IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 25, Issue 2 Ser. 5 (February. 2026), PP 47-53
www.iosrjournals.org

When Menarche Does Not Begin- Clinical Patterns On Primary Amenorrhea

Dr. Sree Mounica L, Dr. Sneha Pawar, Dr. Devdatta Dabholkar
Department Of Obstetrics And Gynaecology, MGM Institute Of Health Sciences, Navi Mumbai, Maharashtra, India.
Department Of Obstetrics And Gynaecology, MGM Institute Of Health Sciences, Navi Mumbai, Maharashtra, India.
Department Of Obstetrics And Gynaecology, MGM Institute Of Health Sciences, Navi Mumbai, Maharashtra, India

Abstract
Introduction: *Primary amenorrhea is when the menarche does not occur by 16 years of age in the presence of normal secondary sexual characteristics, or by 14 years in their absence. It is an uncommon presentation in gynaecology with diverse aetiologies. This often indicates some underlying disorders of the hypothalamic–pituitary–ovarian axis or an abnormality of the reproductive tract. It is essential to identify these concerns early in life as they affect sexual maturity, fertility and psychosocial health.*
Methods: *We present three Primary amenorrhea cases who presented to the hospital A systematic evaluation comprising detailed history, physical examination with assessment of secondary sexual characteristics, hormonal investigations, pelvic imaging, and karyotyping wherever indicated was performed in all patients. Management became personalized depending on the underlying etiology.*
Results: *Assessment was done on three patients. Among the three patients with primary amenorrhea two patients were diagnosed as MRKH syndrome and one patient as Turners syndrome. All three patients received individualized treatment and showed good improvement on follow-up.*
Conclusion:
Primary amenorrhea has many different causes and requires a structured approach to diagnosis. A multidisciplinary, individualized management strategy is essential to optimize clinical outcomes and address the psychosocial impact associated with delayed menarche.

Date of Submission: 18-02-2026 Date of Acceptance: 28-02-2026

I. Introduction

An adolescent gynecology and reproductive medicine problem of major significance is primary amenorrhea. The absence of menarche (the first occurrence of menstruation) is defined as absence of menarche by age 16 in girls who have normal growth and development of secondary sexual characteristics, or by 14 in those without secondary sexual characteristics [1]. This condition conveys an interruption at hypothalamic-pituitary-ovarian axis one or more levels, responsiveness-or end-organ along outflow genital tract. The evaluation of primary amenorrhea should be systematic and stepped. The presence or absence of secondary sexual characteristics directs the initial assessment, which can assist in differentiating disorders of sexual development from endocrine dysfunction or structural abnormalities of the reproductive tract. An accurate diagnosis can be achieved through proper clinical examination and the relevant hormonal, pelvic imaging, and cytogenetic studies. Congenital Mullerian anomalies can cause primary amenorrhea in women that have normal secondary sexual characteristics [2]. MRKH syndrome is one among a group of disorders characterized by congenital absence of the uterus and upper vagina in those with normal female karyotype (46, XX) and normally working ovaries. The patients generally have normal external genitalia and normal puberty but experience primary amenorrhea. This must be identified as early as possible to provide the right counselling and more options. Chromosomal irregularities make up another etiological group. Characterized by gonadal dysgenesis, Turner syndrome is mostly due to complete or partial monosomy X. It results in estrogen deficiency and hypergonadotropic hypogonadism. Individuals afflicted with this condition frequently experience primary amenorrhea, short stature, and halted or absent development of sexual characteristics. Early detection allows commencement of hormone replacement therapy and checking for systemic anomalies [3]. This paper describes three cases of primary amenorrhea, including two with MRKH syndrome and one with Turner syndrome. They exhibited distinct clinical, hormonal, radiological, and genetic profiles. The series emphasizes organized diagnosis and individualized management for patients with primary amenorrhea [4].

