(International Print/Online Journal)

SJIF IMPACT FACTOR: 5.565
PUBMED-National Library of Medicine ID-101739732

ISSN (Print): 2209-2870
ISSN (Online): 2209-2862

International Journal of Medical Science and Current Research (IJMSCR)
Available online at: www.ijmscr.com
Volume 5, Issue 4 , Page No: 107-113
July-August 2022

IJMSCR

Primary Amenorrhea : A Prospective Study For Diagnosis And Management At A Tertiary Care Center

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Type of Publication: Original Research Paper
Conflicts of Interest: Nil

Abstract

Background: Primary amenorrhea is defined as the absence of menarche in females. The basic requirement for normal menstrual function include intact hypothalamus pituitary axis, working ovaries and a functional outflow tract. Abnormality of any component lead to absence of menstruation. Primary amenorrhea has a major impact on the physical, psychological and social life of the girl and her family. This study aims to evaluate the patient characteristics, diagnosis and management of primary amenorrhea.

Methods: A prospective study was conducted in the department of Obstetrics and Gynecology, SMS Medical College Jaipur from july 2019 to October 2021. Patients presented with complaint of primary amenorrhoea were included in the study. Detailed history, clinical examination and investigations done to diagnose underlying cause then further managed accordingly.

Result : A total of 30 patients of primary amenorrhea were included in our study. In our study, most common cause of primary amenorrhea was Mayer Rokitansky Küster Hauser syndrome (MRKH) accounting for 66.6% cases followed by imperforate hymen (10%). Other least common causes were Androgen insensitivity syndrome, turner syndrome, transverse vaginal septum and physiological delay. Out of 20 patients of MRKH, 14 patients undergone vaginoplasty.

Conclusion : Primary amenorrhea is a major health concern among adolescent girls. Systemic evaluation including history, examination and laboratory assessment help to reach the diagnosis. Multidisciplinary approach involving gynaecologist, geneticist and psychologist followed for individualizing the management and counselling.

Keywords: Amenorrhea, Androgen insensitivity syndrome, Turner syndrome, Vaginoplasty

Introduction

Amenorrhea is defined as the absence of menstruation in females of reproductive age. It is classified as primary and secondary amenorrhea. Primary amenorrhea is defined either as the absence of menses in 14-year-old girls without the development of secondary sexual characteristics or the absence of menses in 16-year-old girls with normal development of secondary sexual characteristics.[1] However, a girl with the obvious stigmata of Turner's syndrome or the absence of a vagina should be evaluated right from the time of her presentation rather than this being postponed until she has met the strict definition of primary amenorrhea. Secondary amenorrhea is defined as the cessation of previously regular menses for three cycles or for more than 6 months in previously irregular cycles.

Menstruation in a girl is often celebrated as advent of womanhood and some communities in India even

celebrate this by ceremoniously blessing the girl. The initiation of menses is a complex process involving an intact hypothalamic pituitary axis, working ovaries, and a functional outflow tract. Etiologies of amenorrhea can be categorized as: outflow tract abnormalities, primary ovarian insufficiency, hypothalamic or pituitary disorders, other endocrine gland disorders, sequelae of chronic disease, physiologic[3]

Incidence of primary amenorrhoea is 1-2%.[2] Although uncommon but if left ntreated, it would leave the patient with long-term health risks, including hypoestrogen-related problems, infertility, sexual dysfunction, feelings of defeminization, and innate problems of the underlying cause. Therefore, early diagnosis and appropriate management are necessary in order to prevent or minimize the problems. This study aims to evaluate the patient characteristics, diagnosis and management of primary amenorrhea in a tertiary care hospital from July 2019 to October 2021.

Material And Methodology

A prospective study was conducted in the Department of Obstetrics and Gynecology, SMS Medical College Jaipur from July 2019 to October 2021. The inclusion criteria were female of age >14 years with primary amenorrhea with the absence of secondary sexual characteristics and female with age > 16 years with primary amenorrhea with normal growth and presence of secondary sexual characteristics. The exclusion criteria were female <14 years and secondary amenorrhea.

Demographic characteristics like age, marital status, socioeconomic status were noted. Detailed history regarding breast development, Pubic and axillary hair appearance, cyclical pain abdomen, retention of urine , headache ,visual disturbance, galactorrhea, radiation exposure, sudden weight loss, anosmia, family history of primary amenorrhea, maternal age at menarche, any sexual contact ,general health, and lifestyle were taken.

