07/31/21 Morning Report with @CPSolvers

Case Presenter: *Thiago Mendes (@MendesThiagoB)* ***Case Discussants***: *Gabriela Pucci (@GabiFPucci) and Rabih Geha (@RabihMGeha)*

CC: Lack of menstruation
HPI: 13 y female with no menstruating and undeveloped body. October 20 referred by gynecologist she had infantile uterus. She felt bad comparing with friends

Past Medical History:
None

Meds:
None

Family History:
Menarche of mother 10. Sister 17 and had menarche at 11. Father side had menarche at normal time.

Social History:

Health Related Behaviours:

Allergies:None

Vitals: **T**: **HR**:**BP**: **RR**: **SpO_2**: Height: 1.56 cm, Weight: 39.9 BMI: 16.2
Exam:
Gen: Taner:M1P1, Rest of the examen normal
HEENT: Normal
CV: Normal
Pulm: Normal
Abd: Normal
Neuro: Normal
Extremities/Skin: Normal
Vagina: N, no signs of virilization

Notable Labs & Imaging:
Hematology:
WBC: Hgb: Plt:

Chemistry:
Na: K: Cl: CO2: BUN: Cr: glucose: Ca: Phos: Mag:
AST: ALT: Alk-P: T. Bili: Albumin:
Estrogen less than 20 FSH: 137.3 LH:27.46 TSH: 2.3

Imaging:
EKG:
CXR:
US: Uterus of 3.9 cc right ovary 0.8 cc and left 0.6cc.
Karyotype: XX in all cells
Dx: Gonadal dysgenesis XX
Started in Progesterone and breast development

Problem Representation:
ENG: 13 F p/w lack of menstruation and infantile uterus. Elevated FSH. Normal Karyotype. DX: Gonadal dysgenesis with XX.
ESP: 13 F con ausencia de menstruación y útero infantil. FSH elevado. Cariotipo normal. Dx: Disgenesia gonadal con XX.
POR: 13 M com ausencia de menstruacao e utero infantil. FSH elevado. Cariótipo normal. Dx: Disgenesia gonadal.

Teaching Points (Vale):

- **Delayed Puberty: Is it really delayed? ->** Determine Tanner stage, growth curve, history from parents. The absence of breast suggests an absence of estrogens.
- **Primary Amenorrhea:** Absence of menarche and secondary sexual characteristics by 13 y/o or presence of 2ry characteristics by 15 y/o.
- **Amenorrhea:** 1ry (congenital-gonadal dysgenesis, Turner syndrome vs acquired-autoimmune) vs 2ry.

Hypogonadism: Low estrogen.

- **Peripheral-Hypergonadotropic (High LH, FSH):** Ovarian Insufficiency, Hypothyroidism.
- **Eugonadotropic Eugonadism:** Signs of androgen excess (hirsutism, acne). Includes PCOS.
- **Central-Hypogonadotropic (Low LH, FSH):** Pituitary (autoimmune, empty sella, infiltrative, prolactinoma, medications), Hypothalamus (eating disorder, functional, infection, Kallmann syndrome).

Collecting clues: Presence of uterus and ovaries: Mullerian duct development (rules out Mullerian Agenesis, 5-alpha reductase deficiency). MRKH Syndrome absence of internal organs (1ry amenorrhea), but normal 2ry sexual characteristics.

- **Primary Ovarian Insufficiency:** Most common Turner Syndrome. Other causes: gonadal dysgenesis (normal karyotype).
- Transmasculine patients can prefer gender neutral terms for certain anatomical parts (Ex. Use internal organs, rather than uterus or ovaries, external opening, rather than vagina). But, most importantly: Ask!