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RESEARCH ARTICLE

CASE STUDY: TURNER SYNDROME UNDERSTANDING TURNER SYNDROME: DIAGNOSIS, IMPLICATIONS, AND MANAGEMENT

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Abstract

This case study presents an 18-year-old woman with mosaic Turner syndrome, diagnosed in 2020, who presented with severe short stature and a desire for pregnancy. Complicated by premature ovarian failure and a history of inconsistent hormone replacement therapy, her fertility prospects are significantly impacted. Investigations revealed a severely diminished ovarian reserve, indicated by low Anti-Müllerian Hormone (AMH) and non-visualized ovaries on ultrasound. The case highlights the complexities of managing Turner syndrome, emphasizing the need for a multidisciplinary approach involving endocrinology, cardiology, and reproductive medicine. It underscores the importance of patient education, psychosocial support, and thorough discussion of fertility options, including oocyte donation, while carefully considering the increased risks associated with pregnancy in Turner syndrome. The study emphasizes the critical role of patient-centered care and effective communication in empowering individuals with Turner syndrome to make informed decisions regarding their health and fertility.

Introduction:-

Turner syndrome (TS) is a genetic disorder that affects around 1 in 2500 newborn girls. A partial or full deletion of the X sexual chromosome is one of the extremely varied genetic abnormalities that define Turner's syndrome; it can manifest as a mosaicism with two or three distinct cellular lines or as a monosomy. A 45 XO karyotype is present in 50% of individuals with Turner's syndrome, whereas the remaining instances include mosaicism, X isochromosomes, or partial or whole Y chromosomes. Numerous abnormalities involving physical and cognitive development, particularly in the endocrine, cardiovascular, reproductive, auditory, and visual systems, are what define this illness. Fertility depends on the X chromosome's integrity. Accelerated germ cells are seen in TS.(Porcu et al., 2023a)

Genetic Basis and Pathophysiology:

Most Turner syndrome cases are not hereditary. The chromosome anomaly is a random occurrence during the development of the person's parent's reproductive cells when monosomy X is the cause. Nondisjunction is a cell division error that can produce reproductive cells with an abnormal number of chromosomes. Mosaic Turner syndrome is also not a genetic disorder. A random incident that happens during the cell division stage of the

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affected person's early fetal development is the cause. The outcome is that certain cells in an individual have the typical two sex chromosomes, while other cells have just one copy of the X chromosome.
A key characteristic of TS is ovarian failure brought on by gonadal dysgenesis, which starts in mid-gestation with increased germ cell apoptosis, leading to ovarian degeneration in the fetus and insufficient endogenous estrogen production. Girls with TS do not have the GH-modulatory effects or the local epiphyseal effects of endogenous estrogens, which probably contribute to their subnormal growth rates, decreased sensitivity to GH, and delayed skeletal maturation in early and mid-childhood. They also do not have the pubertal growth spurt and continue to grow slowly into their late teens and early 20s.(Fechner ditor, n.d.)

