VOLUME - 13, ISSUE - 02, FEBRUARY - 2024 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper | Obstetrics & Gynaecology

PREGNANCY IN A CASE OF PURE GONADAL DYSGENESIS: A RARE CASE STUDY

Dr. Suma Varsha Thota	Clinical head and Consultant, Ferty9 Fertility Center, Vijayawada, Andhra Pradesh, India.
Dr. Nallani Veneela*	Consultant, Ferty9 Fertility Center, Vijayawada, Andhra Pradesh, India. *Corresponding Author
Dr. Kanumuri Sindhu	Fellow, Ferty9 Fertility Center, Vijayawada, Andhra Pradesh, India.
Dr. Pandirla Swathi	Fellow, Ferty9 Fertility Center, Vijayawada, Andhra Pradesh, India.

ABSTRACT Gonadal dysgenesis is a group of heterogeneous disorders with very rare presentation. The clinical manifestation of disease includes primary amenorrhoea, failure to develop secondary sexual characters and also infertility. Here we are reporting a case of 29 year old phenotypic female who presented with primary infertility with 46, XX karyotype with hypoplastic uterus and streak ovaries (on imaging), with high gonadotropins level and low estradiol. After evaluation of the couple and proper counseling plan was made to go for cyclic hormone therapy for 3 months followed by donor oocyte and husband sperm IVF due to hypoplastic uterus with gonadal dysgenesis. 3 Day 3 embryos were transferred after hormonal therapy of 3 months. She was conceived with twins and later delivered 2 healthy babies at 9 months. We conclude that, pregnancy can be achieved even in gonadal dysgenesis patients after proper diagnosis of the condition and counseling the couple regarding the best treatment options available.

KEYWORDS : Gonadal dysgenesis, 46-XX,Amenorrhoea, Hypoplastic uterus, Hypergonadotropic hypogonadism, Oocyte donation.

INTRODUCTION:

Gonadal dysgenesis is a term used for a unique subset of disorders of sexual development[1]. It is an infrequent cause for primary amenorrhea (less than 1:100 000).

It is characterized by incomplete or defective formation of gonads and there is progressive loss of germ cells in the developing gonads of an embryo. 46, XX pure gonadal dysgenesis is a rare form of gonadal dysgenesis with wide clinical presentation. It may present with Primary or secondary amenorrhoea with or without normal secondary sex characteristics.

Case Study:

A 29 year-old married, sexually active girl presented to us with complaints of Primary infertility of 2 years. She attained menarche at 14 years after usage of withdrawal medicine and she gets her mensus only with withdrawal medicine.

There was no significant medical and surgical history. She was moderately built, with height of 156 cm and weight of 70 kg and BMI being 29 kg/m^2 . Her general physical and systemic examtination revealed no abnormalities. Abdomen was soft with no organomegaly.

Findings:

On genital examination external genitalia was normal, with ultrasound suggestive of hypoplastic uterus (2×3.8×2.6cm) with thin endometrium and streak ovaries measuring 1.3×1.3 cm.

Hormonal profile was done and it was suggestive of hypergonadotropic hypogonadism (follicle stimulating hormone: 80 IU/l, luteinizing hormone: 49 IU/l, serum estradiol <10 pg/ml, thryoid stimulating hormone: 2.2, Serum prolactin: 12.9 and serum AMH <0.01). Her karyotype was XX, 46. Thus a diagnosis of hypoplastic uterus with gonadal dysgenesis is made.

Semen analysis of the partner was normal.

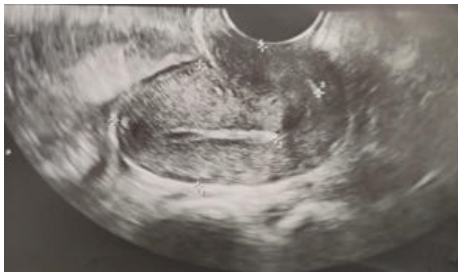

Fig 1: USG image of hypoplastic uterus

Management And Outcome Of Case:

The couple were counselled regarding the diagnosis and the appropriate treatment protocol. As the ovaries were not having any follicles oocyte donation was advised to the couple after 3 months of cyclic hormonal therapy. Benefits and risks were explained about the treatment protocol and treatment was initiated. Patient responded well with hormonal therapy and follow-up ultrasound showed uterine enlargement (2.6×6.0×3.2cm).

