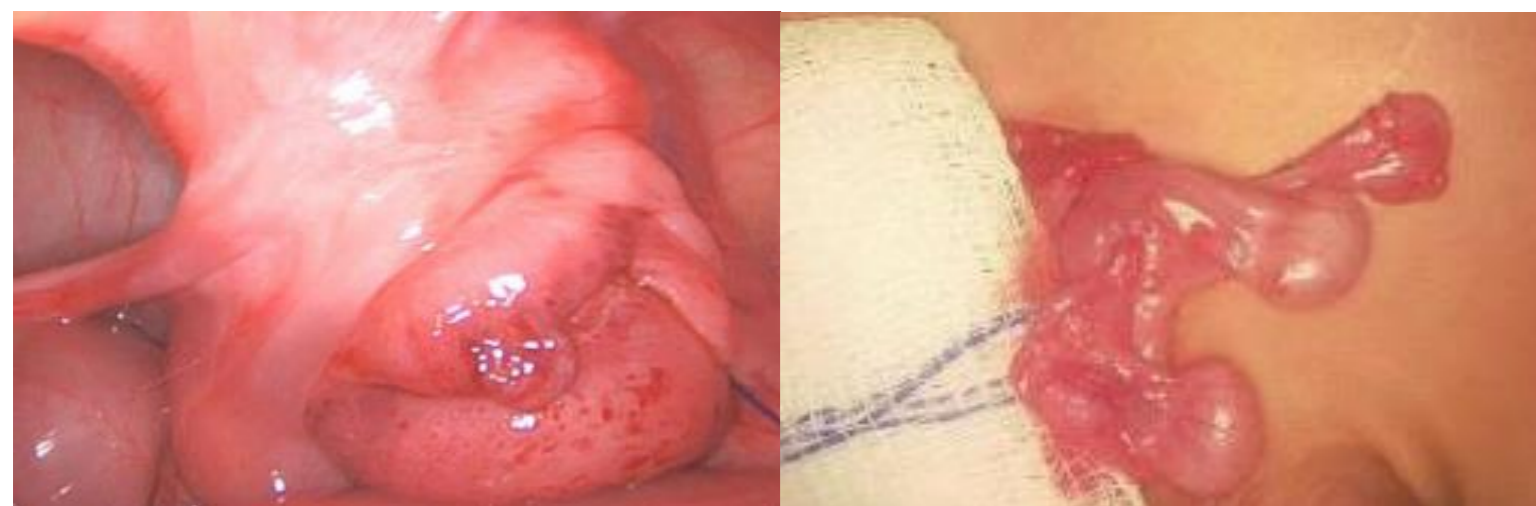


INTRODUCTION

GD: Incomplete or defective gonadal differentiation caused by disturbances in **germ cell migration** or **organization** in the **gonadal ridge** with variable differentiation towards **ovary, testis, or both**.