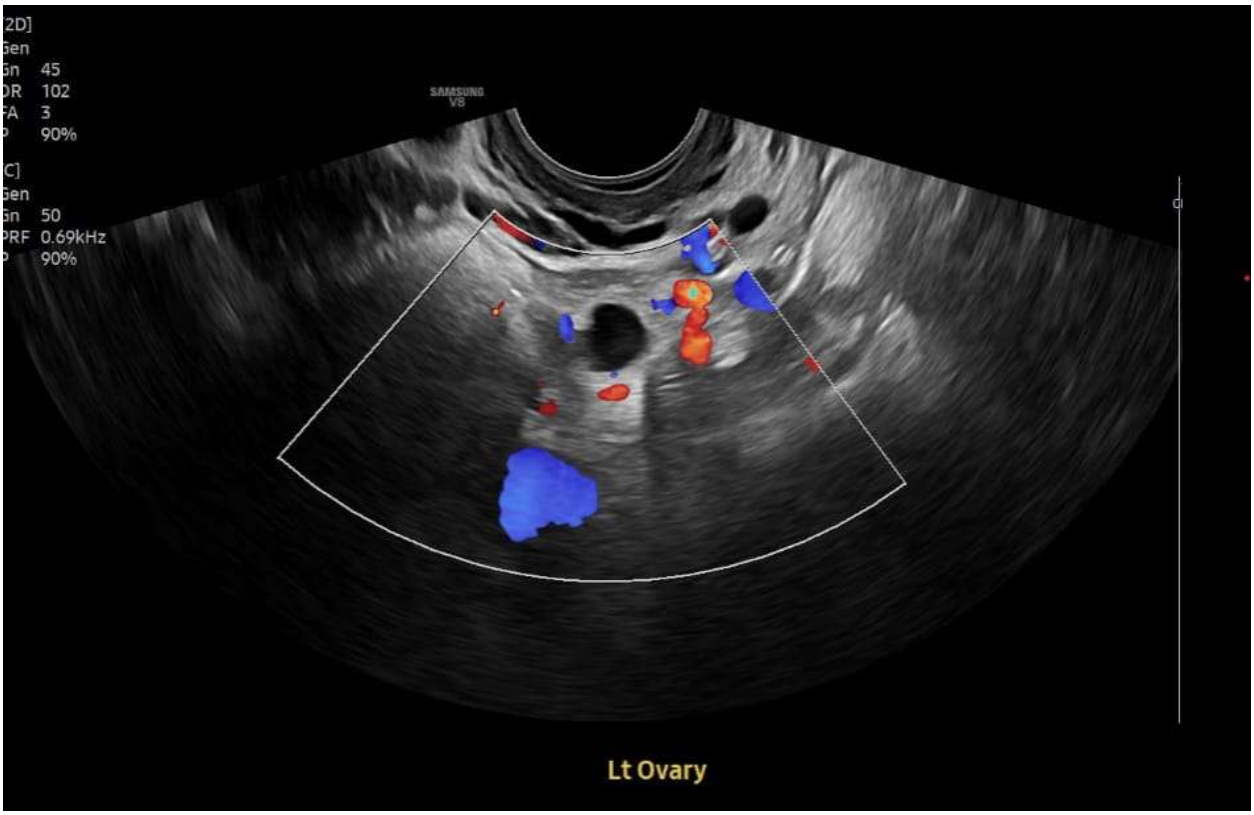
Turner's syndrome can be diagnosed prenatally using invasive techniques like amniocentesis and chorionic villous sampling. Ultrasonography features like increased nuchal translucency and cystic hygroma should raise suspicion. Abnormal maternal blood screening for alfa-fetoprotein, beta HCG, inhibin A, and unconjugated oestriol can also help, but karyotype confirmation is required.(Porcu et al., 2023a)

