BREAKING THE PATTERN: GONADAL GERM CELL TUMOR IN AN ADOLESCENT MALE WITH KLINEFELTER SYNDROME

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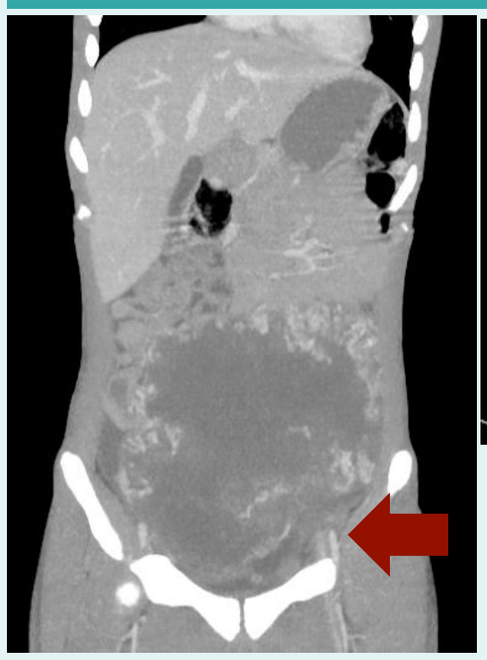
INTRODUCTION

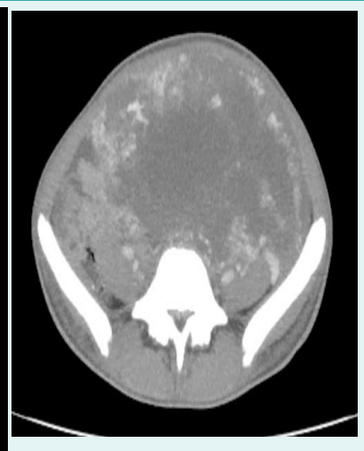
- Klinefelter Syndrome (KS) affects around 1 in 450-600 male births, 50-75% of cases go undiagnosed[1].
- Individuals with KS are more likely to develop mediastinal teratomas or GCTs with mixed histology[2].
- There is a strong association of KS with primary mediastinal non-seminomatous germ cell tumor (PMNSGCT), with reported incidence of 8-18%[2].
- **Aim:** To report the rare association of KS with gonadal choriocarcinoma in an adolescent male.

CASE PRESENTATION

- 14 years old
- Phenotypically male boy
- Bilateral undescended testes
- Abdominal pain and distension-3 months

