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A Rare Case of Coexistence of 46xx Gonadal Dysgenesis and Mayer Rokitansky Küster Hauser Syndrome

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ABSTRACT
Gonadal dysgenesis is a rare genetically heterogeneous disorder characterized by underdeveloped ovaries with consequent, primary amenorrhea, and hypogonadotropic hypogonadism. Mullerian agenesis or Mayer Rokitansky Küster Hauser syndrome is characterized by congenital aplasia of the uterus and the upper part (2/3) of the vagina in a woman with normal development of secondary sexual characteristics and a normal 46, XX karyotype.The association of Mayer-Rokitansky-Küster-Hauser syndrome and gonadal dysgenesis is very rare. We report an 18-year-old phenotypical female student with normal intelligence who presented with primary amenorrhea and underdeveloped secondary sexual characteristics. The hormonal evaluation revealed hypogonadotropic hypogonadism. Her karyotype was 46XX. Her parents were second-degree consanguinity. Family history was not significant.MRI of the pelvis revealed absent uterusfallopian tubes and ovaries, consistent with the findings of gonadal dysgenesis. There were no urinary or skeletal abnormalities.There were no other morphological malformations.The endocrine study revealed hypogonadotropic hypogonadism. The karyotype was normal, 46, XX.

I. INTRODUCTION

Gonadal dysgenesis is described as the absent or insufficient development of ovaries.[1] The patient with gonadal dysgenesis hasprimary amenorrhea and a lack of development of secondary sexual characteristics due to the inability of the ovaries to produce sex hormones. The karyotype in patients with gonadal dysgenesis can be 46XX, 45XO, mosaicism, or deletion of a certain part of the X chromosome.[1]

Mayer-Rokitansky-Küster-Hauser syndrome (MRKHS) is described as having an absent or hypoplastic uterus and upper two third of the vagina in phenotypically and karyotypically normal female with the incidence of approximately 1 in 5,000 new-born girls.[2] The female with MRKHS has normal secondary sexual characteristics due to normally functioning ovaries. It is the second most common cause of primary amenorrhea. Very few cases have been reported with the co-existence of MRKH syndrome and Mullerian agenesis. [1-2]. We report here a case presented to our outpatient department.

II. CASE REPORT

An18-year-old female was evaluated in our clinic because of primary amenorrhea and poor breast development. She is a daughter of second-degree consanguineous parents. Her birth event and perinatal and neonatal periods were uneventful. Her growth and development were normal with normal intelligence. Her developmental history was also normal. Atpresentation, her height was 147 cm, weight30 kg, and BMI 13.8. On examination, there was no facial dysmorphism, Axillary hair was dark and coarse (Wolfs Dorf stage 3), Breast development was infantile (Tanner stage 1),Pubic hair was Curly&black (Tanner stage 4),Normal female external genitalia with an intact hymen. A bimanual rectal examination revealed no palpable uterus& cervix. No skeletal deformity was found. Her blood pressure was 110/70 mmHg in both arms. Her renal function tests and liver function tests were normal. Her FSH and LH levels were elevated, estradiol levels were low and she was hypothyroid with normal TSH levels. Ultrasound abdomen revealed an absent uterus and absent ovaries. On MRI uterus was absent and vaginal lengthwas 2cm and ovaries were not visualized. No otherGenito urinary andSkeletal malformations. Karyotypingrevealeda karyotype of46xx and Fish analysis showedno Ychromosome.The patient was advised to undergo diagnostic laparoscopy, but the patient was unwilling.

Management

The patient was started on oestradiol valerate 2mg per day to trigger the development of secondary sexual characteristics and prevent bone loss.She was scheduled for an increment in dose every 6 months to mimic normal pubertal progress until the adult dose is reached over 2to3 to years. She was also prescribed calcium and vitamin D3 and referred to an endocrinologist regarding treatment with Growth-hormone.

