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Primary Amenorrhea Due to Developmental Defects in Adolescent Girls in Faisalabad

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ABSTRACT

Understanding the frequency of developmental defects in adolescents presenting with primary amenorrhea is crucial for timely diagnosis and intervention, in order to preserve reproductive potential, and address psychosocial impacts. **Objective:** To determine the frequency of developmental defects in adolescent girls presenting with primary amenorrhea. **Methods:** This cross-sectional study was conducted from March 2021 to September 2021 after taking approval from ethical review committee of Faisalabad Medical University. 205 girls having primary amenorrhea were recruited from Obstetrics and Gynecology Department, Allied Hospital, Faisalabad. Non-probability consecutive sampling technique was used. After taking history and physical examination, patients were sent to the hospital Radiology department for transabdominal ultrasound and reported by senior Radiologist. Developmental defects were assessed and noted. Data analysis was done using SPSS version 26.0, quantitative data were presented as mean and standard deviation, while qualitative as frequency and percentages. **Results:** Frequency of developmental defects in adolescent girls presenting with primary amenorrhea was found in 23 (11.22%) participants; with imperforate hymen in 26.09%, Mayer-Rokitansky-Küster-Hauser syndrome in 21.74%, transverse vaginal septum in 8.70% and absent vaginal functioning uterus in 43.48%. **Conclusions:** This study concluded that the frequency of developmental defects in adolescent girls presenting with primary amenorrhea is quite high. It was recommended that public awareness programs should be arranged on national levels for women about primary amenorrhea and their causes through educational training and guidance to take proper and timely treatment in order to reduce the morbidity of these particular patients.

INTRODUCTION

Approximately 2-5% of adolescent girls presents with primary amenorrhea [1]. Its prevalence is rising, and this increase can be attributed to greater access to healthcare services, declining trend in child marriages, and enhanced awareness driven by social media [2]. Primary amenorrhea is a symptom indicative of an underlying condition affecting any part of the hypothalamic-pituitary-ovarian-uterine axis. The causes can be categorized as functional or anatomical defects in the hypothalamus, pituitary gland, uterus, or ovaries, as well as genetic abnormalities at the chromosomal or gene level [3]. Primary amenorrhea often involves developmental anomalies, or imperforate hymen [4]. Among developmental anomalies, Müllerian agenesis and gonadal dysgenesis being commonest [5]. While imperforate hymen may be identified during childhood, it can also remain undiagnosed and present in adolescence with cyclic abdominal pain and primary amenorrhea [6]. Another category of outflow tract anomalies involves absence of Müllerian structures, including Mayer-Rokitansky-Küster-Hauser syndrome (MRKH) and

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Androgen Insensitivity Syndrome (AIS). MRKH, commonly associated anomalies include skeletal, renal, and auditory defects [7]. Diagnosis is typically made using ultrasound or MRI [8]. AIS, arises from androgen resistance in genetic males with functional testes. Both MRKH and AIS share overlapping clinical features, but they are distinguished by karyotype analysis [9]. Primary amenorrhea is a challenging problem in developing countries like Pakistan due to society pressure and unknown fear. It affects physical, mental, psychological and social life of the patients and family and hence delayed diagnosis [10]. There is a misconception that the hormonal defects are the main cause of primary amenorrhea and treatment is given according to this concept.

So, this study was conducted to determine the frequency of developmental defects in adolescent girls presenting with primary amenorrhea in adolescent girls. Proper diagnosis can lead to appropriate treatment of the patients suffered from primary amenorrhea.

