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Mixed Gonadal Dysgenesis – A Case Study

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Abstract

Gonadal dysgenesis refers to various clinical conditions in which abnormal development of the fetal gonad is present. It consists of 46 XY gonadal dysgenesis, mixed gonadal dysgenesis, and 45 X turner syndrome. Mixed gonadal dysgenesis is a condition of unusual and asymmetrical gonadal development leading to unassigned sex differentiation. A number of differences have been reported in the karyotype, most commonly a mosaicism 45 X / 46 XY.1 We herein report an interesting and rare case of mixed gonadal dysgenesis in a 23-year-old nulliparous unmarried woman.

Case: A 23-year-old nulliparous unmarried woman with primary amenorrhea with short stature, webbed neck, breast developed according to Tanner's stage was presented to our clinic. She had normal external genitalia with axillary and absence of pubic hair. The evaluation of her uterus on ultrasound revealed a hypoplastic uterus measuring 2.6 x 1.4 x 0.9 cm with poorly formed endometrium and myometrium with bilateral ovaries being small and hypoplastic. Her follicle-stimulating hormone (FSH), testosterone, luteinizing hormone, and anti-mulleins hormone levels was 120 IU/L, 15 ng/dL, 24.5 IU/L and 0.02 ng/Ml respectively. Karyotype test showed mosaicism mixed gonadal dysgenesis, i.e., 45 X0 and 46 XY. She underwent laparoscopic bilateral salpingectomy (gonadectomy and salpingectomy).The post-operative period was uneventful. Intraoperatively streak gonads with elongated tubes were visualized. An infantile uterus with no pelvic abnormality was also observed intraoperatively. The histopathology report showed a cut dissection of ovaries.

Discussion: Mixed gonadal dysgenesis (MCG) is a rare intersexual disorder, characterized by the presence of a testis and a contralateral streak gonad; in some cases the contralateral gonad may be rudimentary not having differentiated into an ovary or into a testis and in other cases it may be absent. On evaluation of 23-year-old nulliparous unmarried women's uterus on ultrasound revealed a hypoplastic uterus measuring 2.6 x 1.4 x 0.9 cm with poorly formed endometrium and myometrium with bilateral ovaries being small and hypoplastic. Karyotype investigation revealed mosaicism mixed gonadal dysgenesis, i.e., 45 X0 and 46 XY. Johansen and co-workers reported structural rearrangement of the Y chromosome in 63% of mixed gonadal dysgenesis patients.2,3 Arora R et al reported that in 16-year-old person, karyotype analysis revealed 46XY karyotype and diagnosed with 46XY mixed gonadal dysgenesis.4 With this study, treatment recommended was laparoscopic bilateral salpingectomy (gonadectomy and salpingectomy). Sheela S et al also found similar kind of presentation where right sided gonads and adjacent tubal structures were visualized laparoscopically and performed gonadectomy.5 Similarly, Yadav P et al reported right streak ovary, left sided fallopian tube and streak ovary were noted in exploratory laparoscopy and later it was excised by doing gonadectomy.6.

Conclusion: Gonadectomy was done as a prophylaxis measure in a 23-year-old nulliparous unmarried woman presented with gonadoblastoma or dysgerminoma. It was learned from the experience of the present study that patients with mixed gonadal dysgenesis have a different presentation and variant of chromosomal abnormalities. A multidisciplinary approach is essential for early diagnosis and proper management of patients diagnosed with rare mixed gonadal dysgenesis in order to prevent mental and social sequels

Biography

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