WORK UP



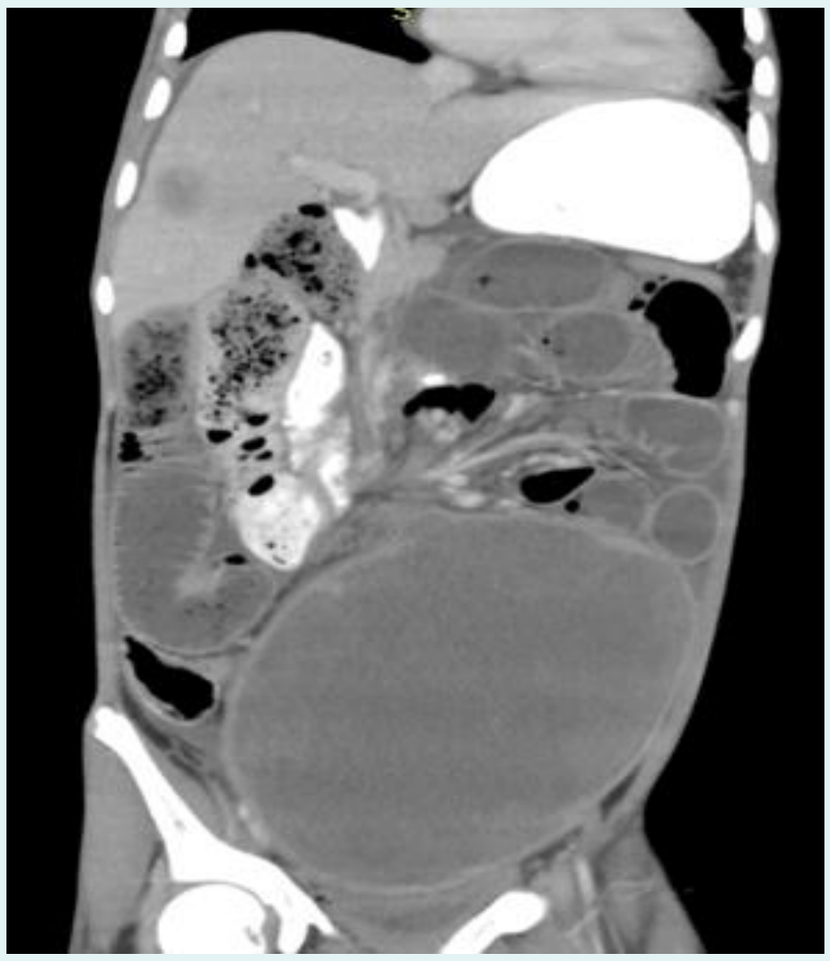


abdomino-pelvic mass

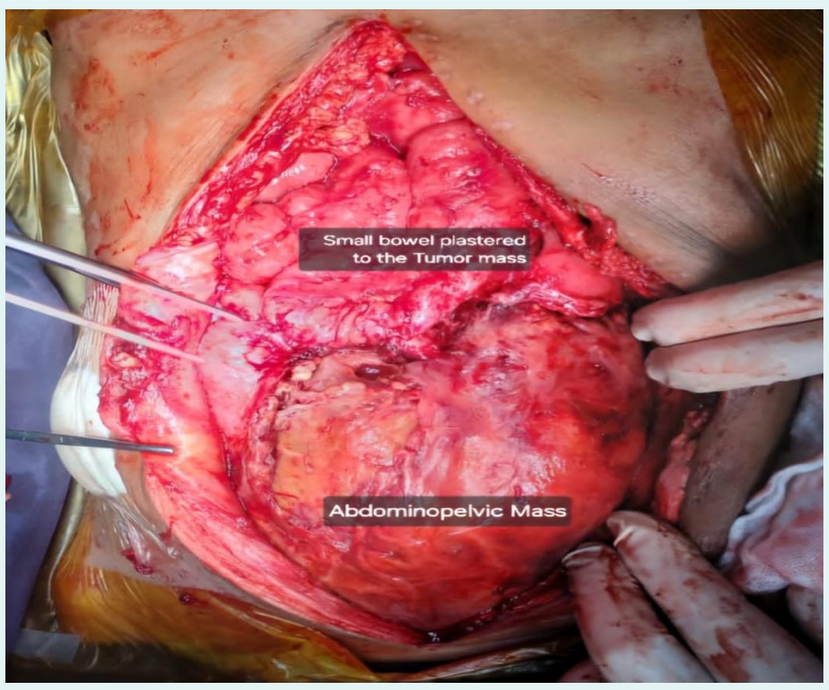
Beta HCG	>20,000 ng/ml
AFP	63 ng/ml
GGT	129 U/L
STAGING WORKUP	Cystic lesion consistent with hemangioma
METASTATIC WORKUP	Negative

- **Biopsy**- Choriocarcinoma

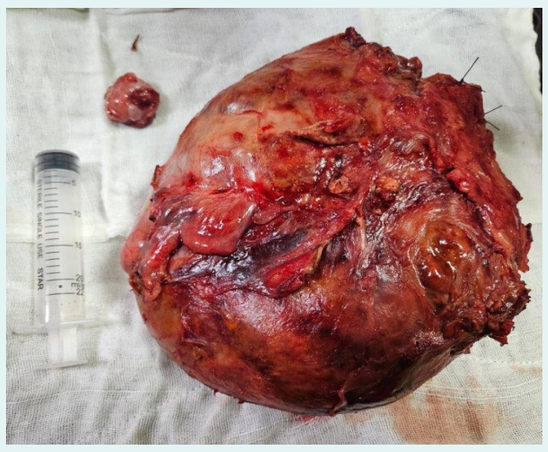
MDT DISCUSSION: 4 cycles of BEP chemotherapy