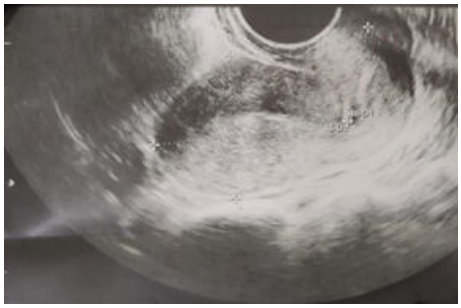

Fig 2: USG image after 3 months of hormonal therapy.

Seeing the response patient was started on frozen embryo transfer protocol medications and 3 day 3 embryos were transferred when endometrial thickness reached 7mm. Beta hcg was done 15 days post embryo transfer and it turned out to be positive. Later on ultrasound she was diagnosed to have

twin gestation. Antenatal care was given to her and later she delivered at term by cesarean section giving birth to a male and one female babies.

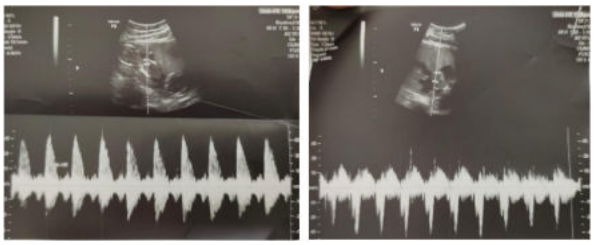

Fig 3:FHR of twin 1 and twin 2

DISCUSSION:

The genetic sex of an embryo is determined at fertilization. Bipotential gonads develop from the gonadal ridge, between 6-7 weeks of intrauterine life along with two sets of wolffian ducts (WD) and mullerian ducts (MD) developing from intermediate mesoderm. Y chromosome has testes determining effect (SRY gene). In the presence of SRY gene testes are formed and they cause regression of MD by secretion of Anti-mullerian hormone from sertoli cells. Androgens secreted from testes causes development of WD. Sex determination is irrespective of X chromosome, however two X chromosomes are necessary for ovary development. Absence of SRY gene causes indifferent gonads to develop into ovaries and MD persistence[2].

Dysgenetic gonads are characterized by variable degrees of immaturity or dysfunction, which can manifest in a wide range of genital ambiguity. Gonadal dysgenesis can be classified as either complete (CGD) or partial (PGD) depending on the gonadal morphology [3,4]. In complete gonadal dysgenesis no gonadal development occurs. These patients have a completely female phenotype due to the lack of any gonadal steroid production. Uterus could attain adult size with cyclic hormonal therapy. In terms of fertility, these people can conceive and have successful pregnancy with in vitro fertilization with donor oocytes[5].

CONCLUSION:

Thus an appropriate diagnosis of Gonadal Dysgenesis with appropriate management can help the patient in achieving pregnancy and delivering a healthy baby.

REFERENCES:

1. Lee PA, Houk CP, Ahmed SF: Consensus statement on management of intersex disorders: international consensus conference on intersex. Pediatrics 2006, 118:e488–e500.
2. Singh PB, Purwar R, Kumar P. Pure gonadal dysgenesis misdiagnosed as Mayer Rokitansky Kuster Hauser Syndrome: a case report. Int J Reprod Contracept Obstet Gynecol 2020;9:4756-8.
3. Ostrer H: 46,XY disorder of Sex development and 46.XY complete gonadal dysgenesis. Seattle: GeneReviewsSeries; 1993.
4. Fallat ME, Donahoe PK: Intersex genetic anomalies with malignant potential. Curr Opin Pediatr 2006, 18:305–311.
5. Thewjitcharoen Y, Veerasomboonsin V, Nakasatien S, Krittiyawong S, Himathongkam T. Misdiagnosis of Mullerian agenesis in a patient with 46, XX gonadal dysgenesis: a missed opportunity for prevention of gonadal dysgenesis in a 46,XX female. J Obstet Gynaecol Res. 2019;45(7):1386-90.