II. Objectives

- To report the clinical manifestations of patients with primary amenorrhea.
- To highlight the importance of clinical examination, imaging, hormonal evaluation, and karyotyping for diagnosis.
- To understand the clinical, hormonal and genetic profile of patients with MRKH syndrome and Turner syndrome.
- To highlight the significance of early diagnosis and adequate counseling in the management

III. Methodology

The study was a descriptive case series conducted at a tertiary care centre in the Department of Obstetrics and Gynaecology. The study was conducted on three young females with primary amenorrhoea. All patients and guardians were informed consented prior to the study. In each case, detailed history included age, pattern of growth and development of secondary sexual characteristics and associated symptoms.

A general physical examination was conducted with particular focus on height, body proportions, and dysmorphic features. Where indicated, local genital examination was performed and per rectal examination was performed too. We conducted a baseline hormonal evaluation of all patients. The evaluation included serum follicle-stimulating hormone, luteinizing hormone, estradiol, thyroid-stimulating hormone, and serum prolactin levels. All cases underwent pelvic ultrasound, and MRI was performed when necessary to delineate Mullerian anomaly. All patients underwent analysis of chromosomes to detect their abnormalities.

Based on clinical findings and investigation results, a final diagnosis was established. Patients were managed according to the underlying etiology and were counseled regarding prognosis, fertility options, and the need for long-term follow-up.

IV. Case Series

Case 1:

A 16-year-old female presented with primary amenorrhea. There was no history of cyclic abdominal pain. General physical examination was unremarkable. On examination there was absent breast development and absent pubic and axillary hair. Local examination revealed normal external genitalia. Per rectal examination showed a short blind vaginal pouch with absence of cervix and uterus. Pelvic ultrasonography demonstrated an absent uterus and upper 2/3rd of the vagina with a thin streak of bilateral ovaries (Image 1). Renal imaging was normal. CT A+P showed non visualization of uterus and ovaries ?MRKH. Karyotype analysis revealed a 46,XX chromosomal pattern (Image 2). A hormonal examination showed raised FSH and LH (Table 1). Based on clinical, radiological, and cytogenetic findings, a diagnosis of Premature Ovarian failure with MRKH syndrome was made. The patient was advised HRT for bone protection and Vaginoplasty. The patient received counseling regarding the condition, sexual health, and future fertility options.

