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Case Series

Three cases of primary amenorrhoea

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ABSTRACT

Three interesting series of cases in young ladies who reported with history of primary amenorrhoea with surprisingly different presentation, symptoms, clinical signs and findings requiring two entirely different algorithms which this article attempts to explain to emphasize the art of history taking and clinical examination in women. This article attempts to present the importance of effective communication to the patient and understand different concerns in different women, communities, age groups and a plethora of presentation.

Keywords: Amenorrhoea, Infertility, Medical genetics, Reproductive endocrinology, Abdominal gonads, Hermaphrodites, Ovotestis, Fragile X

INTRODUCTION

Amenorrhoea is generally defined as the absence of menstruation in a female of reproductive age. It can be classified as either primary or secondary amenorrhoea. Primary amenorrhoea is the failure to reach menarche (i.e., the first menstrual cycle) during normal development. It is clinically diagnosed when there is no history of menstruation by the age of 15 years or 3 years after menarche. Patients meeting the criteria for primary amenorrhoea warrant an evaluation. Additionally, an assessment for delayed puberty is indicated in adolescents aged 13 years and younger without initial breast development or other secondary sex characteristics (e.g., pubic and axillary hair).[1] Most of the underlying causes of primary amenorrhoea can be classified into general groups: anatomic and sexual development abnormalities, ovarian insufficiency, hypothalamic or pituitary disorders, and other endocrine gland disorders. Physiology and medications may also cause primary amenorrhoea; however, they are more commonly associated with secondary amenorrhoea. The initial work-up usually includes a comprehensive history and physical examination, a urine pregnancy test, serum hormone testing, and pelvic imaging. Additional testing may also be indicated based on the clinical presentation.[1,2] Treatment depends on underlying etiology and may include lifestyle interventions, hormone therapy/other medications, surgery, and mental health services. Therefore, this activity for healthcare professionals is designed to enhance the learner's competence when managing primary amenorrhoea, equipping them with updated knowledge, skills, and strategies for timely diagnosis, effective interventions, and improved care coordination, leading to better patient outcomes.

CASE SERIES

Case 1

A 17-year-old unmarried girl brought by mother with chief concerns of absence of menarche. No other associations, GPE-was WNL, Height was 155 cm and weight was 54 kg with no androgenizing features or coarse facies. She had attained thelarche, adrenarche, pubarche, with absence of menarche. She was not on any chronic medication. On examination all her secondary sexual characters were well developed, abdominal examination did not reveal any mass/lump. Examination of external genitalia revealed a blind vagina with dimple and no bulge. TVS could not be

performed, bedside TAS (pelvis) on full bladder was performed where presence of uterus could not be demonstrated, ovaries were present with follicles. Systemic examination was unremarkable. Provisional diagnosis was MRKH syndrome was made; confirmation of which required detailed anatomical survey, normal female karyotype, normal endocrinological values in HPG/TOPHA axis. All biochemical, endocrinological and haematological parameters were within normal limits, karyotype was 46/XX, USG and CEMRI abdomen and pelvis revealed blind/absent vagina, small infantile hypoplastic T shaped uterus, cervix could not be identified, normal ovaries with peripheral follicles; fallopian tubes and adnexa absent. On retrospective questioning of mother revealed taking unknown medication from village quack for undiagnosed bleeding commencing from end of first trimester till around 36 weeks. A possible DES association was made clinically and patient referred to tertiary care centre for vaginoplasty, possible metroplasty and HRT for her sexual fulfillment as a biological woman. She was also counseled regarding other reproductive options and fertility issues which included surrogacy. Type of malformation is always confirmed by gold standard which remains diagnostic laparoscopy. Patient and her parents agreed to share CEMRI images.