Physical examination included measurement of height, weight, BMI, presence of secondary sexual characters, Breast tanner staging, Abdominal examination, examination of the external genitalia. Rectal examination for presence of pelvic organs. Stigmata of Turner's syndrome was explored, including short stature, webbed neck, widely spaced nipples, cubitus valgus, low hairline, high arched palate, multiple pigmented naevi, and short fourth metacarpals.

Laboratory investigations, which vary from patient to patient depending on the provisional diagnosis derived from the medical history and physical examination, were sent. Pregnancy test was done wherever necessary. All the patients were subjected to pelvic ultrasound examination and the reports were reviewed not in isolation but with respect to the clinical findings. Further investigations like karyotyping, hormonal assays, MRI pelvis were done in selected cases according to provisional diagnosis.

After final diagnosis patient and her attendants were counselled regarding their condition and their menstrual, coital and reproductive function.

All the cases of MRKH syndrome were counselled about the need of vaginoplasty and vaginoplasty was done in 14 cases out of 20. Two sisters, elder one was already diagnosed with AIS with history of bilateral gonadectomy and younger one presented with bilateral inguinal swelling and primary amenorrhea further diagnosed with AIS. Bilateral orchidectomy with vaginoplasty was done at same sitting for first case and vaginoplasty done for second case. Both were advised for estrogen replacement therapy.

Result

A total of 30 patients of primary amenorrhea were studied during the period of July 2019 to October 2021. Maximum patients presented in the age group of 15 to 19 years (70 %) as shown in table no.1.Only one patient (3.3 %) presented above 25 years. out of these, 24 patients were unmarried and six patients were married. Twenty two patients were from the lower socioeconomic class and seven from lower middle class and another one from upper middle class.

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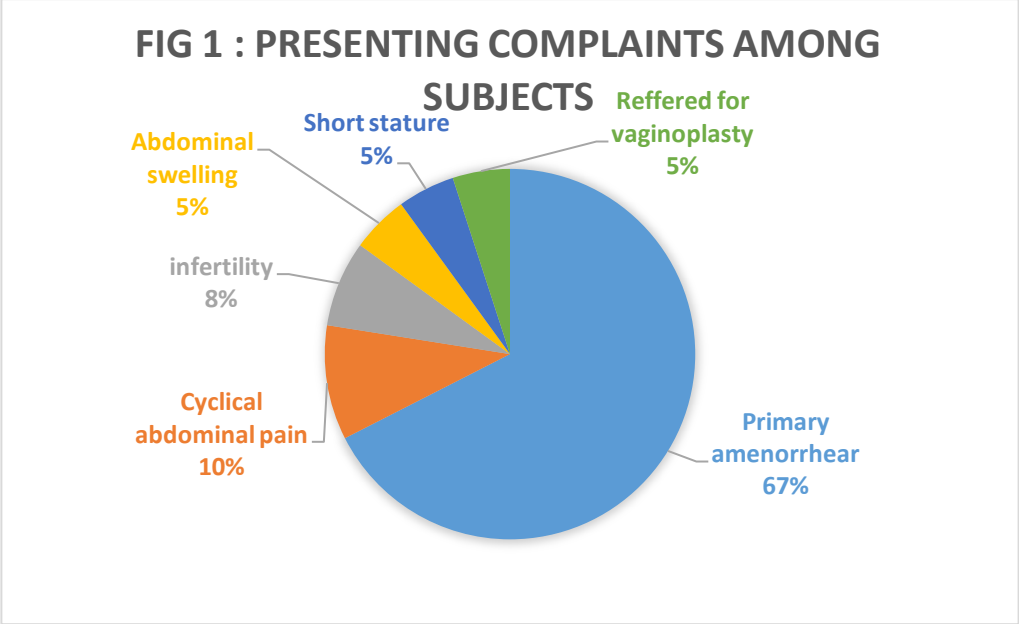
Table 1- Age at presentation

AGE	SUBJECT	PERCENTAGE
15-19	21	70
20-25	8.00	26.6
>25	1	3.3

Table 2 - Marital status at presentation

Marital status	Subjects	Percentage
Unmarried	24	80%
Married	6	20%

The most common presenting complaints among all subjects was primary amenorrhea (67%) followed by cyclical abdominal pain (10 %) as shown in fig 1 .