Image 1

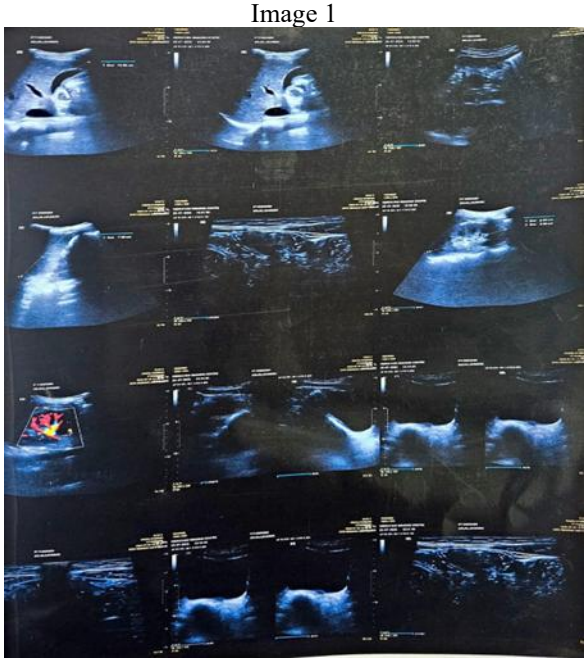


Image 2

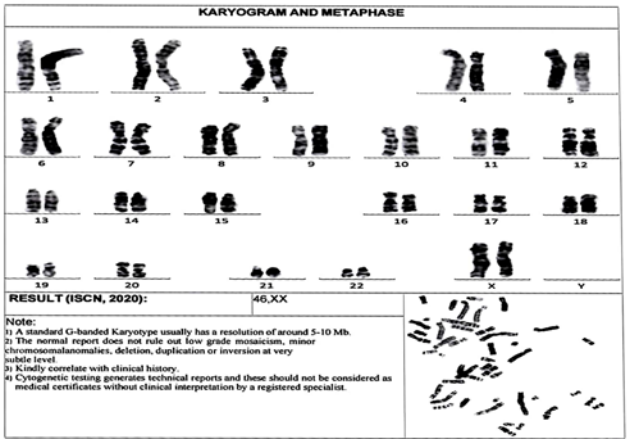


Table 1: Comparative Investigations of Three Patients with Primary Amenorrhea

Parameter	Case 1	Case 2	Case 3
Age (years)	16	20	17
Height	Normal	Normal	Short
Secondary sexual characteristics	Absent (Tanner I)	Normal (Tanner V)	Absent (Tanner I)
External genitalia	Normal	Normal	Normal
Vaginal length	Short blind pouch	blind ending vagina	Short blind pouch
Uterus (PR exam)	Not palpable	Not palpable	Small uterus
Ultrasound pelvis	Absent uterus, thin streak of bilateral ovaries	Aplastic uterus with hypoplastic vaginal canal with B/l streak ovaries	Diffusely atrophic uterus with non visualization of B/L ovaries ?Agenesis
CT A+P/ MRI	Non visualization of uterus and ovaries ?MRKH	Absent uterus and cervix with non visualization of vagina s/o MRKH	
Renal imaging	Normal	? left atrophic kidney	Normal
Estradiol (20-145)	16	98	2.0
Serum FSH Menopause: 21.5-131 Follicular: 1.98-11.6 Luteal:1.38-9.53	69.2	4.5	40.89
Serum LH Follicular: 2.58-12.1 Luteal: 0.85-15.5	26.6	3.97	34
AMH		2.6	
TSH (0.12-3.12)	1.277	2.591	5.28
Prolactin Non pregnant: <25	820.6	2.73	102.8
Karyotype	46XX	46XX	45X
Final diagnosis	Premature ovarian failure + MRKH	MRKH syndrome	Turner's syndrome

Case 2:

A 20 year old female presented with failure to attain menarche and dyspareunia. She had normal development of secondary sexual characteristics with no dysmorphic features. External genitalia were normal on inspection. On per speculum examination there was blind ending vagina. On per vaginal examination, the tip of the finger is admitted. Per rectal examination revealed absence of uterus. Pelvic ultrasound showed, A 1.5X0.8 cm sized short tubular hypoechoic structure is seen between bladder and rectum probably representing cervix with visualized endocervical canal. The vaginal canal appears hypoplastic measuring upto 1.5cm, however the uterine body could be visualized likely suggestive of aplastic uterus (Image 3). B/L Ovaries appeared small in size. On renal imaging, the right kidney is normal in size, shape and echotexture but the left kidney is not visualized in the left renal fossa suggesting ? atrophic/absent left kidney (Image 4). MRI pelvis showed absent uterus and cervix with non visualization of vagina, bilateral ovaries are visualized and appear normal. Findings are suggestive of MRKH syndrome. Hormonal profile was within normal limits. Karyotyping showed 46,XX (Image 5), confirming MRKH syndrome. The patient also underwent vaginal examination under anaesthesia which was suggestive of an extremely small blind pouch in the posterior fourchette, uterus not felt per rectal examination and transperineal ultrasound showed no evidence uterus. The patient and her family were counseled regarding the diagnosis and the patient was counselled regarding Vaginoplasty to create a functional vagina. For fertility the patient was counselled regarding embryo donation and surrogacy. Psychological support was offered, and long-term follow-up was advised.

