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Original research article

A STUDY ON MULLERIAN ANOMALIES A CAUSE OF PRIMARY AMENORRHEA, AT TERTIARY CARE CENTRE

[1]Dr. Chitti Bala Bhavana Sai Kirthi, [2]Dr. Katari Narayana Madhavi, [3]Dr. Thota Susmitha,
[4]Dr. Potharaju Jayanthi, [5]Dr. Dontamsetti Anjana Devi

[1]Junior Resident, Department of Obstetrics and Gynecology, Guntur Medical College, Guntur,
Andhra Pradesh, India

[2]Associate Professor, Department of Obstetrics and Gynecology, Guntur Medical College, Guntur,
Andhra Pradesh, India

[3]Assistant Professor, Department of Obstetrics and Gynecology, Guntur Medical College, Guntur,
Andhra Pradesh, India

[4]Professor, Department of Obstetrics and Gynecology, Guntur Medical College, Guntur,
Andhra Pradesh, India

[5] Assistant Professor, Department of Obstetrics and Gynecology, Guntur Medical College, Guntur,
Andhra Pradesh, India

Corresponding Author:
D. Anjana Devi

Abstract

Background: Initiation of menstruation is important milestone in reproductive life of women. Amenorrhea means absence of menstruation. Primary amenorrhea means absence of menstruation by 14 years with no secondary sexual characteristics or by 16 years with presence of secondary sexual characteristics.

Primary amenorrhea occurs when there is mullerian anomalies, outflow tract obstruction, hormonal changes. True incidence of mullerian anomalies is unknown but believed between 0.1% to 3.8% and second most common cause of primary amenorrhea is mullerian anomalies.

Methods: This was an observational study conducted in tertiary centre, government general hospital, Guntur over a period of 14 months (Jan 2023 to Feb 2024). Ethics approval was obtained from institutional ethics committee.

Results: Out of total patients presenting with primary amenorrhea (n=12) have outflow tract obstruction and anomalies.

Conclusion: Mullerian anomalies and imperforate hymen are most prevalent causes of amenorrhea in our study.

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Keywords: MRKH syndrome, primary amenorrhea, septum, MRI, renal anomalies

Introduction

Primary amenorrhea is defined as failure of onset of menstruation by age of 16 years, regardless of development of secondary sexual characters or absence of menstruation by 14 years of age when there is no development of secondary sexual characters [1]. Uterus can be identified as early as by end of 3rd month. Uterus, fallopian tube, cervix and upper 2/3rd of vagina are derived from mullerian duct in absence of Y chromosome [2].

The incidence of primary amenorrhea is 0.3% [3]. Amenorrhea occurs if there is blockage of the outflow tract or non-functioning/absent uterus or outflow tract missing, this include transverse blockage, mullerian anomalies, and absence of functional endometrium.

Early diagnosis and specific management are essential for planning treatment and achievement of healthy sexual relationship and successful reproductive outcome.

True incidence of obstructive mullerian anomalies is unknown, but believed between 0.1-3.8% and second most common cause of primary amenorrhea is mullerian anomalies [4].

Causes of amenorrhea are multiple and can be due to problems at different level-it could be due to chromosomal abnormalities, altered HPO axis, streak gonads, endocrine disorders (thyroid, prolactin), uterine/vaginal anomalies.

Aim of the study is to know etiology, causes of amenorrhea, different varieties of mullerian anomalies causing primary amenorrhea.

Methods

This study was done at government general hospital, Guntur from January 2023 to February 2024 over a period of 14 months

Inclusion Criteria

- Women of reproductive age group presenting with primary amenorrhea.
- Normal hormonal levels.

Exclusion Criteria

- Pregnant women.
- Women with causes other than mullerian anomalies.

History of family for genetic anomalies taken. General examination focused on secondary sexual characters, per abdomen, height of patient, local hymen examination, per vaginal, per rectal done to determine cause of primary amenorrhea.

RBS, LH, FSH, PROLACTIN, TSH, USG done as part of routine protocol of primary amenorrhea. MRI was advised to all and diagnosis was done based on MRI, associated renal anomalies were confirmed by MRI.

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Results

Table 1: Age at Presentation

AGE	n (n=12)	Percentage
12-14 years	5	41.6
15-17 years	4	33.3
18-20 years	1	8.33
20-23 years	2	16.66

In table 1 most common age group are between 12 to 14 years, comprising about 41.6%, 15-17 years were 33.3% and 20-23 years were 16.66%.
38.4% were in 19-21 years age group, in Chandrayan P *et al*. study [6].

