Endometrioid adenocarcinoma in a Lynch patient with subseptate uterus – a case report

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Background

Endometrial cancers in women with a congenital uterine malformation are rare and may be diagnostically challenging

Case history

A 39-year old Lynch patient, presented with vaginal bleeding and abdominal pain. Endometrial biopsy was normal. On ultrasonography and computed tomography a uterine malformation was suspected with filling of the right horn and a left-sided ovarian mass (**Figure 1**). A total hysterectomy and bilateral salpingo-oophorectomy were performed.

Macroscopy

Final histopathological examination revealed a 13,5 cm ovarian mass **(Figure 2)**. Furthermore a subseptate uterus was observed with filling of the right uterine horn (diameter 5,5 cm) (**Figure 3**). In addition more than half of the myometrium was invaded by tumor. Cervix, left uterine horn, right ovary and both parametria were free of tumor.

Microscopy

Microscopy revealed a FIGO IIIA high grade endometrioid adenocarcinoma of the right uterine horn (diameter 5,5 cm) and left ovary (diameter 13,5 cm) **(Figure 4).**

Positive		Negative	
CK7	PR	CK20	WT-1
PAX8	Vimentin	Napsin A	p16
ER	Ki67 (high)		

p53 (mutation type-overexpression),

Molecular analyses

Multiple classifier endometrial cancer (POLEmut-MMRd-p53abn)