Case 2

A 16-year-old unmarried girl brought by mother with chief concerns of absence of menarche. No other associations, GPE- revealed tall well-built girl height was 170 cm and weight was 60 kg with sharp features and feminine body habitus. She had attained thelarche, adrenarche, pubarche as per patient; however, menarche was absent. She was not on any chronic medication. On examination her breasts were tanners stage II, external genitalia revealed small clitoris, flat mons pubis, infantile labia and vaginal orifice with complete absence of rugosities and pubic hair. Body hair and axillary hair were scant and fine. Abdominal examination did not reveal any mass/ lump. TVS was not performed as patient did not consent. Bedside TAS (pelvis) on a full bladder was performed where presence of uterus could not be demonstrated, ovaries were absent. Systemic examination was unremarkable. Provisional diagnosis was DSD/ MGD; confirmation of which required detailed anatomical survey, karyotyping, endocrinological evaluation of HPG/TOPHA axis, laparoscopy with gonadal biopsy. All biochemical, and haematological parameters were within normal limits, FSH and LH levels were high, testosterone levels were low. Karyotype was 47/XXY, USG and CEMRI abdomen and pelvis revealed infantile vagina, absent uterus, and bilateral streak abdominal gonads suggestive of ovotestis. Patient was referred to higher centre for laparoscopy, intra-op frozen section of streak gonads for ovum harvesting for future fertility if possible in DSD; and gonadectomy in absence of primordial follicles to prevent future malignancy in MGD. She returned for followup after bilateral gonadectomy from a premier research institute and histopathology confirmed Bilateral Ovotestis with absence of primordial follicles and no malignancy. She was placed on HRT and is under counseling for gender dysphoria. These patients with streak gonads and ovotestis are referred as hermaphrodites/intersex. Those with both gonads and both genitalia are called true hermaphrodites. Patient and her parents declined to share images.

Case 3

A 34-year-old P1L1, male child (IVF-ET) (DO), reported with complaints of presumed secondary amenorrhoea. She was already on HRT with withdrawal spotting of scanty drops requiring only one sanitary towel around once a year. On detailed history taking and evaluation of all previous documents she revealed she did not attain menarche and was placed on HRT at 18 years prior to matrimony. She failed to conceive after 7 years of regular cohabitation and reported to fertility specialist. She was evaluated completely including laparoscopy and was diagnosed as primary ovarian insufficiency (POI) after six cycles of OI on HRT along with protocol for super-ovulation and was advised donor ovum harvesting for IVF-ET to save on time and cost. Her initial endocrinological workup revealed elevated FSH, and low AMH. All other biochemical, haematological and endocrinological parameters were normal. She had primarily reported for possibility of ART for her own biological child. Her height was 152 cm and weight 72 kg, she had mild hirsuitism, her anatomical survey and TVS were normal. She complained of mood swings, vasomotor symptoms, social anxiety, lack of libido, depression, lack of attention and forgetfulness. She was evaluated again and her HPG/TOPHA axis revealed very low AMH. All other biochemical, haematological and endocrinological parameters including serum cortisol were WNL. She was tested for fragile X mutation (FMR1 gene) which revealed expanded number of CGG repeats value of 86. She was clinically diagnosed as a case of fragile X-associated POI (FXPOI) and was advised to continue HRT and placed on antidepressants. She was advised genetic, psychiatric and neurophysician counseling to address her depressive episodes and prevent neurodegenerative disorders and ataxia. Full spectrum of her disease could not be assessed. Patient was lost to follow up and consent could not be obtained.

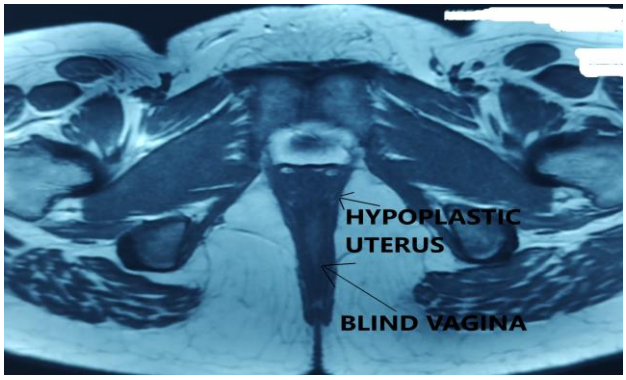

Figure 1: MRI showing hypoplastic uterus and blind vagina in MRKH syndrome.

