International Scholars Journals

International Journal of Obstetrics and Gynecology ISSN: 2736-1594 Vol. 9 (2), pp. 001-003, February, 2021. Available online at www.internationalscholarsjournals.org © International Scholars Journals

Case Report

Is Mullerian Agenesis a common association with Mosaic Turner Syndrome, a case report

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Abstract

Accepted 06 October, 2020

We report a young girl who presented with delayed menarche and uterine absence as detected by ultrasonography and Magnetic Resonant Imaging. Evaluation of the case revealed Mosaic Turner Syndrome (45X/46XX). Early loss of gonadal function is the usual presentation of Turner Syndrome and will be thought-about in any case of delayed puberty. Follow-up imaging for apparently absent uterus particularly once begin of sex hormone replacement medical aid to verify a traditional uterus. It is traumatic for both the patient and her family to be informed by the absence of her uterus which has vital implications for long-run fertility.

Keyword: Mosaic Turner Syndrome, absent uterus, primary amenorrhea.

CASE REPORT

A young girl 14 years old presented to outpatient gynecology clinic at Benha Teaching Hospital with primary amenorrhea and delayed puberty (stage I Tanner for both breast development and pubic hair), this age is considered late as her female relatives (mother and sisters), where their age of menarche was between 10-12 years.

Physical examination revealed no abnormalities, investigations were requested and revealed; hyper-gonadotropic hypogonadism with (FSH) follicle-stimulating hormone 13m IU/ml (menopausal range), (LH) luteinizing hormone 10 mIU/ml (menopausal range), and estradiol 24 pg/ml (pre-pubertal range), serum prolactin 6. 11 ng/ml, serum TSH 39 mIU/ml, anti-thyroid antibodies (Anti thyroglobulin thyroid peroxidase antibodies were negative) antinuclear antibodies were negative & her Echo was free from any abnormality, a pelvic ultrasound revealed an absent uterus, both ovaries were traditional and visualized in their traditional site, kidneys and other pelvic organs showed no To verify the absence of the uterus; MRI was requested and reported an entire absence of uterine tissue above the vagina denoting uterine agenesis (uterine aplasia) with both ovaries within the pelvic cavity. Patients were referred for cytogenetic testing that showed revealed a pattern coinciding with Turner Syndrome 46XX (77%) 45X0(23%) Counseling the patient and her family about the choice of her management and estrogen replacement as a line of therapy, they agree to start treatment with low dose oral conjugated estrogen therapy for 25 days and in conjugation with progesterone (norethisterone acetate 10 mg) in the last 14 days for 12 months on this protocol withdrawal, bleeding To confirm its presence; an Ultrasound examination was done and it confirmed the presence of a small uterus (5× 4× 3) cm.

DISCUSSION

This is a case report of a patient with delayed puberty, amenorrhea, and absent uterus with average-sized ovaries by ultrasound and MRI. Cytogenetic studies reveal mosaic Turner syndrome which is a genetic disorder that only affects women. Approximately 1:2,500 babies underwent monosomy (45, X), 5-10% had a copy of the long arm X (46X) (Xqi) (isochromosome), and most others presented mosaicism for 45X with one or more additional this condition

can cause various physical, emotional, and educational disorders. This occurs when one of the two chromosomes found in women is missing or lacks (1)

The clinical picture of Turner syndrome:

A Turner case shows certain physical characteristics includes;
Slow growth rate (< 10 %) with the resultant short stature, short “fourth metacarpals, and metatarsals”, hypermetropia “farsightedness”, defenses, low set hair, small mandible, abnormal ear, webbed neck, “cubits valgus”, “congenital hydrops”(puffy back of toes and fingers), “hyper-convex nails”, “broad chest with widely spaced nipples”, “pigmented nevi.”
In addition to “horseshoe kidney, double or cleft ureters”.
Orthodontic evaluation reveals; “flattened cranial base angle reduced posterior cranial base length high arched palate, narrow maxilla Micrognathia”
Increased risk of autoimmune disorders and autoimmune thyroiditis.
Cardiovascular disorders include;” hypertension”, “bicuspid aortic valve defect”,” abnormal aortic valve” “ascending aortic dilation aneurysm.”
Also, premature gonad failure, increased risk of diabetes is characteristic of those cases. (1)
There is typically an early loss of gonadal function in Turner Syndrome (6) the uterus is usually present however it's going to be smaller in size (2, 3). To the contrary of that our case was presented by premature gonadal failure with no Mullerian development, we tend to review the causes of the absent female uterus, and few syndromes are related to Mullerian agenesis, to verify or rule out the diagnosis of Turner. “ Mayer-Rokitansky-Küster-Hauser syndrome” (MRKHS) is a congenital malformation that is characterized by normal Karyotype (46, XX), and absent female Mullerian duct (uterus, tubes, and vagina). However, not like Turner Syndrome where ovarian function is preserved in these patients (4), this didn't coincide with our patient as cytogenetic study (45xo).

Key points;

- Early loss of ovarian function is a typical presentation of” turner syndrome”.
- When primary gonadal insufficiency is present in addition to an apparently absent uterus as detected by ultrasound examination, “turner syndrome” should be excluded.
- Follow-up of any case with an uterine absence after hormonal replacement therapy is mandatory to confirm the presence of a normal uterus.
- The absent uterus is psychologically traumatic for patients and families, accurate diagnoses are important. (6).

Similar to this case, a 17 years old girl presented with “primary amenorrhea” differed from our patient as she had absent both ovaries and uterus on ultrasound and MRI (5).
Karyotyping reveals “turner syndrome", and like our case, six months estrogen therapy MRI revealed a (4.5 cm) long uterus (5).
In Turner syndrome the ovaries typically are atretic, which was not the case of our patient which showed normal size ovaries with premature loss of ovarian function, this led us to consider our case as an atypical presentation of mosaic Turner syndrome.
Another atypical presentation of Turner syndrome is the absent uterus as usually, the uterus is present in turner cases, in support of our diagnosis a case report of "a 14-year-old girl" presented with pubertal developmental delay in association with an absent uterus on ultrasound examination which is considered as “Turner Syndrome.”

CONCLUSION

This is a case report of a female patient with atypical mosaic “Turner syndrome” with an absent uterus shown to appear after hormonal replacement therapy.

For which we recommended:

Every case of “mosaic Turner syndrome” should be examined by Ultrasound and MRI for association with Mullerian agenesis and follow up every 3-6 months.
Careful evaluation of “under- estrogenized” patient by ultrasound and MRI for uterine which should be repeated for assessment of hormonal therapy efficacy.

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