Image 3

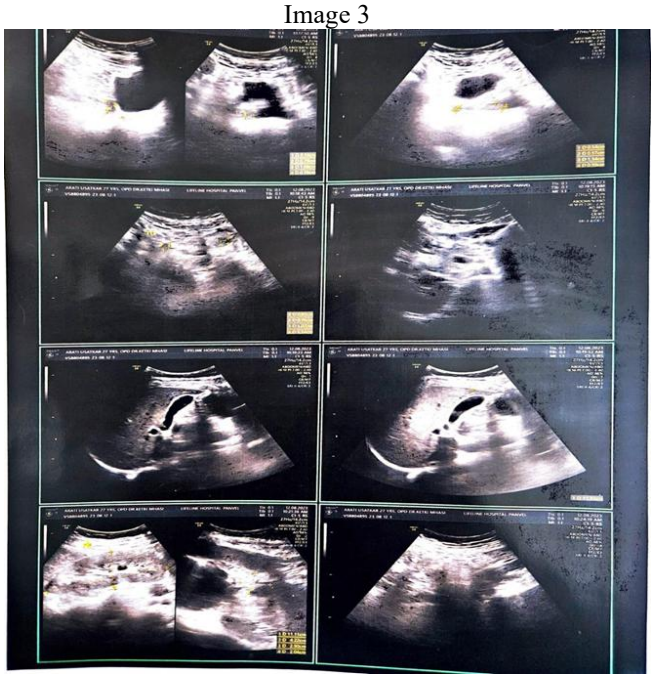


Image 4

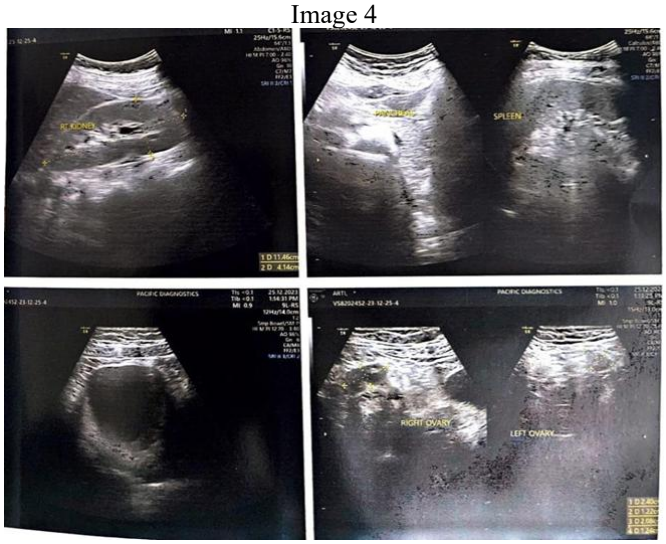
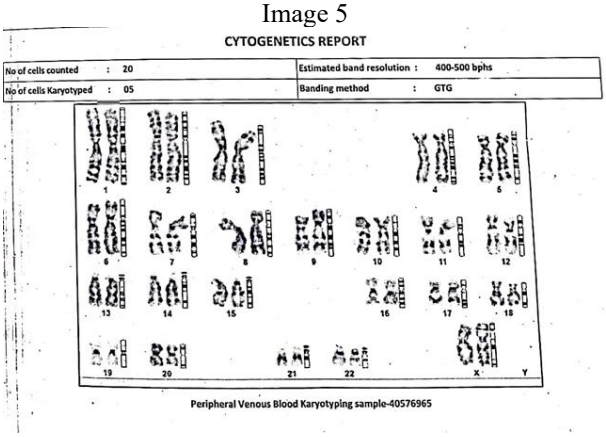


Image 5



Case 3:

A 17-year-old girl presented with primary amenorrhea and short stature. She had poor breast development and sparse pubic hair. Physical examination revealed short stature, webbed neck and widely spaced nipples (Image 6). On examination there were absent secondary sexual characteristics. A per rectal examination revealed a small uterus. Pelvis Ultrasonography showed the uterus is deeply atrophic and is seen as a linear hypoechoic area measuring approximately 3cm in length and 0.5cm in breadth and bilateral ovaries were not visualized (Image 7). Hormonal evaluation revealed that FSH and LH levels were high while the estradiol levels were low, indicating hypergonadotropic hypogonadism. The karyotype study illustrated monosomy X (45,X) (Image 8) indicating Turner syndrome. The patient started on estrogen replacement therapy and referred for multidisciplinary management including cardiology and endocrinology evaluation.