METHODS

This cross-sectional study was conducted at Obstetrics and Gynecology Department, Allied Hospital, Faisalabad, from 30th March 2021 to 29th September 2021. After obtaining approval from Faisalabad Medical University ethical review board (F.No.48-ERC/2020-21/PHRC/FMU/56), 205 eligible patients meeting the selection criteria were enrolled in the study. Informed consent was obtained from all participants. A sample size of 205 was calculated using the WHO sample size calculator with confidence level of 95%, an anticipated proportion 7%, and absolute precision 3.5%. A non-probability consecutive sampling technique was used [11]. Females aged 12–18 years, presenting with primary amenorrhea were included. Primary amenorrhea was defined as menstrual onset failure by the age of 14 in individuals without secondary sexual characteristics or absence of menstruation by the age of 16 despite normal growth and the development of secondary sexual characteristics. Females with hypopituitarism, weight loss, anorexia nervosa and isolated GnRH deficiency, constitutional delay of puberty, or chronic systemic disease /acute illness were excluded. A detailed history was taken, and physical examinations were conducted on all participants. Patients were referred to the hospital's Radiology Department for transabdominal ultrasound, which was interpreted by senior radiologist with minimum experience of 4 years in relevant filed. Developmental defects were assessed and noted including Imperforate hymen, MRKH syndrome, Transverse vaginal septum and Gonadal dysgenesis. Gonadal dysgenesis was labelled if; Ultrasound showed "streak" of fibrous tissue seen in the expected location of the ovaries and may contain no or very few ovarian follicles [12]. Transverse vaginal septum appears on ultrasound as a shortened, blind vaginal pouch with positive transillumination, septum can occur at any point along the vaginal cavity, although the vulva typically appears normal if the septum is located in the mid or upper vagina [13]. Imperforate hymen was labelled if; Presence of bluish bulging membrane at the entrance of vagina that allows positive transillumination at introitus on examination and confirmed on ultrasound as distended fluid-filled vagina and uterus with internal echoes [13]. Ultrasound findings of absent uterus and upper two-thirds of vagina, accompanied by normal ovaries and fallopian tubes, suggest Müllerian agenesis [14]. The study data were entered and analyzed by SPSS version 26.0. Quantitative variables, were presented as mean and standard deviation, while qualitative variables, as frequencies and percentages. Data were stratified for age, BMI and marital status, post stratification chi square was applied, p value ≤ 0.05 was taken as significant.

RESULTS

Table 1 shows demographic characteristics of study population. Mean age of 205 study participants noted was of 15.39 ± 1.83 years, among them majority of the patients 110 (53.66%) were between 16 to 18 years of age. Mean BMI noted was 27.31 ± 2.90 kg/m2. 188 (91.71%) participants found to be unmarried while only 17(8.29%) were married.

Table 1: Demographic Characteristics of Study Population(n=205)

Variables	Frequency (%)/Mean ± SD
Age	
12-15 (Years)	95 (46.34)
16-18 (Years)	110 (53.66)
Mean Age (Years)	15.39 ± 1.83
BMI (Kg/m^2)	27.31 ± 2.90
Marital Status	
Unmarried	188 (91.71)
Married	17 (8.29)

In current study, developmental defects in adolescent girls presenting with primary amenorrhea was found in 23 (11.22%) as shown in figure 1.

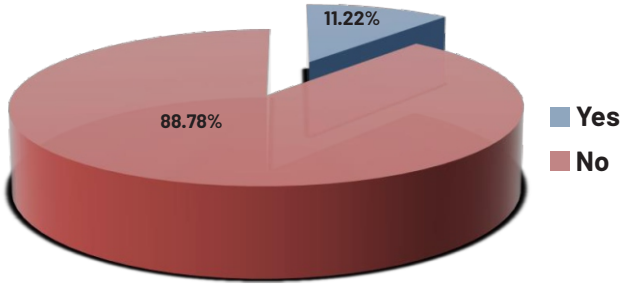

Figure 1: Frequency of Developmental Defects in Adolescent Girls Presenting with Primary Amenorrhea(n=205)

Among 23 patients found to have developmental defect, most common defect found was absent vaginal functioning uterus 43.48%, followed by imperforate hymen 26.09%,

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MRKH syndrome 21.74%, and transverse vaginal septum 8.70% as shown in table 2.

Table 2: Frequency Distribution Developmental Defect Etiology (n=23)

Types of Defects	Frequency (%)
Imperforate Hymen	06 (26.09%)
MRKH Syndrome	05 (21.74%)
Transverse Vaginal Septum	02 (8.70%)
Absent Vaginal Functioning Uterus	10 (43.48%)

Stratification of developmental defects with respect to age groups, BMI, and marital status is shown in Table 3 respectively, p-value >0.05 found to be statistically insignificant. In the age group of 12-15 years, 7.37% had developmental defects, compared to 14.55% in the 16-18 years' group, with a p-value of 0.105, suggesting a potential increase in prevalence with age despite the lack of statistical significance. For BMI, 13.86% of individuals with a BMI ≤27 kg/m^2 had developmental defects, while 8.65% of those with BMI >27 kg/m^2 did, with a p-value of 0.228 indicating no significant difference but suggesting a slight trend toward fewer defects in higher BMI individuals. Marital status showed that 12.23% of unmarried individuals had developmental defects, while none of the married individuals did, with a p-value of 0.126, suggesting no statistical association, though the absence of defects in married individuals might reflect other socio-economic factors.

