Background

- Primary amenorrhea defined as the absence of menstruation at 15 years of age in the presence of normal growth and secondary sexual characteristics.  
- Rare, occurs in <1% such as Turner Syndrome. Mosaic Turner syndrome is one of challenging type.  
- This is a case report of 15-year-old woman presenting with primary amenorrhea with the result of chromosome 45X/46, XY Mosaic Turner.

Case Report

- A 15 years old woman, no menstruation, no breast growth and pubic hair, short stature, no acne, with loud voice and hirsutisms. Tanner 2 and female external genitalia appearance.  
- Her height 139 cm and weight 45 kg  
- LH 13.18, FSH 45.64, prolactin 23.18 and estradiol < 9, testosterone 2.83 ng/dl, chromosomal analysis mos 45X(23)/46,XY  
- The MRI examination revealed normal uterus size (3.4 x 2 x 4.2 cm Vol 14.8 cc), anteflexed, no masses or cysts were seen. Normal left and right adnexa.  
- Laboratory results within normal range  
- Oral contraceptive was given for 6 months and later, thus her height increased to 145 cm and had regular menstrual period.

Discussion

- Primary amenorrhea is a matter of concern for Obstetrics and Gynecology regarding their reproductive life  
- The diagnosis of Turner syndrome based on phenotype and presence of hypergonadotropic hypogonadism; This mosaic type occurs in about 25% of Turner syndrome  
- Y chromosome is identified, increasing the risk of gonadoblastoma about 20-30% but her laboratory revealed low testosterone level, hence gonadectomy not yet recommended  
- The patient also did not have prominent virilization signs such as acne, deep voice, enlarged clitoris. This is in accordance with the theory presented by Guedes et al. (2006) that girls with presence of a Y-chromosome has been associated with virilization only 5% and/or turner-like clinical picture except for growth retardation.  
- The goal of replacement are to mimic the normal progression especially menarche in this girl while maximizing growth potential and minimizing risks.  
- Combined oral contraceptive is an option for treat this patient whereas her height has increased to 145 cm and has been having monthly menstrual period.  
- The administration of growth hormone had no function anymore. Therefore, the treatment aims to patient satisfaction, growth and development measures.  
- Patients with 45X/46,XY mosaic type Turner syndrome have reproductive potential problems, so age-appropriate counseling can significantly reduce the adverse psychological impact of diagnosis  
- Successful pregnancy outcomes have occurred in patients with 45X/46,XY mosaicism as well as 46,XY gonadal dysgenesis following oocyte donation and in vitro fertilization with donor egg and/or gestational surrogacy

Conclusion

- Physicians should pay special attention to patients, treat them individually  
- Recommend a comprehensive psychoeducational assessment involving a team of gynecology, genetics, psychology and counsellors.  
- The management and prognosis of the patient depend on the primary etiology of the primary amenorrhea.

References