Etiology of Primary Amenorrhea

In our study twenty patients were diagnosed with MRKH accounting for 66.6% (n=20) followed by imperforate hymen (10%). Two cases of AIS were diagnosed and one case was of transverse vaginal septum. Two cases of constitutional delay and turner syndrome recorded during study.

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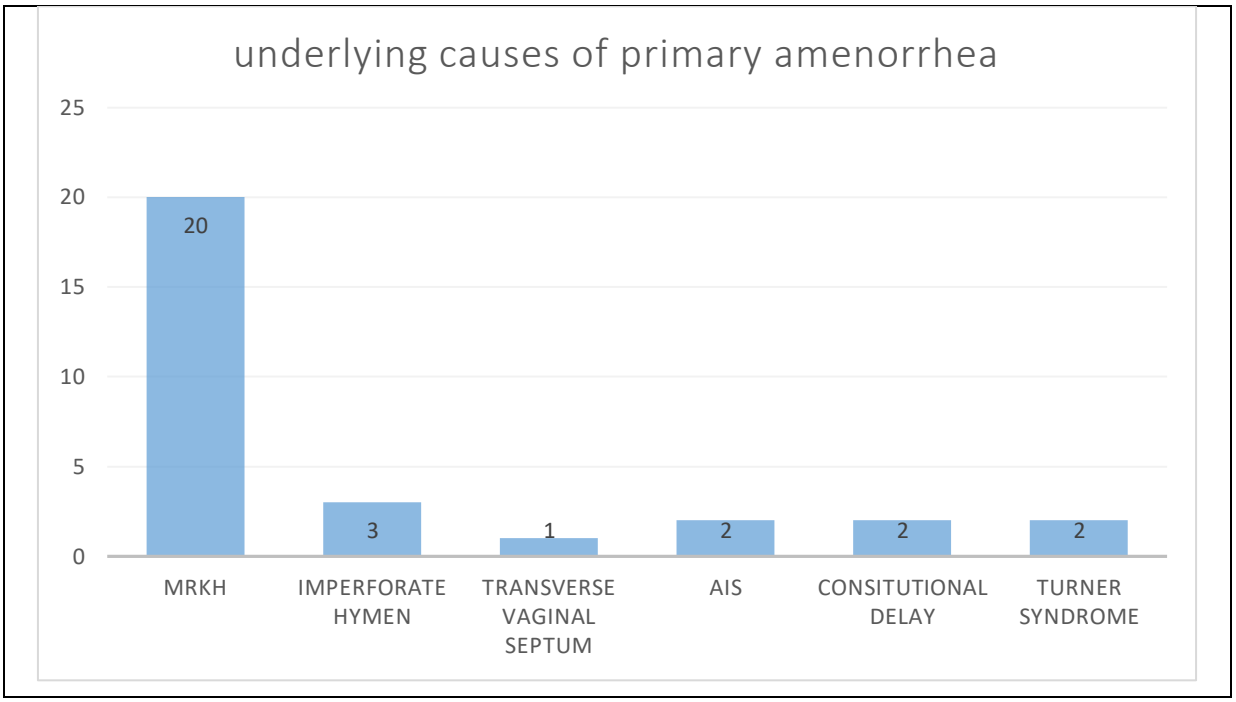

