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GONADAL DYSGENESIS WITH MAYER-ROKITANSKY-KUSTER-HAUSER SYNDROME

Radio diagnosis

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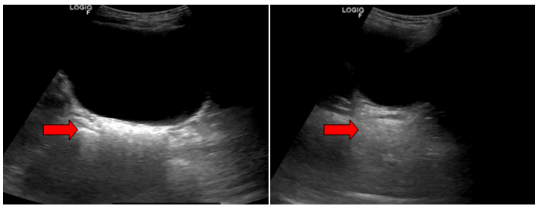
ABSTRACT

The association of gonadal dysgenesis with Mullerian tract anomalies is extremely rare and appears to be coincidental, independent of chromosomal anomalies. Gonadal dysgenesis is characterized by underdeveloped ovaries with consequent, impuberism, primary amenorrhea, and hypergonadotropic hypogonadism. Mullerian agenesis or Mayer-Rokitansky-Kuster-Hauser (MRKH) 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. We report a case of 16 year old female patient who presented with Primary amenorrhea and Impuberism. Endocrine study revealed, hypergonadotrophic hypogonadism with normal level of TSH. The pathogenesis of the association of both is still unknown. The treatment is based on hormonal therapy, however the fertility is unfortunately compromised.

KEYWORDS

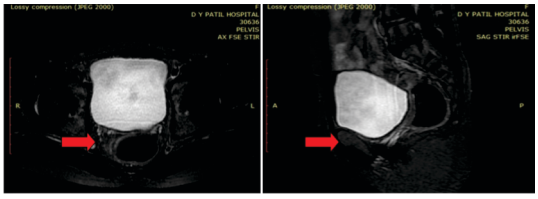
16 years old female patient come with the complaints of Primary amenorrhea and Impuberism. Patient was examined by gynecologist, approximately 1 cm vagina ending into blind pouch was found. Bimanual pelvis and rectovaginal examination revealed absence of cervix and uterus. First the patient was advised USG abdomen and pelvis On USG uterus and ovaries were not visulaized. The diagnosis was ? Absent uterus and ovaries ? Atrophic uterus and ovaries. Patient was advised for MRI to rule out mullerian duct agenesis. Chromosomal analysis from peripheral blood and endocrine study including ovarian and thyroid evaluation was also performed.

On USG: No uterus indentified behind bladder.

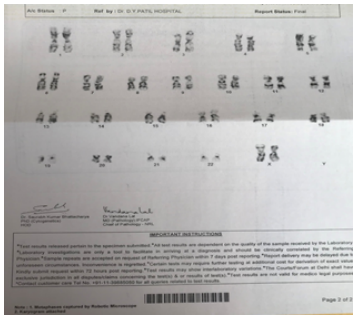


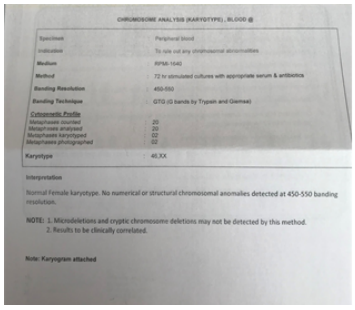
On MRI-

1) Axial plane cut showing bladder and rectum without interposition of uterus. (on right)
2) Sagittal plane cut: absence of uterus and ovaries. (on left)



On chromosomal analysis: No Y chromosome detected.





FINDINGS

- Endocrine study revealed, hypergonadotrophic hypogonadism (LH-13.0mIU/ml, FSH-50.6mIU/ml and oestradiol-11.3pg/ml) with normal level of TSH.
- On chromosomal analysis (karyotype) from peripheral blood the karyotype was 46XX, no Y chromosome material was detected.
- On USG uterus and ovaries were absent.
- MRI study revealed complete absence of the uterus, cervix and both ovraies.

DIAGNOSIS

Gonadal dysgenesis with associated mayer-rokitansky-kuster-hauser syndrome.

DISCUSSION

- Our patient had two anomalies affecting reproductive performance, 46, XX gonadal dysgenesis and MRKH.
- 46, XX gonadal dysgenesis is a primary ovarian defect leading to premature ovarian failure in otherwise normal 46, XX females due to failure of the gonads to develop or resistance to gonadotropin stimulation. Prevalence is unknown but is thought to be less than 1/10.000. Patients are born as females without ambiguity. However, affected individuals present during adolescence or young adulthood with either delayed or absent puberty resulting in primary amenorrhea. The internal and external genitalia are normally developed. Although the underlying etiology of ovarian dysgenesis remains unknown in most cases, several genes have been implicated including homozygous or compound heterozygous inactivating mutations of the follicle-stimulating hormone receptor gene (FSHR), mutations in the BMP15 gene, and mutations in the NR5A1 gene.
- MRKH syndrome is characterized by Müllerian duct structures agenesis, vaginal atresia, rudimentary or absent uterus with normal ovaries and Fallopian tubes in a female with normal genetic (46 XX), phenotypic and developmental features. The

prevalence has been reported as 1 in 4.500 female births.

- The etiology of MRKH syndrome is unknown but it is believed that embryological development is interrupted during the sixth or seventh week of gestation. Since the mesonephros (which give rise to kidneys), the Mullerian ducts, and the skeleton all originate from the mesoderm, it is believed that a deleterious event occurs during this phase of gestation, giving rise to the abnormalities seen in the MRKH syndrome.
- Estrogens may influence development of the Mullerian ducts. Absence of anti-Mullerian hormone is also essential for its development. The existence of activating mutations of either the gene for the anti-Mullerian hormone or the gene for the anti-Mullerian hormone receptor, and the lack of estrogen receptors during embryonic development have been hypothesized to cause MRKH syndrome.
- The association of gonadal dysgenesis with Mullerian tract anomalies is extremely rare. It was first described by McDonough *et al*. in a girl with gonadal dysgenesis, duplication of the Mullerian duct system, and a 46, XX karyotype. The coexistence of these two anomalies was then reported in patients with 45, X0; 46, XX; 46, XY; 45, X/46XX; 45,X/46X/del(X) (p22.2); 46, Xi (Xq) karyotypes.
- Theoretically, an undifferentiated gonad could produce anti-Mullerian hormone in earlier embryologic period, leading to a subsequent regression. But this hypothesis cannot be valuable without the presence of the chromosome Y. The chromosomal analysis of our patient demonstrated the absence of chromosome Y.
- An association between these two conditions is very exceptional and appears to be coincidental independent of chromosomal abnormality.

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