39 cases of soft tissue saromas
  - Liposarcoma,
  - Synovial sarcoma
  - Leiomyosarcoma

- Saito et al reported a MSI low phenotype and loss of MLH1/MSH2expression in five of 11 alveolar soft part sarcomas (ASPS)

- Myoepithelial neoplasms represent a rare group of tumors which is still incompletely characterized clinically, pathologically and genetically

- The histogenesis of myoepithelial neoplasms is not well understood, because:
  - They appear not only at sites with normal myoepithelial cell lining of glandular/ductal structures (skin, lung, larynx, and breast)
  - but also, though rarely, within *soft tissues* and bone, where myoepithelial cells are usually absent

- Myoepithelial carcinoma, also known as malignant myoepithelioma, is a rare aggressive tumor that has been recently described in soft tissue

- Rare tumors are often difficult to diagnose and lack the necessary treatment for disease control, with myoepithelial carcinomas being one of them
- Unless diagnosed at a very early stage, where a complete surgical resection is feasible with wide surgical margins, these tumors have been known to have adverse outcomes
- This case has been presented in view of the rarity of myoepithelial carcinoma of soft tissue, and additionally, it's synchronous presentation with adenocarcinoma of the colon
- The association of myoepithelial neoplasms with loss of MMR protein expression has not been well documented
- There have been cases of myoepithelial carcinoma metastasizing to the colon, however, cases with concurrent presentations of these two tumors has been rarely documented

- For these reasons it seems significant to advertise to the medical community that soft tissue tumors could be related to MMR gene(s) deficiency, particularly to mutations on hMSH2

THANK YOU