Image 6

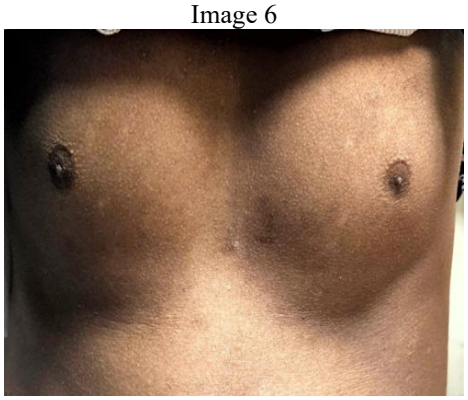


Image 7

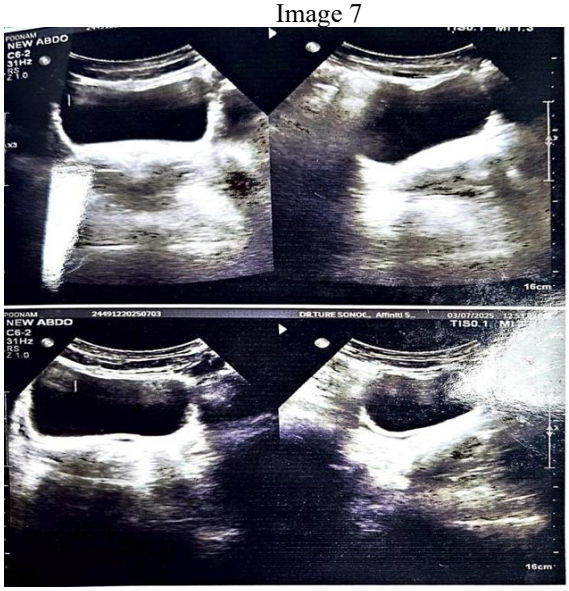
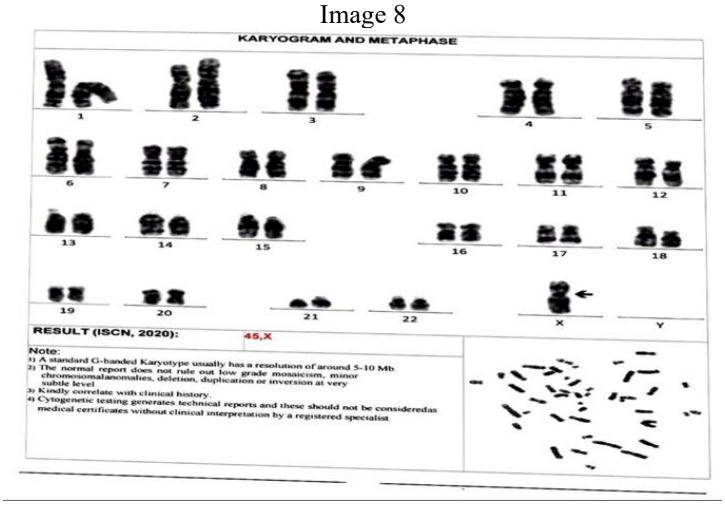


Image 8



V. Results

Three patients having primary amenorrhea were evaluated. Two patients experienced a missing uterus along with a blind-ending vagina. Their karyotyping exhibited chromosomal pattern of a female (46, XX) The findings were along the line of MRKH syndrome. The third patient showed short stature with poorly developed secondary sexual characters. Ultrasound scan showed a small lesser-developed uterus and streak gonads. Hormonal testing revealed high gonadotropins with low estrodial indicating hypergonadotropic hypogonadism. Karyotype study confirmed monosomy X (45,X) of Turner's syndrome. Table 1 depicts a comparative summary of clinical characteristics, imaging findings, hormonal profiles, and genetic results. All patients were diagnosed early, which allowed timely counseling and initiation of appropriate management.

VI. Discussion

Diagnosis of primary amenorrhea is done using a stepwise approach based on secondary sexual characters, genitalia morphology, hormonal evaluation and karyotyping. MRKH syndrome occurs in many cases who have normal secondary sexual characteristics and normal ovarian function. Instead, Turner syndrome commonly presents with gonadal dysgenesis and absent or incomplete puberty[5]. An early diagnosis is essential to avoid medical complications; start hormone therapy and psychosocial support.