Fig 1 Vaginal membrane with bluish hue suggestive of imperforate hymen

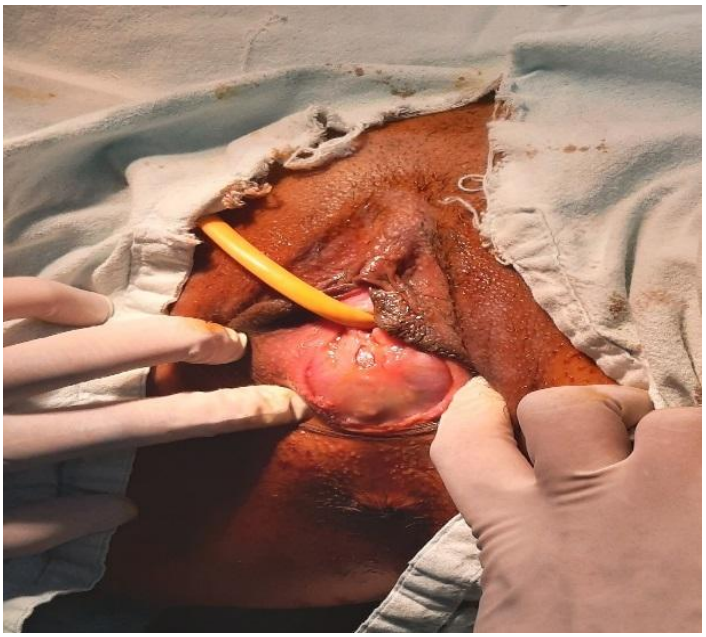

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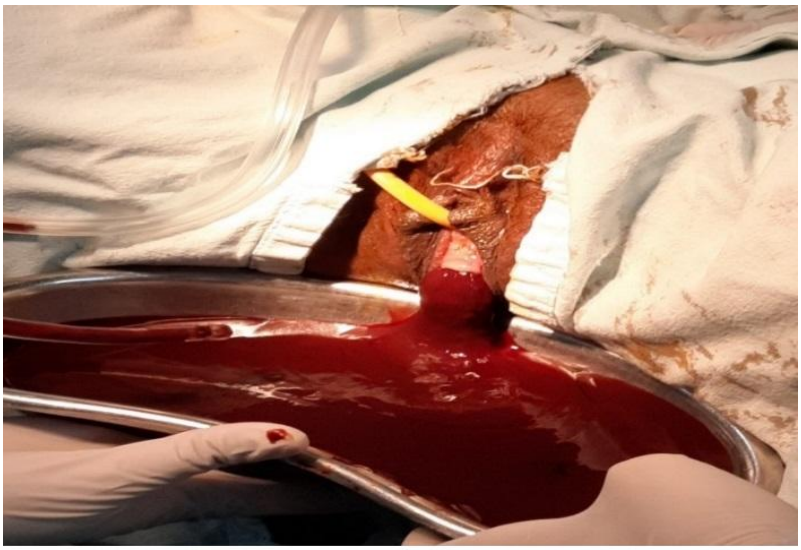
Fig 2 Drainage of hematometra secondary to imperforate hymen



Androgen insensitivity syndrome

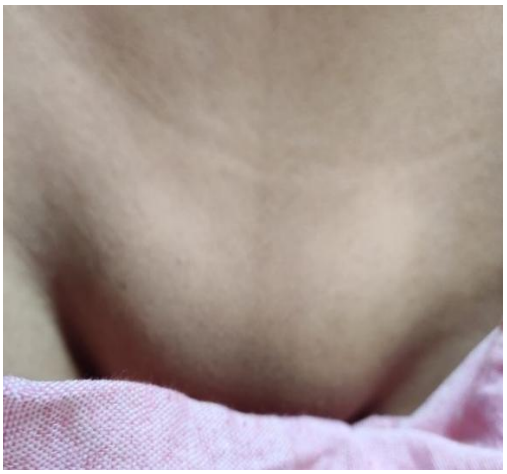


Fig 3 bilateral inguinal swelling

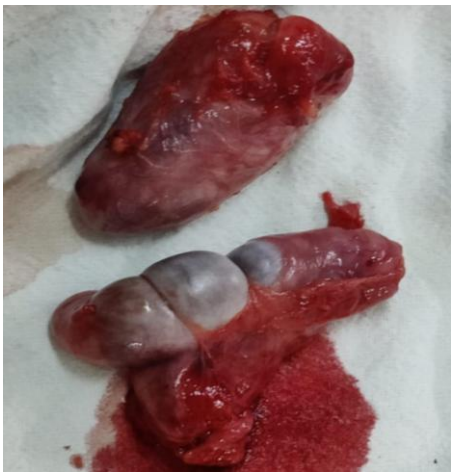
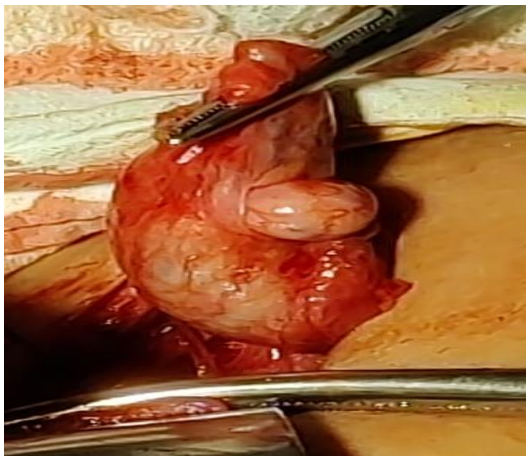


Fig 4: sample retrieved after orchidectomy

Figure 5: intraop picture of orchidectomy



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Discussion

Primary amenorrhea is a symptom of many potential causes, including developmental anomaly of the genital organs, failure of the ovaries to receive or maintain oocytes, and delayed pubertal development. Absence of menses in a pubertal girl create a lot of anxiety among parents. It requires sympathetic handling and multidisciplinary approach. Detailed history and physical examination help to make suspected diagnosis.