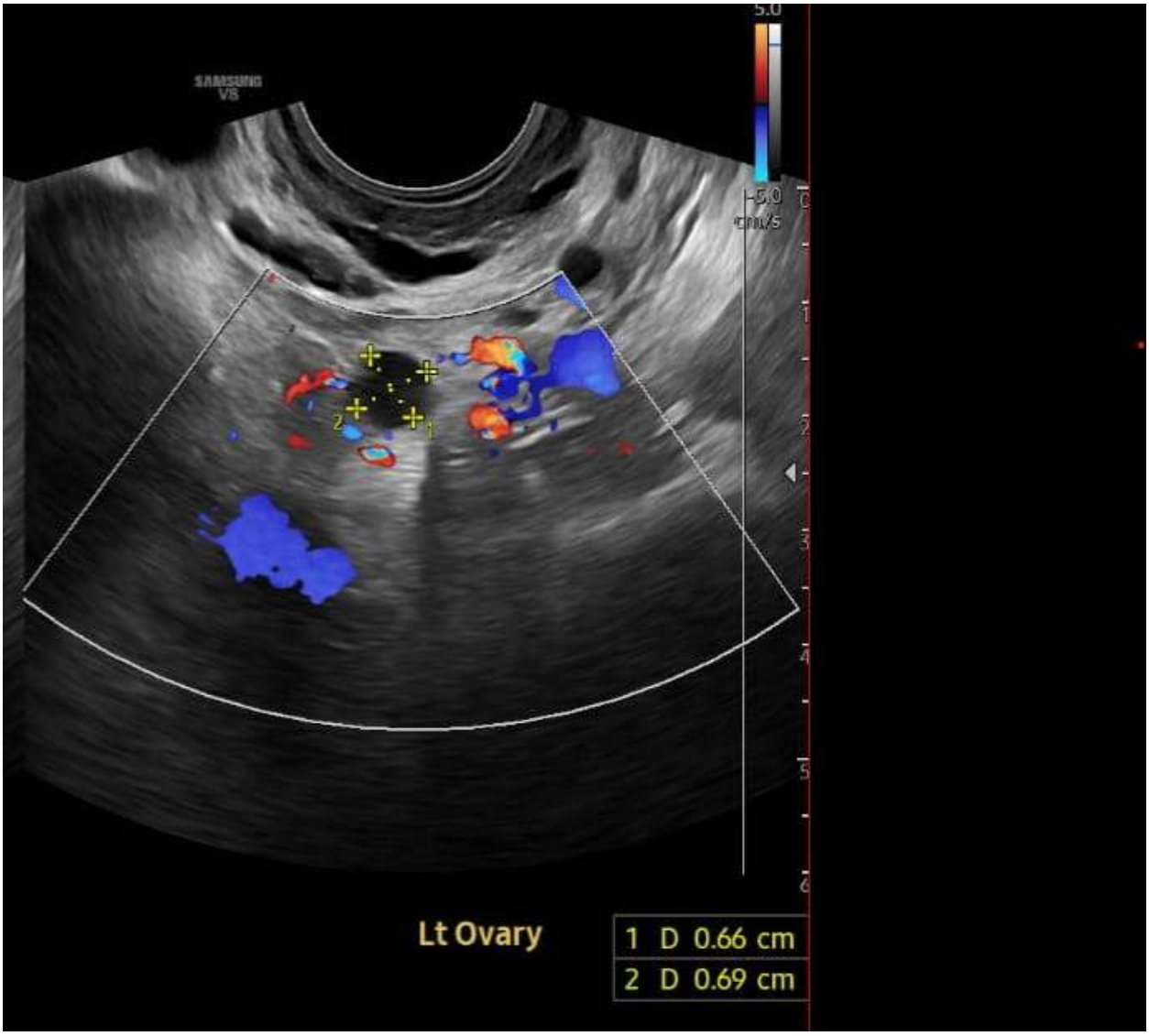
Early diagnosis of this condition enables the detection of pubertal development retardation, cardiac and auditory problems, and the establishment of an appropriate treatment to avoid consequences. Most TSs with mosaic karyotypes lack the typical TS traits. Should there be a history of small height, delayed puberty, lymphoedema, or aortic coarctation, karyotype investigation should be conducted.(Porcu et al., 2023a)

Case history:
Ms. S is 18 years old
College student
Married for 2 months
Diagnosed as a case of turner syndrome
Chiefcomplain sever short stature
Seen in an endocrinology clinic as a case of teenagerwith delay growth for investigation
Diagnosed as a case of Mosaics Turner Syndrome 2020
Her height was below 3.5, the standard
Had evidence of pubertal development but she was premenarchal
She didn't show up to her doctor till 2021 the work up completed including baseline pelvis and renal ultrasound
Started on growth hormone in March 2021, which she tolerated well, and her growth velocity has improved, catch up growth although her final adult height is short, discontinued growth hormone by herselfFebruary 2023
Based on her diagnosis as primary amenorrhea and FSH, LH, estradiol level she developed premature ovarian failure.
Started on estradiol patches 14 mcg daily in June 2021
Had menarche in January 2023 after she turned 16 years old
Then she stopped any medication and refused to received sex hormone due to misunderstanding from her family to the diagnosis
Then February 2025 she stared sex hormone replacement therapy with estradiol patches 0.075mg daily, and prometrium 200 mg for 12 days each month
Ms. S has poor compliance with her medication