- MSI high (instable)
- POLE (p.P286L) mutation
- TP53 (p.P278H) mutation

The ovarian and endometrial mass are clonaly related

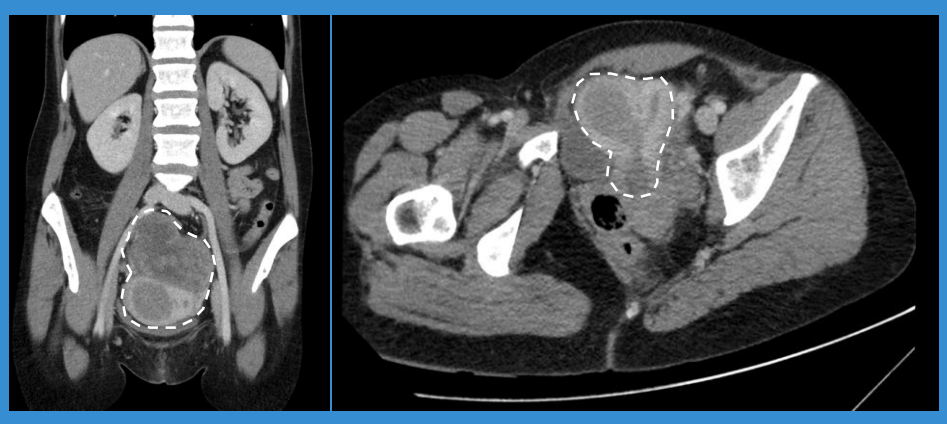

Figure 1. Radiology – ovarian mass and uterine malformation

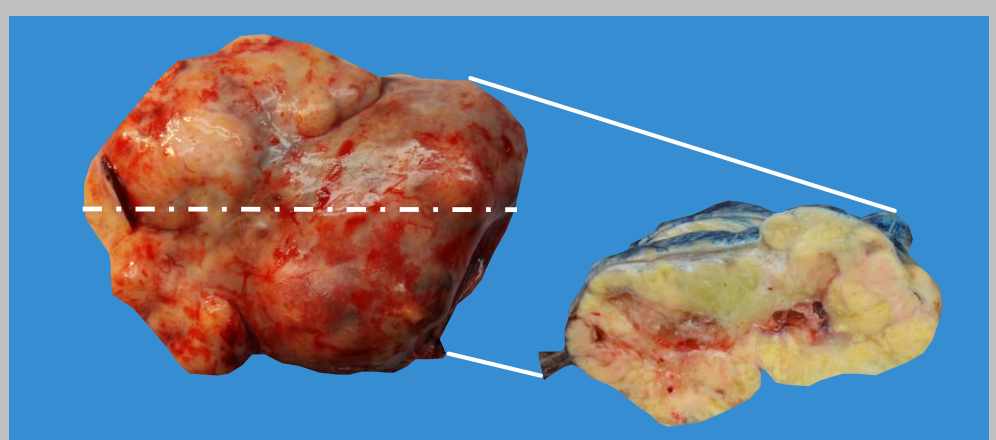

Figure 2. Macroscopy – gross appearance of a 13,5 cm left sided ovarian mass

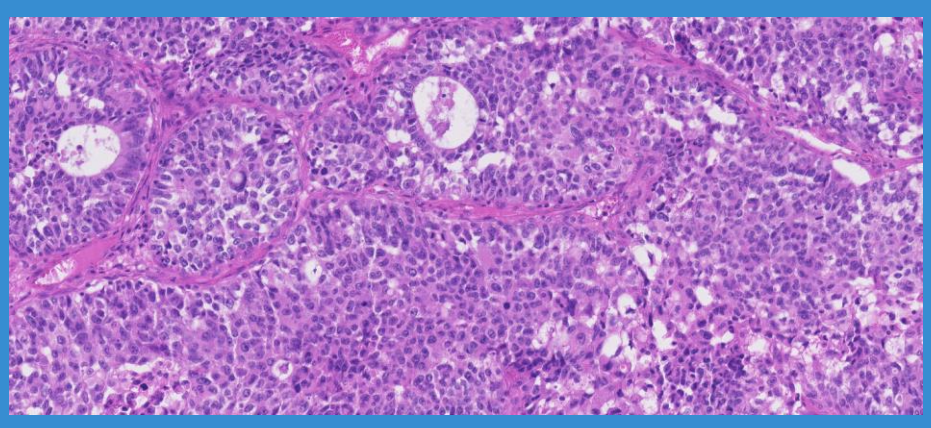

Figure 4. Microscopy – Grade 3 endometrioid adenocarcinoma (ovary + uterus)

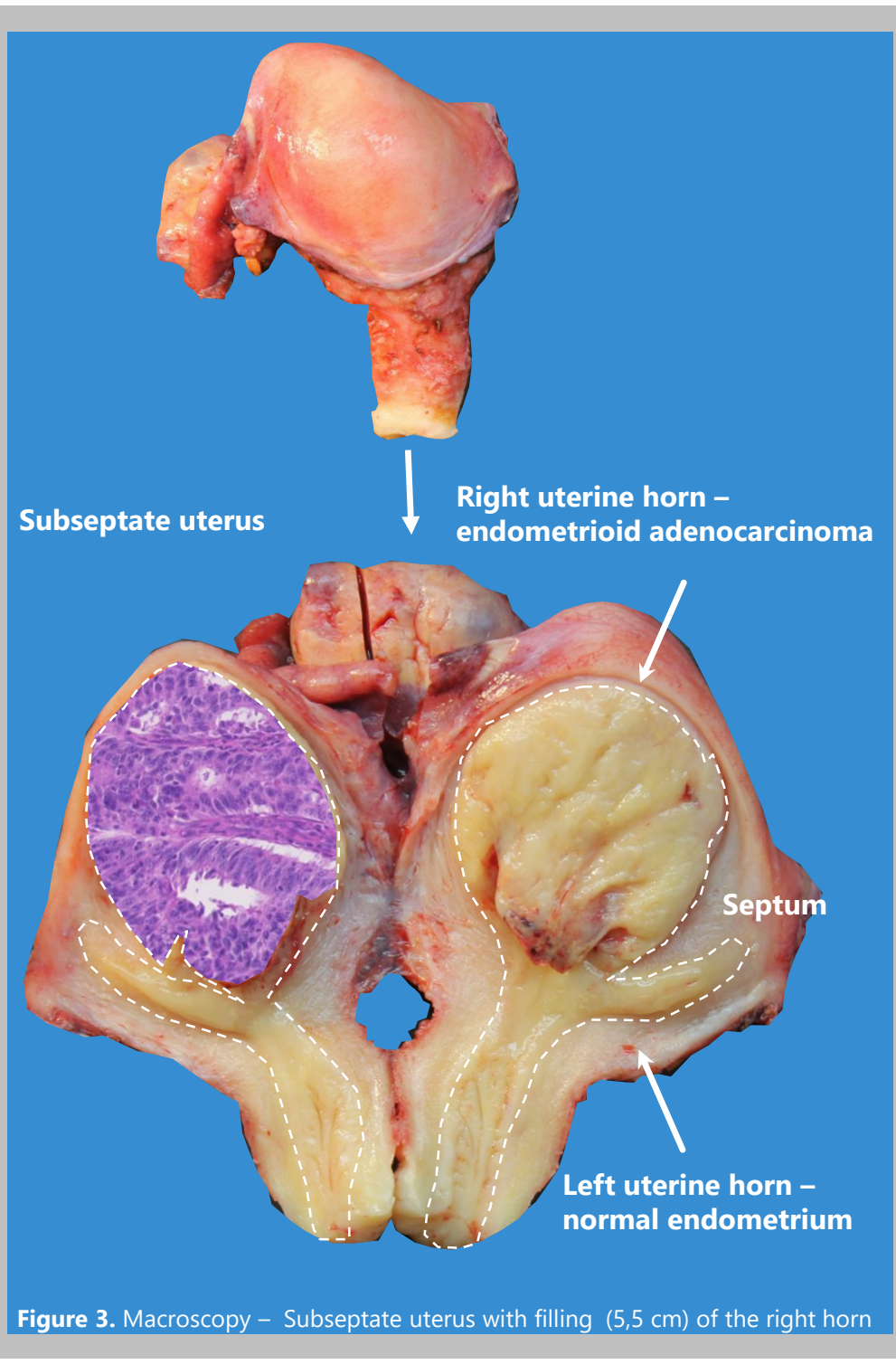

Figure 3. Macroscopy – Subseptate uterus with filling (5,5 cm) of the right horn

Conclusion

Clinicians should be aware that congenital uterine malformations and uterine malignancies can coincide. Uterine malformations may cause false negative results upon initial histopathological examination if only one horn is affected. In these cases - clinical, radiological and histopathological correlation is key.

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