

# Case Series of Primary Amenorrhea at Nmch, Patna

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**Abstract:** Primary Amenorrhea Is Defined As The Absence Of Menses Till 13 Years Of Age In The Absence Of Secondary Sexual Characters , Or, Absence Of Menses Till 15 Years Of Age Regardless Of Secondary Sexual Characters. Its Incidence Is 0.1-2% In Reproductive Age Group Of Women .Identification Of Primary Amenorrhea Should Always Prompt A Thorough Evaluation To Identify The Cause And Provide Treatment Modalities Accordingly. This Study Provides Us With Knowledge Of Incidence And Findings Of The 10 Cases Of Primary Amenorrhea Presenting To Out Patient Department Of Obstetrics And AAAGynecology In Nmch ( Nalanda Medical College And Hospital ), Patna , Bihar.

**Keywords:** Primary Amenorrhea, Turners Syndrome, Cryptomenorrhea, Meyer Rokitansky Syndrome

## I. INTRODUCTION

Absence of menses by 13 years of age in the absence of growth or development of secondary sexual characteristics *or* Absence of menses by 15 years of age regardless of the presence of normal growth and development including secondary sexual characteristics • *novak. Amenorrhoea. Clinical Gynecologic Endocrinology And Infertility. 9th ed. Philadelphia: Wolter Kluwer (Replica Press); 2015:435-93.[1]*

II. AETIOLOGICAL	III. ENDOCRINOLOGICAL CLASSIFICATION
CLASSIFICATION OF PRIMARY AMENORRHEA	1. UTERUS ABSENT + BREAST PRESENT
	• MULLERIAN AGENESIS
	• ANDROGEN INSENSITIVITY SYNDROME
	• 5 ALPHA REDUCTASE DEFICIENCY ABSENT
COMPARTMENT 1: DISORDER OF OUTFLOW TRACT	• 2. UTERUS PRESENT + BREAST PRESENT
	• IMPERFORATE HYMEN
	• CERVICAL AGENESIS
	• TRANSVERSE VAGINAL SEPTUM
COMPARTMENT 2: DISORDER OF OVARY	• RESISTANT OVARY SYNDROME
	• 3. UTERUS PRESENCE + BREAST ABSENT (POOR)
	• TURNERS SYNDROME
	• MIXED GONADAL DYSGENESIS
COMPARTMENT 3: DISORDER OF ANTERIOR PITUITARY	• 17 ALPHA HYDROXYLASE AND AROMATASE ENZYME DEFICIENCIES
	• 4. UTERUS + ABSENT BREAST ABSENT AGONADISM
COMPARTMENT IV: DISORDERS OF THE HYPOTHALAMUS	

**IV. MATERIALS AND METHOD**

- TYPE OF STUDY : PROSPECTIVE
- DURATION OF STUDY : 1 YEAR (JUNE 2021 TO JUNE 2022)
- STUDY SIZE : 10
- THE STUDY WAS PERFORMED USING 10 PATIENTS WITH COMPLETE MEDICAL BACKGROUND WITH PRIMARY AMENORRHEA , VISITING OUTPATIENT DEPARTMENT OF OBSTETRICS AND GYNECOLOGY DEPARTMENT OF NMCH , PATNA, BETWEEN JUNE 2021 TO JUNE 2022.
- ALL PATIENTS UNDERWENT URINE PREGNANCY TEST BEFORE INCLUSION IN THIS STUDY.
- WITH FULL INFORMED CONSENT, A PROSPECTIVE ANALYSIS OF DATA WAS DONE ON BASIS OF DETAILED HISTORY TAKING, PHYSICAL EXAMINATION, PELVIC ULTRASONOGRAPHY , MRI , HORMONAL EVALUATION AND KARYOTYPING.

**TABLE 1 -THIS TABLE IS SHOWING DIAGNOSTIC WORKUP OF THE 10 CASES**

NAM E OF PATI ENT	A G E	SECON DARY SEXUA L CHAR ACT ERISTI CS	PER ABDO MEN	PELV IC EXA MIN A TION	PER VAGI NAL EXA MINA TION	PER REC TAL EXA MIN A TION	USG FIND ING	KARY OTY PING	FSH / LH LEV ELS	FINAL DIAGNO SIS
1. JULIE	14 Y R S	WELL DEVEL OPED	SOFT, UTER US	BLUIS H TENSE BULGI NG MEMBR ANE	NOT DONE	SMALL BULDGE FELT	UTE RUS AN D OV ARI ES PRE SEN T	46+XX	NO RM AL LEV EL	CRYPTOMENO RRHEA
2.PRIYA NKA	12 Y R S	WELL DEVEL OPED	SOFT	BLUIS H MEMBR ANE	NOT DONE	BULDGE FELT	UTE RUS AN D OV ARI ES PRE SEN T	46+XX	NO RM AL LEV EL	CRYPTOM ENORRHE A
3.SALO NI	12 Y R S	WELL DEVEL OPED	SOFT	BLUIS H MEMBR ANE	NOT DONE	NO FINDING	UTE RUS AN D OV ARI ES PRE SEN T	46+XX	NO RM AL LEV EL	CRYPTOMENO RRHEA
4.NISHA	15 Y R S	WELL DEVEL OPED	SOFT	NO SIGNIFI CANT FINDING	VAGI NA SHOR T	SMA LL NOD ULE FELT	UTERU S HYPOP LAST IC OVARI	46 XX	NO RM AL LEV EL	MEYER ROKITANS KY KUSTER