Ms. S has the outcome of vaginal delivery without any complaints as per her and her records.
Normal sexual life, no pain with sexual intercourse
No significant findings in her medical, surgical or family history
Doing well in her college study
Review of system:
Had normal BMI
Endocrine: growth failure on growth hormone, since March2021, menarche January 2023. On full sex hormone replacement.
Genetics: mosaic turner syndrome
Other system: no abnormalities detected
Physical examination:
Vitally stable
General: short, no dysmorphic feature.
No midline defects

No palpable thyroid, no lymphadenopathy
Chest: clear
Breast well developed
cardiovascular system: normal sinus rhythm
abdomen: soft, no palpable mass or organomegaly
pubic hair present
genitalia: looks normal
extremities: wide carrying angle in the elbow joint
back, spine: normal
investigation:
FollicleStimulated Hormone: 6.1
Estradiol: 374
Anti Mullerian Hormone: <0.01
Inhibin: <10.0 ng/l which is postmenopause level
Thyroid Function Test: normal
Ultrasound Scan: both ovaries could not be visualized, however a small left adnexal cyst noted can be ovarian follicle
She was assessed by endocrinologists and cardiologists





Assessment:
turner syndrome with mosaic, with primary amenorrhea and premature ovarian failure on sex hormone replacement. Without cardiac manifestation
MS. S main concern is to have normal life and get pregnant.

Turner syndrome and fertility:
The rate of spontaneous pregnancy in TS ranges from 2% to 7%.
Girls with Turner syndrome have impaired oogenesis and folliculogenesis; follicular atresia was expedited, and a significant percentage of aberrant follicles was seen. Follicles are dependent on karyotype (mosaicism), age, and FSH and AMH concentrations. Indeed, there is a correlation between high levels of AMH and inhibin B and spontaneous puberty.(Porcu et al., 2023b)
The evaluation for potential fertility in girls with TS consists of:
- Evaluation of karyotype
- Evaluation of ovarian reserve (AMH and antral follicles count)(Porcu et al., 2023b)

Four techniques can be used to preserve fertility:

- heterologous ovarian tissue transplantation
- oocyte cryopreservation
- oocyte donation
- and embryo cryopreservation.(Porcu et al., 2023b)

risk associated with pregnancy in patients with turner syndrome should be discussed including:
preeclampsia, metabolic disorders, diabetes, placental insufficiency, and intrauterine growth restriction. Both spontaneous pregnancy and pregnancies resulting from oocyte donation carry a higher risk of fetal loss and congenital abnormalities.(Porcu et al., 2023b)

Discussion:-

Ms. S, who has early ovarian failure and mosaic Turner syndrome, wants to get pregnant. Given her poor ovarian reserve, a spontaneous pregnancy is improbable, underscoring the necessity of carefully weighing the risks of pregnancy when considering reproductive methods such as oocyte donation. Her patchy hormone treatment highlights the value of psychological support and patient education. To satisfy both physical and emotional requirements, the case highlights a multidisciplinary strategy that combines cardiology, reproductive medicine, and endocrinology. A patient-centered treatment plan and effective communication are essential to her health and competence to make well-informed choices about her fertility.

Conclusion:-

A multidisciplinary approach centered on well-informed decision-making, careful risk assessment (particularly endocrinology, cardiovascular), and strong psychosocial support is necessary in Ms. S's case, which involves mosaic Turner Syndrome, premature ovarian failure, and a desire for pregnancy, especially when considering oocyte donation.

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