Table 3: Data Stratification

Variables		Developmental Defects		p-value	CI
		Yes Frequency (%)	No Frequency (%)		
Age	12-15 (Years)	07 (7.37%)	88 (92.63%)	0.105	(-1.2% - 15.56%)
	16-18 (Years)	16 (14.55%)	94 (85.45%)		
BMI	≤27 (Kg/m^2)	14 (13.86%)	87 (86.14%)	0.228	(-3.45%- 13.87%)
	>27 (Kg/m^2)	09 (8.65%)	96 (91.35%)		
Marital Status	Unmarried	23 (12.23%)	165 (87.77%)	0.126	(7.84% - 16.62%)
	Married	00 (0.0%)	17 (100.0%)		

DISCUSSION

I have conducted this study to determine the frequency of developmental defects in adolescent girls presenting with primary amenorrhea. In current study, frequency of developmental defects in adolescent girls presenting with primary amenorrhea was found in 11.22% with imperforate hymen in 26.09%, MRKH syndrome in 21.74%, transverse vaginal septum in 8.70% and absent vaginal functioning uterus in 43.48%. In contrast to current results, study conducted by Kim *et al,* on 1060 females with primary ammenorrhea found higher frequency (30.96%) of outflow tract abnormality; among them Müllerian agenesis was most common cause (26.17%), followed by gonadal dysgenesis (22.4%), imperforate hymen (2.57%) and transverse vaginal septum (0.47%) [11]. This is further supported by another study conducted in Pakistan by Bibi *et al*, anatomical defect was noted in 60% females presenting with primary amenorrhea, and found Müllerian agenesis as most frequent cause (46%), followed by transverse vaginal septum and imperforate hymen (7% each)[15]. However, study conducted by Fowler *el al.*, found anatomical defect in only 1.5% participants [16]. In study conducted by Javed *et al.*, cases, most common anatomical defect found was gonadal dysgenesis (24%) followed by Mayer Rokistansky Kuster Hauser (21.9%), Imperforate hymen (8.5%), and vaginal septum (6.1%)[17]. These variances were found in results because factors including racial, genetic, and environmental seem to play a role in pathophysiology of primary amenorrhea [18]. The primary goals of treatment for young women with these anomalies are to alleviate obstructive symptoms, restore normal menstrual flow, and ensure sexual function while preserving reproductive potential [19]. It was proposed earlier that if regular menstruation has not commenced within two years after the onset of otherwise normal puberty, it is essential to rule out congenital absence of the uterus or vagina, provided this has not already been identified clinically[20]. In cases where puberty is atypical, or if abnormal gonadal tissue is identified before puberty, comprehensive investigations should be conducted promptly [21]. The clinical implications of these findings are significant for improving early detection, diagnosis, and treatment of primary amenorrhea, particularly among populations at higher risk for developmental defects. Health professionals should consider incorporating screening for developmental defects into routine evaluations for primary amenorrhea. Moreover, these findings highlight the need for targeted educational campaigns to raise awareness about primary amenorrhea, its causes, and available treatments. Furthermore, incorporating this data into treatment guidelines can help create a more personalized approach to care, ultimately enhancing the quality of life and reproductive health of affected girls. This study has certain limitations. It focused solely on structural anomalies and did not explore other potential etiologies of primary amenorrhea, such as endocrine abnormalities, genetic mutations, or environmental influences. Hormonal assessments and advanced genetic testing were not included, which may have provided a more comprehensive understanding of the condition. Additionally, the study relied on transabdominal ultrasound for diagnosis, which, while effective, may not capture subtle abnormalities detectable by more advanced imaging modalities like MRI.

CONCLUSIONS

This study concluded that the frequency of developmental defects in girls with primary amenorrhea is quite high. It

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was recommended that public awareness programs should be arranged on regional and national levels for educating women about primary amenorrhea and their causes through educational training and guidance to take proper and timely treatment in order to reduce the morbidity of these particular patients.

Authors Contribution

Conceptualization: SS
Methodology: SS, SH, NK, SNZ
Formal analysis: HR, SK
Writing, review and editing: HR, BZ, SNZ

All authors have read and agreed to the published version of the manuscript

Conflicts of Interest

All the authors declare no conflict of interest.

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