The patient and her parents were counselled regarding marriage and fertility options were explained.

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III. DISCUSSION

In reported similar 23 cases in the literature,5 wererereported with a history of consanguinity. In 11 cases ovaries were absent, while in 9 cases ovaries were present but were dysgenetic. In 15 case reports,the uterus was absent & in other present but hypoplastic. Urological abnormalities are present in 6 cases. Skeletal abnormalities were present in 6 cases. All the studies revealed that 46, XX gonadal dysgenesis with Mullerian agenesis commonly presents with normal female phenotype but absentuterus and ovaries with primary amenorrhoea and hyper-gonadotropic hypogonadismwith or without somatic abnormal malformations [2].

IV. CONCLUSION

The association between two entities is considered may be coincidental. There may be a mutation or Deletion of common genes involved in the development and migration of germ cells and Mullerian derivatives. Some parts of the X chromosome deletion or mutation ofthe Regulatory gene may be responsible for the coexistence of ovarian agenesis and MRKH.

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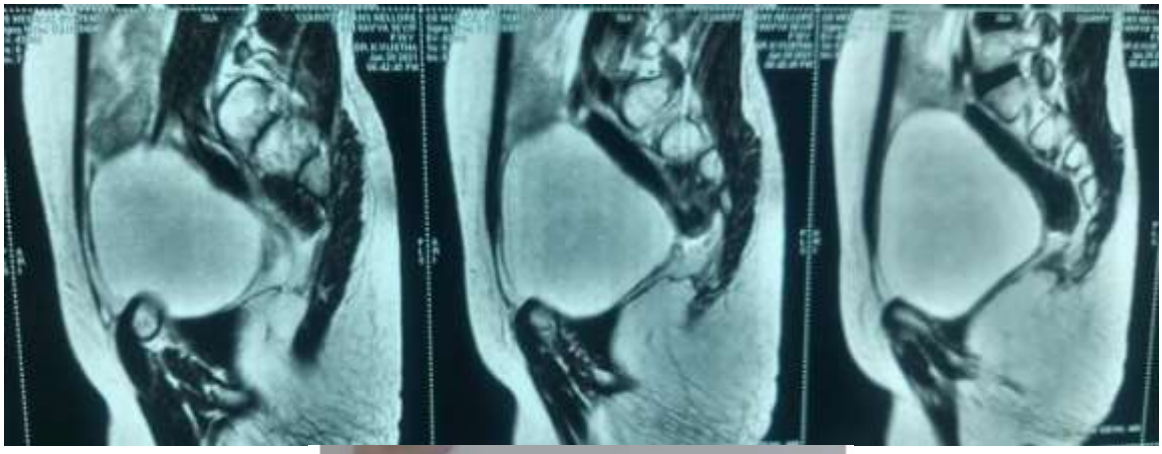

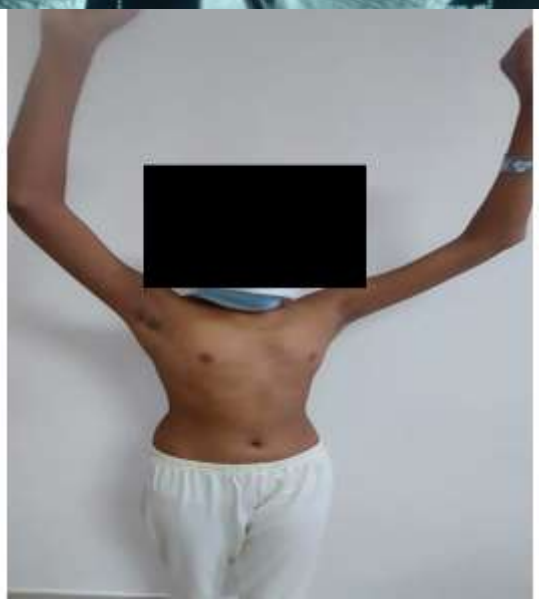

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