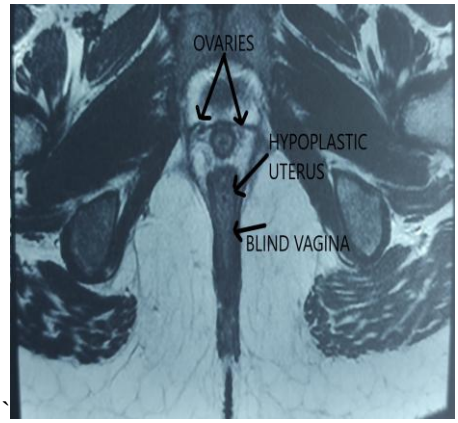

Figure 2: MRI image of MRKH syndrome showing hypoplastic uterus, blind vagina and small bilateral ovaries in pelvis.

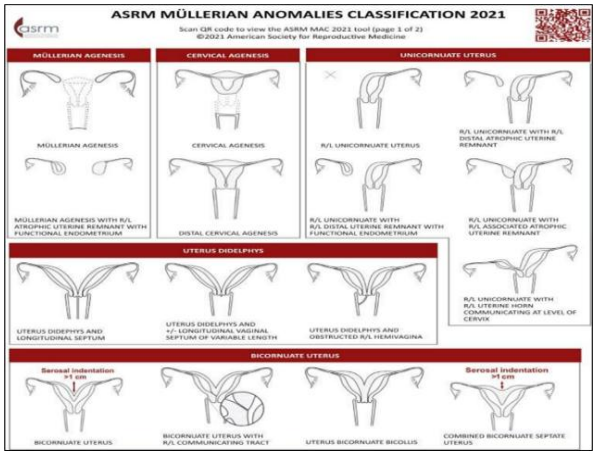

Figure 3: ASRM classification 2021 (detailed representation in QR code).[13,14]

Table 1: Clinical and diagnostic findings of primary amenorrhoea aetiologies.[9]

Etiology	Thelarche (T) and adrenarche (A)	FSH	Müllerian structures	Other findings
Gonadal dysfunction	T: Absent A: Absent	High	Present	Prepubertal appearance
Müllerian agenesis	T: Present A: Present	Normal	Absent	Associated with other urogenital anomalies
Obstructive vaginal anomaly	T: Present A: Present	Normal	Present	Cyclic pelvic pain
CDGP	T: Absent A: Absent	Low	Present	Delayed bone age Consistent family history. Resolves with time
FHA	T: Absent or delayed A: Absent or delayed	Low	Present	Delayed bone age History of eating disorder or vigorous exercise
PCOS	T: Present A: Present	Normal	Present	Hirsutism, acne Polycystic ovaries on imaging
CAIS	T: Present A: Absent	Normal	Absent	Well-developed breasts

T: thelarche, A: adrenarche, FSH: follicle-stimulating hormone, CDGP: constitutional delay of growth and puberty, FHA: functional hypothalamic amenorrhoea, PCOS: polycystic ovary syndrome, CAIS: complete androgen insensitivity syndrome, USG: ultrasound.

Table 2: ASRM classification.[13,14]

Classes	Name	Description
I	MRKH syndrome Mullerian agenesis	Hypoplasia of fallopian tubes, cervix, uterus or vagina.
II	Unicornuate uterus	No rudimentary horn. Non-cavitatory rudimentary horn. Cavitatory rudimentary horn (communicating/non-communicating).
III	Uterus diadelphys	Two separate uterine horns and two cervices.
IV	Bicornuate uterus	Complete septum extends from uterus to the internal and external os. Partial septum is confined to fundal region. Two separate uterine horns and single cervix. Fundal depression separating the two horns >1 cm. Intercornual distance >4 cm.
V	Septate uterus	Complete-septum extends from uterus to the internal os. Partial-septum does not reach the internal os. Fundal depression separating the two horns >1 cm. Intercornual distance >4 cm.
VI	Arcuate uterus	Normal variant. Endometrial cavity is not divided. External contour of the uterus is flattened or minimally concave.
VII	T- shaped uterus	In utero exposure to DES.