In our study, maximum patients presented in the age group of 15 to 19 years (70%). Similar results correlated with two studies conducted by Anita et al and Pavanaganga A et al.[4,5] Compatible with two Indian studies, Müllerian agenesis was the most common cause in the present study.[6,7] However, this was in contrast to the study by Reindollar *et al.*, which showed that the most common cause was gonadal dysgenesis (48.5%).[8]

In our study 20 (66.66%) patients were diagnosed with MRKH. This syndrome is defined as a failure of any of the structures along the mullerian tract, which includes the formation of the fallopian tubes, uterus, cervix, and proximal two-thirds of the vagina. Patients with MRKH will typically present late in adolescence with their only complaint as amenorrhea. Secondary sexual characteristics and external genitalia are normal on examination. Imaging reveal absence or ramant of uterus and mullerian duct derivatives. In our study 12 patients had absent uterus, eight patients had hypoplastic uterus. Two patients had associated renal anomalies. The treatment for these patients is multidisciplinary. Vaginal agenesis can be treated with serial vaginal dilation or surgery .Vaginoplasty is preferred choice because more girls are ready to get themselves surgically treated. Most important aspect is to do the procedure of vaginoplasty when the patient is mentally prepared to do it and to keep the patient motivated to continue doing postoperative vaginal dilatation. In our study ,out of 20 patients of MRKH , 14 patients undergone vaginoplasty, four patients were not mentally prepared to get operated, two patients were unfit for surgery. Vaginoplasty was performed using amnion graft in 10 cases and using skin split graft in 4 cases.

Three patients of imperforate hymen were diagnosed in our study. These patients presented cyclical abdominal pain, primary amenorrhea and normal secondary sexual characteristics. The bulging vaginal membrane with bluish hue noted on perineal examination was suggestive of imperforate hymen. Imaging reveals presence of all mullerian derivatives with normal ovaries. Massive hematometra, hematocolpos, noted in one case. All three patients of imperforate hymen underwent virginity preserving hymenotomy with drainage of menstrual blood. Transverse vaginal septum managed with septal resection with placement of vaginal mould.

Two cases (6.66%) of AIS were recorded during study period. Two sisters presented to gynecology OPD, first one was 19 years old unmarried female presented with bilateral inguinal swelling and primary amenorrhea further diagnosed with complete AIS. Second case was, 21 years old presented with primary amenorrhea and history of orchidectomy 3 years back. Karyotyping of both sisters revealed 46 XY. After evaluation, the patient along with parents were explained about the condition. Both patients were raised as girl-child since birth and wanted to live rest of life as woman. For them the realization that they are genetically male and have only partial female characteristic and would not be able to bear child was stressing factor. Bilateral orchidectomy with vaginoplasty was done at same sitting for first case and vaginoplasty done for second case. Both were advised for estrogen replacement therapy.

Turner syndrome was diagnosed in two cases (6.66%) in our study. Both patients presented with short stature, primary amenorrhea and absent secondary sexual characteristics. Hormonal assay showed increased level of FSH. USG revealed streak ovaries. Renal ultrasound, an echocardiogram and ophthalmologic examinations performed .They were also monitored for hypertension, diabetes, and thyroid dysfunction. No other anomaly detected in both cases. Karyotyping done which showed 45XO pattern. Patients were advised for combined hormone therapy with oestrogen for 21 days (conjugated equine oestrogen 0.625 mg daily) and progesterone (medroxyprogesterone acetate 10 mg daily) in last 12 days of the month. Calcium and vitamin also prescribed to prevent bone loss. Counselling was done regarding their reproductive life.

Conclusion

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Attainment of menarche in a girl brings in a lot of confidence and feminism in a girl. Primary amenorrhea accounts for significant amount of psychological trauma Primary amenorrhea poses a diagnostic dilemma. These patients present with specific signs and symptoms which point towards the underlying etiology. The presence of a normal reproductive tract and examination is followed by step by step investigations protocol lead to early diagnoses and management of underlying cause. Empathy is an important aspect in management of the same.

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