Re-assessment CT

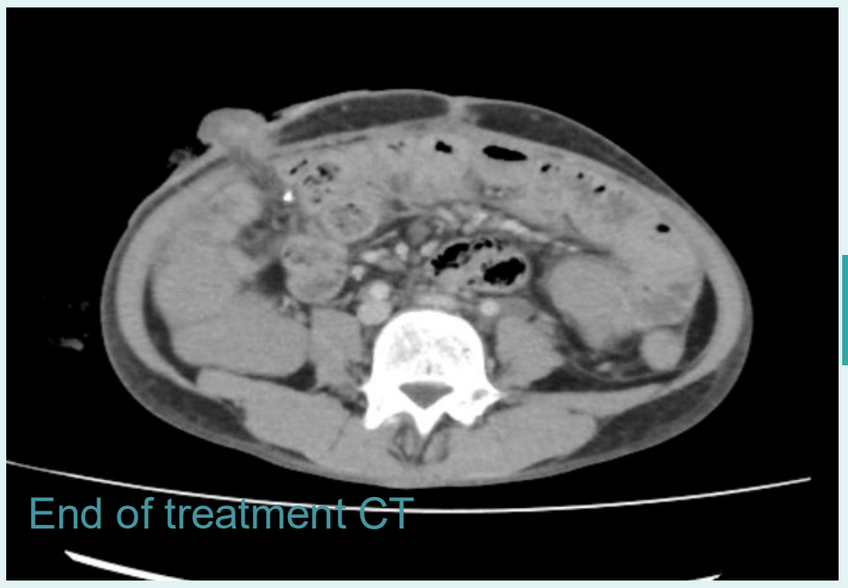


Emergency exploration with excision of abdomino-pelvic mass and ileostomy



- **Biopsy**- Residual mature teratoma with extensive necrosis

MDT DISCUSSION: Follow up with tumor markers and CT scan

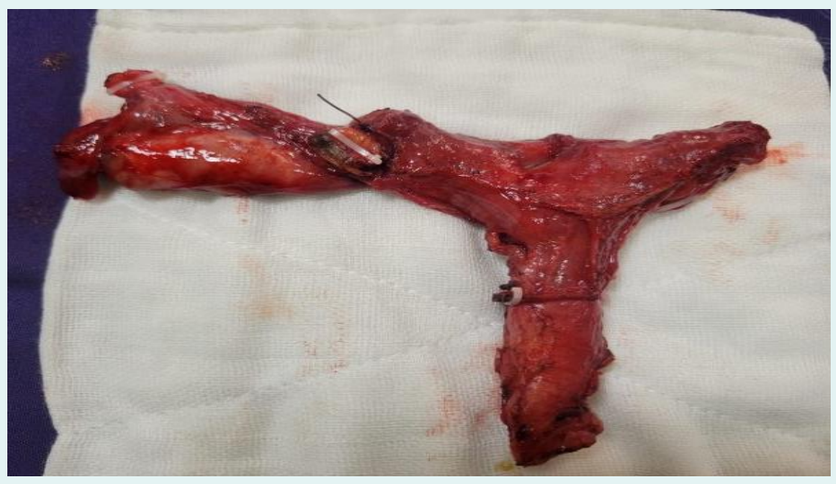


- Exploratory laparotomy for ileostomy reversal:
- **Right gonad** with mullerian duct remnants in pelvis (**right fallopian tube and rudimentary uterus**).
- No left gonad visualized.

- **Gonadal biopsy- testicular parenchyma**, lined by spermatocytes and Leydig cells.
- **Periphery - ovarian** type stroma (metaplastic).

MDT DISCUSSION: Karyotyping, Endocrinologist review

Karyotyping: 47 XXY (klinefelter)
Gender Rear up: Rear up as male.
Right gonadal remnant excision.
Hormone replacement.



Right gonad and rudimentary uterus

Follow-up: Lifelong Testerone supplementation.

CONCLUSION

- This case demonstrates complex clinical spectrum and to rule out chromosomal anomalies in patient with any diagnostic challenge.
- Multi disciplinary management with chemotherapy, surgical excision and hormonal replacement is essential to achieve favorable oncological and long-term functional outcomes.
- This case is an effort to enrich current literature regarding such rare association.

REFERENCES

1. Mehmet B, Dwyer AA, Jayasena CN, Gillard S, Llahana S. Update on Physical, Psychological and Quality of Life Management in Klinefelter Syndrome. The Journal of Clinical Endocrinology & Metabolism. 2025 May 2:dgaf261.
2. Bokemeyer C, Nichols CR, Droz JP, Schmoll HJ, Horwich A, Gerl A, Fossa SD, Beyer J, Pont J, Kanz L, Einhorn L. Extragonadal germ cell tumors of the mediastinum and retroperitoneum: results from an international analysis. Journal of Clinical Oncology. 2002 Apr 1;20(7):1864-73.

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