Table 2: Presenting Symptoms/Complaints

Presenting Symptom	n (n=12)	Percentage
Primary amenorrhea	5	41.6%
Primary amenorrhea with pain abdomen	6	50%
Primary amenorrhea with urinary retention	1	8.3%

In above table 2 all presented with primary amenorrhea but many also presented with associated symptoms, 50% presented with pain abdomen, 8.3% presented with urinary retention 31% presented with primary amenorrhea in chandrayan P *et al*. study [6].

Table 3: Per Vaginal Examination

Per vagina	n (n=12)	Percentage
Vagina as blind pouch	7	58.3
Imperforate hymen	3	25
Vaginal septum	2	16.6

In table 3, on per vaginal examination, significant information was obtained 58.3% had vagina as a blind pouch, 25% had imperforate hymen, 16.6% had vaginal septum (though rare) 15.3% presented with vagina as blind pouch on per vaginal examination, and 15.3% have transverse vaginal septum on per vaginal examination in Chandrayan P *et al*. study [6].

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Table 4: MRI Features

MRI features	n (n=12)	Percentage	Renal Anomalies	Percentage
Mullerian agenesis/hypoplastic uterus	5	41.6%	2	16.6%
Transverse septum in upper 1/3rd vagina	1	8.3%	1	8.3%
Hypoplastic cervix	1	8.3%		
Bicornuate uterus with septum upto cervix	2	16.6%		
Imperforate hymen	3	25%		

In table 4, MRI was performed to all patients, in our study 41.6% have mullerian agenesis/hypoplastic uterus i.e. patient have absent uterus, fallopian tube, cervix and upper 2/3rd vagina.

- 25% have imperforate hymen with normal uters, fallopian tube, cervix and vagina.
- 16.6% patient MRI reveal transverse septum with bicornuate uterus.
- 8.3% have hypoplastic cervix and 8.3% have transverse vaginal septum.
- 25% have associated renal anomalies like ectopic kidney, absent left kidney seen in patients with mullerian agenesis. Other conditions were not associated with renal anomalies.
- 61.5% have mullerian agenesis and 75% have associated renal anomalies in Chandrayan P *et al*. study [6].

Table 5: Comparison with Other Studies

Diagnosis	Our study (n=12) (%)	Parikh RM *et al*. (n=14) (%)	Tanmahansut *et al*. (n=295) (%)	Chandrayan P *et al*. (n=13) (%)
Mullerian agenesis	41.6	57.14	39.7	61.5
Transverse vaginal septum	24.9	7.14	0.3	31
Imperforate hymen	25	28.57	2.0	7.6
Hypoplastic cervix	8.3	0	0	0
AIS	0	7.14	5.1	0

In our study mullerian agenesis (41.6%) was the most common cause, this was compatabile with Parikh RM *et al*. study (57.14%) [5] and Chandrayan P *et al*. (61.5%) [6]. This is in contrast to American study by Reindollar *et al*. [5] gonadal dysgenesis was commonest (48.5%)-American study [7].

Discussion

The fusion of two mullerian ducts (paramesonephric ducts) forms the upper two thirds of the vagina, as well as the cervix and uterus. Embryonically, the female reproductive tract develops from the genital ducts and urogenital sinus. The mullerian ducts grow downwards and extend to enter the urogenital sinus, after which they get fused. Uterine

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cavitation occurs at site of fused mullerian ducts and the uterus continues to grow downward. Cavitation is complete and the lower segment and cervix, along with the upper part of vagina are formed [8].

In our study 25% have imperforate hymen, entirely urogenital origin, failure of canalization may lead to formation of mucocolps, this often remain unrecognized and present as amenorrhea with presence of secondary sexual characters, cyclical abdominal pain and retention of urine.

In our study 41.6% have mullerian agenesis/ hypoplastic uterus. MRKH syndrome occurs in 1in 5000-1 in 20000. It occurs due to defect in development of mullerian duct. It is often associated with other anomalies, most common being renal anomalies. It is the second most common cause of primary amenorrhea [9].

In our study 25% in total have transverse septum, it is a rare condition and incidence is 1 in 84,000 cases. Common site is upper and middle 1/3rd of vagina. The exact etiology of transverse vaginal septum is unknown but its incidence has been reported to be 2 in 100,000 female live births making it one of the rarest anomalies of the female genital tract [10].

Differential diagnosis of mullerian anomalies are ANDROGEN INSENSTIVITY SYNDROME and TURNER SYNDROME.

Conclusion

Mullerian agenesis or MRKH SYNDROME is most common in our study associated with normal secondary sexual characters and it is one of the common cause of primary amenorrhea. They are detected when patient present with primary amenorrhea or associated symptoms like cyclical abdominal pain, urinary retention. Gold standard to diagnose this anomalies is MRI.

Revealing about mullerian agenesis require multidisciplinary approach, it generates anxiety and psychological distress to patient and their family members. One way to treat is create neovagina for normal sexual activity and since they have normal ovaries, they can have children by surrogacy or encourage to adoption.

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