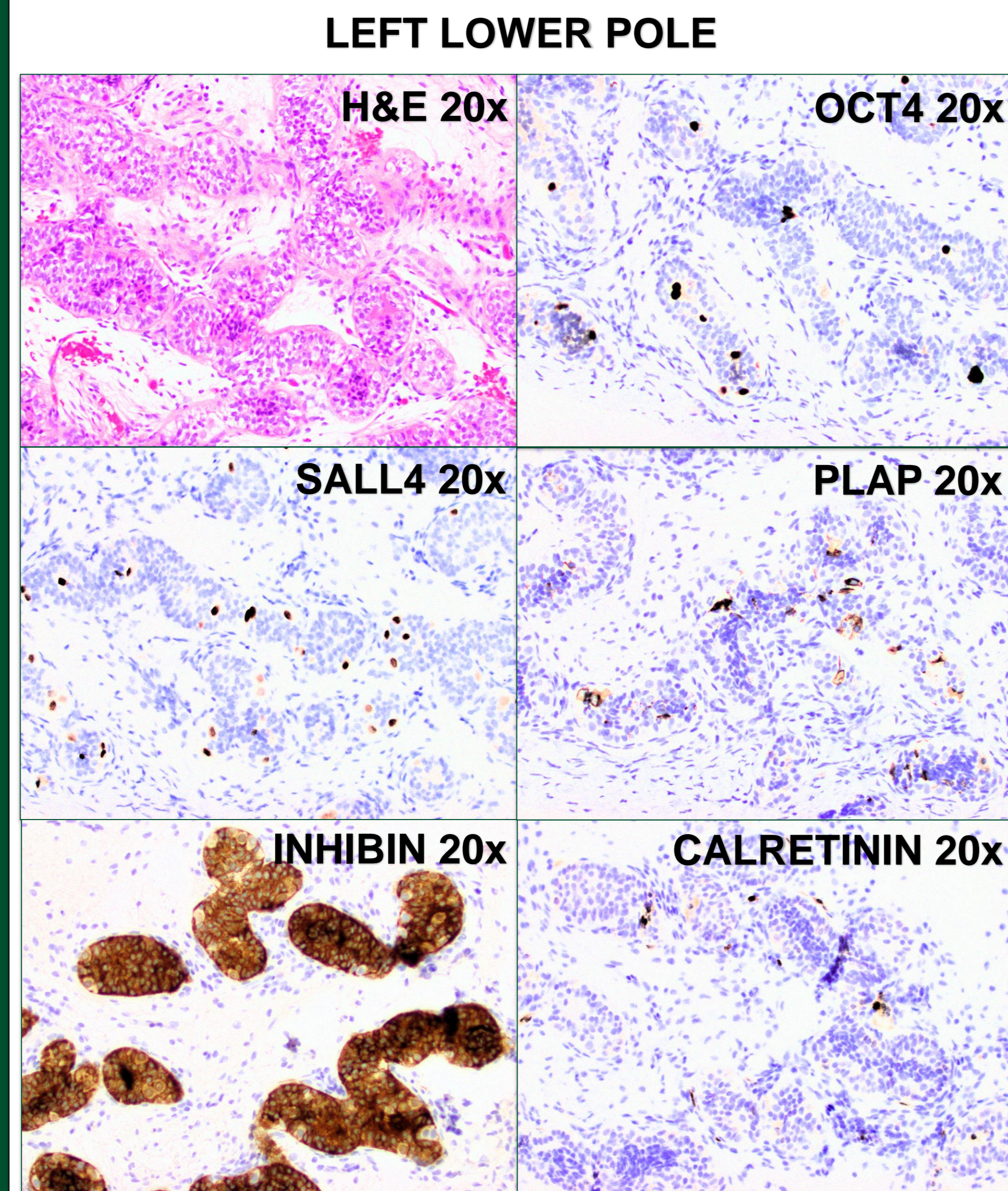
CASE PRESENTATION

Patient: 2-year-old male with recently discovered undescended testicles in the right scrotum and incarcerated non-reducible hernia sent for urgent surgery.



Intraoperative findings: Suspected persistent Müllerian duct syndrome (PMDS) with a rudimentary uterus between two gonads described as “testicular-like” in appearance with islands of ovarian tissue with no obvious vas deferens or fallopian tubes, biopsies of the gonads taken.

