INTRODUCTION:
- 46,XX Pure gonadal dysgenesis is a rare, genetically heterogeneous condition causing primary amenorrhoea.
- Gonadal dysgenesis is a genetic condition due to errors in cell division and or alterations in genetic material, leading to complete or partial loss of gonadal development.
- The development of gonadal dysgenesis begins early either at fertilization or shortly after in the early stages of the embryo and foetus. Complete gonadal dysgenesis genotypes are known as either 46, XX or 46, XY.
- The 46, XX type may develop due to genetic mutations or environmental factors affecting ovarian development leading to ineffective ovaries. 46, XX type gonadal dysgenesis is on the spectrum of conditions that cause premature ovarian failure.

CASE DETAIL:
- 17 year old girl presented to the GP with lack of menarche and breast development.
- There was no family history of consanguinity, miscarriages or any other family member with primary amenorrhoea.
- Her birth and neonatal period were uneventful and she had achieved target milestones during developmental stage. Personal medical history did not reveal any serious illness.
- Physical examination showed normal stature without any phenotype of Turner syndrome or signs of hyperandrogenism. She presented complete armpit and pubic hair development and a stage 3 mammary development on Tanner scale. Her external genitals were normal and her clitoris was of normal size.

INVESTIGATIONS:
- Endocrine evaluation indicated hypergonadotropic hypogonadism with raised Follicle Stimulating Hormone (FSH) and Leutening hormone (LH) levels with low oestradiol level.
- Subsequent MRI showed rudimentary uterus with absent ovary.
- She did not have complete absence of genitourinary system with the presence of vagina ruling out a Mullerian agenesis picture.
- Ovarian and adrenal antibody testing was negative.
- Karyotyping revealed normal 46 XX pattern and she was diagnosed with 46 XX Pure gonadal dysgenesis.

TREATMENT:
- She was commenced on oestrogen replacements and went on to combined oestrogen and progesterone replacement to help withdrawal bleeding. Sterility is an obvious consequence and she was provided appropriate counselling regarding available options of assisted fertility.
- She was also given Vitamin D and Calcium supplement for bone protection.

CONCLUSION:
- Early diagnosis is extremely important to begin prompt treatment with hormone replacement and reduce risks associated with long term lack of female sexual hormones and it is also essential to provide adequate emotional support and guidance for the patient and family.

Declaration:
The authors confirm no conflict of interest.