Genetics and Hormones Evaluation

A complete genetic and hormonal evaluation will help establish the cause of primary amenorrhea and help in the management.

Hormone Assessment

In all study subjects, Serum FSH, LH, Estradiol, TSH and serum prolactin were done as baseline hormonal assessment. In Case 2 diagnosed with MRKH syndrome, serum FSH, LH and estradiol levels were within normal limits indicating normal ovarian function. Thyroid function tests and prolactin levels were normal, ruling out endocrine causes of amenorrhea. The patient's findings and normal secondary sexual characteristics were considered consistent with an outflow tract abnormality rather than hormonal dysfunction. In contrast, the patient with Turner syndrome (Case 3) showed significantly increased FSH and LH levels along with low serum estradiol, reflective of hypergonadotropic hypogonadism due to gonadal failure. These hormonal tests explained the pubertal delay and primary amenorrhoea and justified the initiation of hormone replacement therapy. The patient in case 1 with MRKH syndrome and premature ovarian failure had high levels of FSH and LH, and without secondary sexual charecters along with absent vagina and uterus.

Genetic Assessment

Karyotype analysis of all three patients was done to rule out chromosomal abnormalities. Both of the MRKH patients had a female karyotype, 46, XX. In fact, this is characteristic for MRKH syndrome. Thus, it can help distinguish this condition from other disorders like androgen insensitivity syndrome [6]. Imaging also showcased the presence of ovaries for confirmation. The third patient showed a karyotype of monosomy X (45, X), diagnosis of Turner syndrome. The discovery of this chromosomal anomaly warranted investigation of concurrent systemic anomalies and management by a long-term multidisciplinary approach.

Clinical Importance

In patients with hormonal and genetic characteristics that most likely differ, clinical examination should be correlated with biochemical and cytogenetic studies [7]. MRKH syndrome is basically a structural defect where the ovaries are fine. On the other hand, Turner syndrome is a chromosomal defect with endocrine deficiency that requires initiating hormones early and monitoring for life.

Management & Follow Up

Counseling for vaginal lengthening options, sexual health, and assisted reproductive technologies (ART) like surrogacy using their own oocytes was provided to MRKH patients. A patient with Turner syndrome was started on estrogen replacement therapy to induce secondary sexual characteristics and reduce the risk of osteoporosis. All patients were suggested routine follow-up and counseling.

VII. Conclusion

The primary amenorrhea case series describes various etiologies of primary amenorrhea occurring within the common practice setting. As response, it can be diagnosed with clinical examination, image and karyotyping. Identification of MRKH and Turner syndrome enables early treatment, proper advice, and customized long-term care.

References

[1]. Master-Hunter T, Heiman DL. Amenorrhea: Evaluation And Treatment. Am Fam Physician. 2006;73(8):1374–1382.
[2]. Reindollar RH. Primary Amenorrhea: Investigation, Management, And Prognosis. Fertil Steril. 2003;80(5):1264–1273.
[3]. Griffin JE, Edwards C, Madden JD, Harrod MJ, Wilson JD. Congenital Absence Of The Vagina: The Mayer–Rokitansky–Küster–Hauser Syndrome. Ann Intern Med. 1976;85(2):224–236.
[4]. Morcel K, Camborieux L, Guerrier D. Mayer–Rokitansky–Küster–Hauser (MRKH) Syndrome. Orphanet J Rare Dis. 2007;2:13.
[5]. Bondy CA. Turner Syndrome 2008. Horm Res. 2009;71(Suppl 1):52–56.
[6]. Gravholt CH, Andersen NH, Conway GS, Et Al. Clinical Practice Guidelines For The Care Of Girls And Women With Turner Syndrome. Eur J Endocrinol. 2017;177(3):G1–G70.
[7]. Klein DA, Poth MA. Amenorrhea: An Approach To Diagnosis And Management. Am Fam Physician. 2013;87(11):781–788.