Table 3: VCUAM classification.[17]

Organ	Description
Vagina (v)	0-Normal 1a-Partial hymenal atresia 1b-Complete hymenal atresia 2a-Incomplete septate vagina <50% 2b-Complete septate vagina 3-Stenosis of the introitus 4-Hypoplasia 5a-Unilateral atresia 5b-Complete atresia S1-Sinus urogenitalis (deep confluence) S2-Sinus urogenitalis (middle confluence) S3-Sinus urogenitalis (high confluence) C-Cloacae
Cervix (c)	0-Normal 1-Duplex cervix 2a-Unilateral atresia/aplasia 2b-Bilateral atresia/aplasia
Uterus	0-Normal 1a-Arcuate 1b-Septate <50% of the uterine cavity 1c-Septate >50% of the uterine cavity 2-Bicornuate 3-Hypoplastic uterus 4a-Unilaterally rudimentary or aplastic 4b-Bilaterally rudimentary or aplastic
Adnexa (~ including tubal malformation if appropriate)	0-Normal 1a-Unilateral tubal malformation, ovaries normal 1b-Bilateral tubal malformation, ovaries normal 2a-Unilateral hypoplasia/gonadal streak~ 2b-Bilateral hypoplasia/gonadal streak~ 3a-Unilateral aplasia 3b-Bilateral aplasia
Other malformations	0-None R-Renal S-Skeletal C-cardiac N-neurologic

+(O-Other) # (U-Unknown) (included in all individual descriptions)

DISCUSSION

Amenorrhoea is abnormal uterine bleeding characterized by the absence of menstruation in a female of reproductive age.[1] Amenorrhoea can be classified as either primary or secondary amenorrhoea. Primary amenorrhoea is defined as having no history of menstruation by the age of 15 years or 3 years after thelarche; secondary amenorrhoea is defined as the absence of menses for ≥3 months in a woman with previously regular menstrual cycles or ≥6 months in any woman with at least one previous spontaneous menstruation.[1,2] The median age of menarche is approximately 12.4 years, though this varies somewhat by patient-specific factors (e.g., ethnicity, weight, and nutrition status).[3-5] Menarche typically occurs within 2 to 3 years of initial breast development, which occurs between the ages of 8 and 10 years, known as thelarche.[6,7] Patients meeting the criteria for either primary or secondary amenorrhoea warrant an evaluation. However, an evaluation for delayed puberty is also indicated in adolescents aged 13 years with primary amenorrhoea and no breast development or other secondary sex characteristics (e.g., pubic and axillary hair).[8]

Most underlying causes of primary amenorrhoea can be classified into general groups: anatomic and sexual development abnormalities, ovarian insufficiency, hypothalamic or pituitary disorders, and other endocrine gland disorders. Physiology and medications may also cause primary amenorrhoea; however, they are more commonly associated with secondary amenorrhoea. The

initial work-up usually includes a comprehensive history and physical examination, a urine pregnancy test, serum hormone testing, and pelvic imaging. Additional testing may also be indicated based on the clinical presentation. Treatment depends on the underlying etiology and may include lifestyle interventions, hormone therapy or other medications, surgery, and mental health services. Therefore, all healthcare professionals should strive to enhance their competence when managing primary amenorrhoea and equip themselves with updated knowledge, skills, and strategies for timely diagnosis, effective interventions, and improved care coordination, leading to better patient outcomes.[9]

