Title: SWYER SYNDROME (46,XY COMPLETE GONADAL DYSGENESIS): A RARE CASE OF PRIMARY AMENORRHEA

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Abstract (up to 300 words)
Swyer syndrome is a type of gonadal dysgenesis wherein a 46,XY karyotype presents with a female phenotype. It is a rare cause of disorder in sexual development that occurs in 1:100,000 births. Local studies are currently limited to few case reports. Sex-determining region on the Y chromosome gene mutation is the root cause of nonfunctional gonads with no hormonal or reproductive potential. They are born with normal female external genitalia but not suspected until puberty when menses do not occur or if secondary sexual characteristics do not develop. This report presents the case of a 23-year-old phenotypically female presenting with primary amenorrhea and hypogastric discomfort. Ultrasound revealed an infantile cervix and uterus with streak left ovarian tissue and a cystic mass on the right pelvic area. Gonadotropin levels were elevated, and the karyotype showed a normal male 46,XY. Laparoscopic bilateral gonadectomy with salpingectomy was done, which revealed dysgerminoma on bilateral ovarian tissues. In conclusion, this report describes a rare case of Swyer syndrome associated with ovarian dysgerminoma. Accurate and prompt diagnosis, using a systematic approach in evaluating primary amenorrhea, is crucial in initiating treatment. Our goal is to ensure hormonal replacement, fertility preservation, psychosexual and emotional stress reduction, and overall patient survival.

Biography (up to 150 words)
Pamela Mallari is currently a third-year resident in training in St. Luke's Medical Center, Quezon City, Philippines.

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