INTRODUCTION

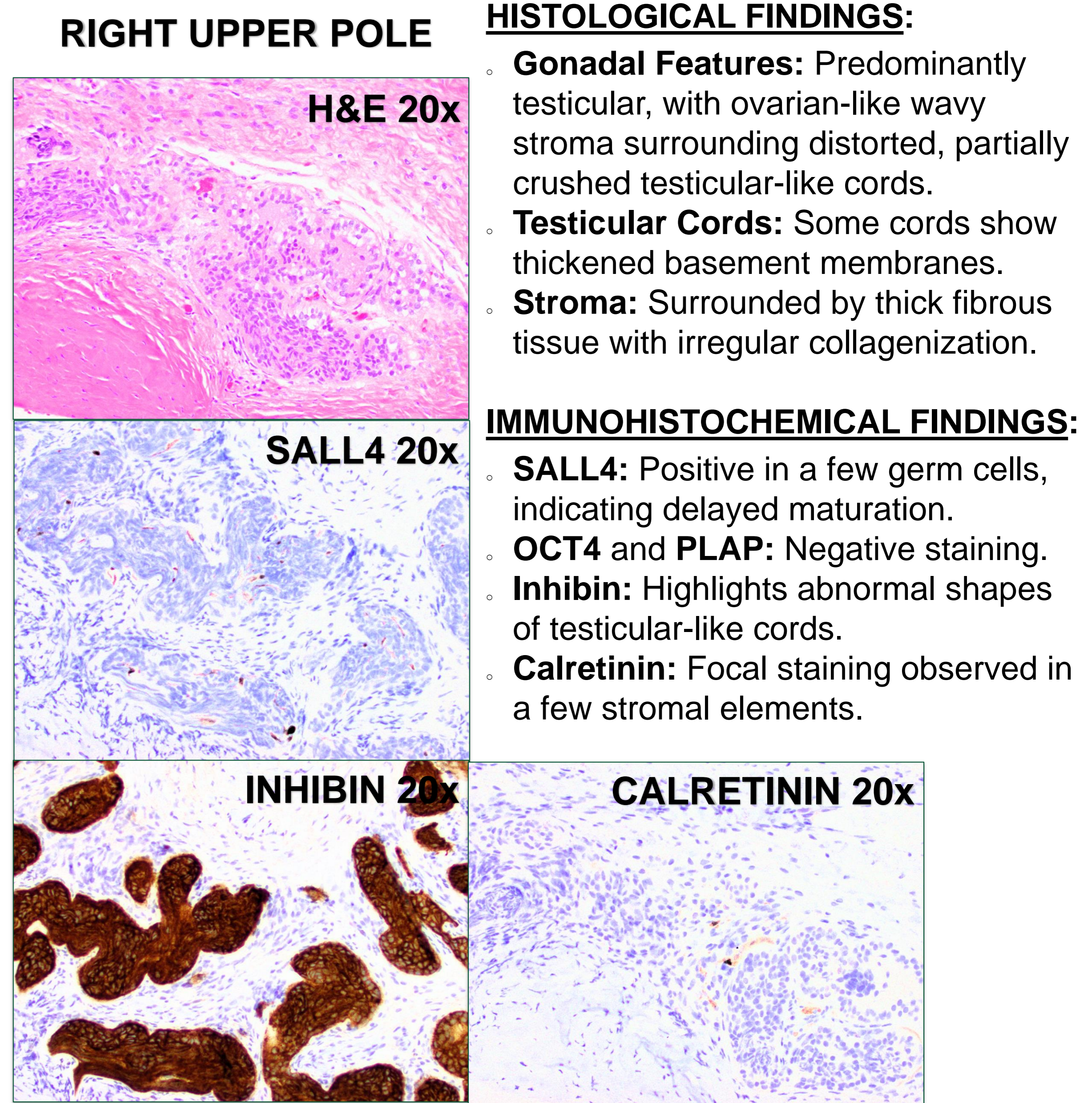


HISTOLOGICAL FINDINGS:

- Testicular Parenchyma:** Surrounded by irregular collagenized tunica albuginea.
- Testicular Cords:** Dysmorphic, anastomosing, and irregular with hypocellular intertrabecular tissue.
- Germ Cells:** Pale, large cells within the cords, consistent with germ cells, but predominant Sertoli cells.
- Leydig Cells:** Absent in the interstitium.
- Tunica Propria:** Variably thickened around the cords.

IMMUNOHISTOCHEMICAL FINDINGS:

- SALL4 and OCT4:** Scattered positive nuclei, indicating delayed germ cell maturation.
- PLAP:** Membranous germ cells staining.
- Inhibin:** Strong positivity in Sertoli cells, emphasizing their dysplastic and irregular architecture.
- Calretinin:** Positive in scattered stromal elements (Leydig cells) and mesothelium in the tunica albuginea, with aberrant staining in some cords.



HISTOLOGICAL FINDINGS:

- Gonadal Features:** Predominantly testicular, with ovarian-like wavy stroma surrounding distorted, partially crushed testicular-like cords.
- Testicular Cords:** Some cords show thickened basement membranes.
- Stroma:** Surrounded by thick fibrous tissue with irregular collagenization.

IMMUNOHISTOCHEMICAL FINDINGS:

- SALL4:** Positive in a few germ cells, indicating delayed maturation.
- OCT4 and PLAP:** Negative staining.
- Inhibin:** Highlights abnormal shapes of testicular-like cords.
- Calretinin:** Focal staining observed in a few stromal elements.

DIAGNOSIS AND DISCUSSION

The observed features are characteristic of gonadal dysgenesis, with a **dysgenetic/dysplastic testis** on the **left** side, and a **streak gonad (streak testis)** on the **right** side.

Dysgenetic Testis Characteristics	Streak-Testis
<ul style="list-style-type: none"> Compact seminiferous cords/tubules with variable germ cell presence. Sertoli and Leydig cells express inhibin, AMH, and calretinin. Poorly collagenized tunica albuginea with ovarian-like stroma. Features intratubular germ cell neoplasia in some cases. 	<ul style="list-style-type: none"> Combination of dysgenetic testis and streak gonad with epithelial cords or ovarian follicles. Linked to specific syndromes, such as mixed gonadal dysgenesis (MGD) and persistent müllerian duct syndrome (PMDS).

Pathology **bridges genetic** mutations and **morphological** gonadal **abnormalities** offering dual insight explaining variability in clinical presentations through gonadal phenotypes and complementing chromosomal or genetic findings with histopathological observations aimed at supporting clinical syndrome correlation.

REFERENCES

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