The basic requirements for normal menstrual function include four anatomically and functionally distinct structural components: the hypothalamus, anterior pituitary gland, ovary, and the genital outflow tract composed of the uterus/endometrium, cervix, and vagina. If any of these components are nonfunctional or abnormal, menstrual bleeding cannot occur. Various other rare etiologies are also possible, and because ovulation occurs before menstruation, pregnancy must always be considered as well. Furthermore, any etiology of secondary amenorrhoea may also present as primary amenorrhoea. Determining the underlying cause of amenorrhoea will assist in guiding management decisions.[10,11] Despite the numerous potential causes of primary amenorrhoea, the majority of cases are caused by gonadal dysfunction (43%), müllerian agenesis (10%-15%), and constitutional delay of growth and puberty (14%). Other relatively common etiologies, accounting for between 2% and 7% of cases, include functional hypothalamic amenorrhoea (FHA), transverse vaginal septum, polycystic ovary syndrome (PCOS), and hypopituitarism.[12]

Case 1

Our first index patient presented with a female genotype and phenotype, definitive scientific association with DES (Diethylstilboestrol) could not be made and presentation was not of a classical "T-shaped uterus". There are two forms of MRKH syndrome: a) typical form (Type I) is characterized by only congenital absence of uterus and upper vagina with normal appearing ovaries and fallopian tubes; and b) atypical form (Type II) includes Mullerian anomalies associated with non-gynaecological anomalies of urological, skeletal, vertebral or cardiac systems or a detailed explanation of uterine anomalies based on various systems of classifications, author encourages readers to refer to latest updates on ASRM classification (Table 2 and Figure 3).[13,14] For better understanding of imaging and MRI findings one can refer to radiology assistant for simplicity and explanation with imaging and PUMCH classification.[15,16] Our first index patient is therefore a case of "MRKH syndrome" and can be best classified by VCUAM classification as $V_{5b}C_{2b}U_{4b}A_{1b}M_0$ (Figure 1 and 2) (Table 3).[17].

Case 2

Our second index patient can be best described as "ovotesticular DSD" with female external genitalia with XXY karyotype or in colloquial parlance "chimera/hermaphrodite/intersex"; where the male sexual characteristics did not probably manifest due to dysgenetic testicular tissue in abdomen. Ovotesticular DSD occurs when an individual is born with both ovarian and testicular tissue present. Ovotestis is the most common gonad present and cases of ovotesticular DSD can be classified into three groups based upon the gonads present: (1) lateral with one testis and one ovary, (2) bilateral with two ovotestes, and (3) unilateral with an ovotestis and either an ovary or testis contralaterally. The development of the internal duct structures is determined based upon the local hormonal effects of the ipsilateral gonad and the external genitalia are ambiguous with hypospadias, cryptorchidism, and incomplete fusion of the labioscrotal folds. The ovarian portion of ovotestis may be normal whereas the testicular portion is typically dysgenetic. Rare ovulation and pregnancy has been reported for female 46XX ovotesticular DSDs but no clear male fertility documented.

The most common karyotype is 46XX, followed by 46XX/46XY mosaicism and 46XY. Most 46XX ovotesticular DSD patients are SRY negative and the genes responsible have not yet been identified. Although sex chromosome mosaicism arises from mitotic or meiotic errors, 46XX/46XY chimerism is usually a result of double fertilization (an X sperm and a Y sperm) or, less commonly, fusion of two normally fertilised ova. Thus, chimeric patients have two distinct cell populations. The least common form of ovotesticular DSD, 46XY, may result from a cryptic 46XX cell line or gonadal mosaicism with a mutated sex determination gene. While ovotesticular DSD is rare, accounting for 3-10% of DSD worldwide, it seems to be more common in South Africa representing approximately 50% of DSD in this population.[18]

Ovotesticular DSD is defined as the presence of ovarian tissue with follicles and testicular tissue with seminiferous tubules in the same individual. External genital appearance does not predict gonadal histology. The gonads can be an ovotetis, ovary, and/or testis. The most common histology is an ovotestis. An ovotestis is typically located on the right side and is often intra-abdominal. An ovotestis can appear ovoid or bilobed on gross appearance. In most ovotestes, ovarian and testicular tissue show distinct separation in an end-to-end arrangement. In this situation, the testicular zone typically shows poor differentiation of the tunica albuginea with atypical interstitial tissue. In some instances, oocytes are interspersed among seminiferous cords/tubules. The proportion of ovarian and testicular tissue differs among patients; uterine development also varies. The most common karyotype is 46,XX. Mosaic karyotypes, such as 46,XX/46,XY, 46,XX/47,XXY, 45,X/46,XY, 47,XYY/46, and XY/45,X have been

described. In some cases, the amount of Y chromosomal material in peripheral blood lymphocytes is limited, such as the *SRY* gene can be detected only by polymerase chain reaction (PCR) amplification. In other instances, *SRY* was apparently absent in peripheral blood lymphocytes and detected in testicular tissue. Several pregnancies have been reported among women with ovotesticular disorder.[19] A detailed discussion of ovotestis, sexual dysmorphism or disorders of sexual development (DSD) is beyond the scope of this case report and author suggests further reading to fully understand the variable presentation with lack of coherent classification of this rare disease.

Case 3

Our third index patient can be labelled as FXPOI and she is a case of "fragile-X premutation carrier" with XX genotype and a normal female phenotype. An individual with the fragile X premutation is a male or female who has between 55-200 CGG repeats in the fragile X (FMR1) gene. The full mutation is defined as over 200 CGG repeats. (A typical FMR1 gene has 6-54 CGG repeats.) Occasionally a female with a full mutation shows little or no effect of the full mutation and is sometimes referred to as a "full mutation carrier." However, most of the time, the term "carrier" is used and associated with those with a premutation. Some individuals have what is called an "intermediate" or "gray area" sized allele. These are alleles with 45-54 CGG repeats. They are not considered to be mutations and do not appear to be associated with any clinical or medical issues, developmental disabilities, or social/emotional difficulties. These alleles are identified as such because there is a small chance that they are mildly unstable and may expand to a premutation in future generations. There is no reported risk for an individual with an intermediate sized allele to have a child with a full mutation. Generally, we don't use the term "carrier" for those with an intermediate allele. The FMR1 gene is on the X chromosome, which is why fragile X syndrome is called an X-linked condition or disorder. Often in these conditions or disorders, only females are carriers and only males are affected. However, in fragile X, both males and females can be carriers or have the Fragile X premutation, and both can be affected by the condition.

About 20% of women with the fragile X premutation develop POI over their reproductive life span, compared with only 1% in the general population. Approximately 3% of women with the fragile X premutation will have menstrual cycle irregularities in their teens or 20s due to FXPOI. The 1% of women with the fragile X premutation will stop having periods prior to age 18, and about one-third-equivalent to 7% of women with the fragile X premutation-stop having periods at or before age 29. Not all women with the fragile X premutation experience FXPOI. One well-documented risk factor is the premutation repeat size: the highest risk for ovarian dysfunction is for women carrying premutation alleles in the 80-100 CGG repeat range, not the highest alleles of >100 repeats. Women with the full mutation do not experience FXPOI or increased twinning rates.[20,21]

CONCLUSION

Primary amenorrhoea is a challenging case for any ob-gyn practitioner. All of us must have encountered a variety of cases during our clinical practice, most of us missing out on diagnosis by often repeating a plethora of investigations as per "protocol" without detailed history taking or a nuanced examination. The article attempts to convey futility of repeated investigations by different fertility specialists based on protocol without proper history taking and a full examination, which leads to wastage of time, with delay in initiation of therapy/surgical intervention and has monetary ramifications for patients who then resort to unscientific treatments which seldom leads to answers or cure leading to hopelessness and ostracization of these women. Our aim should be to provide honest scientific and truthful closure, however unpalatable and avoid giving false hopes of treatment and unrealistic therapies for profits and business. These women deserve honest answers and a viable solution with extensive psychological support for gender dysphoria.

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