							ES NORM AL			HAUSER SYNDROME
5.PUJA	17 YRS	<u>WELL DEVELOPED</u>	SOF T	NO SIGNIFI CANT FINDING	<u>VAGINA SHORT</u> AND BLIND	NO SIGNIFI CANT FINDING	UTERUS ABSENT OVARI ES NORMAL	46 X X	NORMAL LE VEL	MEYER ROKITANSKY KUSTER HAUSER SYNDROME
6.NEHA	16 YRS	<u>WELL DEVELOPED</u>	SOF T	NO SIGNIFI CANT FINDING	VAGINA SHORT AND B LIND	NO SIGNIFI CANT FINDING	UTERUS ABSENT OVARIES NORMAL	46 X X	NORMAL LE VEL	MEYER ROKITANSKY KUSTER HAUSER SYNDROME
7. SEEMA	14 YRS	<u>WELL DEVELOPED</u>	SOF T	NO SIGNIFI CANT FINDING	VAGINA SHORT AND B LIND	NO SIGNIFI CANT FINDING	UTERUS ABSENT OVARIES NORMAL	46 X X	NORMAL LE VEL	MEYER ROKITANSKY KUSTER HAUSER SYNDROME
8. PHULA	15 YRS ( <u>SHORT</u> HEIGHT)	POORLY DEVELOPED BREAST WIDELY SPACED NIPPLES	SOF T	NO SIGNIFI CANT FINDING	NO SIGNIFICANT FINDING	NO SIGNIFI CANT FINDING	UTERUS PRESENT VER Y SMALL OVARIES	45 X O	HIGH	TURNERS SYNDROME
9. URMILA	13 YRS ( <u>SHORT</u> <u>HEIGHT</u> )	POORLY DEVELOPED BREAST WIDELY SPACED NIPPLES	SOF T	NO SIGNIFI CANT FINDING	NO SIGNIFICANT FINDING	NO SIGNIFI CANT FINDING	UTERUS PRESENT UNDERDEVELOPED OVARIES	45 X O	HIGH	TURNERS SYNDROME
10. SUGNI	14 YRS	POORLY DEVELOPED BR EAST	SOF T	NO SIGNIFI CANT FINDING	NO SIGNIFICANT FINDING	NO SIGNIFI CANT FINDING	UTERUS PRESENT <u>OVARIES ABSENT</u>	45 X Y	HIGH	GONADAL DYSGENESIS

## V. TREATMENT MODALITIES

For the treatment of congenital anomalies:[8] [9]

Treatment of imperforate hymen involves making a cruciate incision to open the vaginal orifice.

If a transverse septum is present, surgical removal is required.

Hypoplasia or absence of the cervix in the presence of a functioning uterus is more difficult to treat than other outflow obstructions. Surgery to repair the cervix is rarely successful, and hysterectomy is typically required. The ovaries should be retained to provide the benefits of estrogen and to allow for the possibility of future childbearing by removing mature oocytes for in vitro fertilization and transfer of embryos to a gestational carrier. If the vagina is absent or short, progressive dilation is usually successful. Females diagnosed with primary amenorrhea associated with all forms of gonadal failure and hypergonadotropic hypogonadism require cyclic estrogen and progestogen therapy for the initiation, maturation, and maintenance of secondary sexual characteristics. Therapy can be initiated with conjugated estrogens per day or estradiol per day. Females with short stature should not receive high doses of estrogen as this can cause the epiphyses to close prematurely. Hyperplasia can occur due to unopposed estrogen stimulation of the endometrium. To prevent hyperplasia, estrogen can be provided daily in combination with progestogen. Patients with gonadal streaks and mosaicism may be able to ovulate and conceive either following the initiation of estrogen therapy or spontaneously. If the diagnosis of 17 alpha-hydroxylase deficiency is established, treatment can be initiated with exogenous corticosteroid replacement therapy with either hydrocortisone or dexamethasone. Therapeutic measures should aim to target the underlying cause or primary amenorrhea. Craniopharyngiomas may be resected either during craniotomy or with a transsphenoidal approach. Germinomas are very radiosensitive; thus, surgery is seldom indicated. Hyperprolactinemia and prolactinomas may respond to dopamine agonists. Specific therapies can target underlying causes such as anorexia nervosa or malnutrition. Individuals diagnosed with Kallman syndrome or other etiologies for hypothalamic amenorrhea may be treated with hormone replacement therapy.

## VI. RESULTS

TOTAL 29 PATIENTS WERE DIAGNOSED AS PRIMARY AMENORRHEA IN OUTPATIENT DEPARTMENT OF OBSTETRICS AND GYNECOLOGY NMCH PATNA FROM JUNE 2021 TO 2022 JUNE. OF WHICH 10 CASES WERE FOLLOWED UP. THE MOST COMMON AGE GROUP OF PRESENTATION WAS 16 TO 18 YEARS.

- THE INCIDENCE IS 0.1 – 2 % IN REPRODUCTIVE AGE GROUP WOMEN.
- OUT OF THE 10 CASES :
- 4 HAD MEYER ROKITANSKY SYNDROME
- 3 HAD CRYPTOMENORRHEA
- 2 HAD TURNERS SYNDROME
- 1 HAD GONADAL DYSGENESIS
- 

## VII. CONCLUSION

- DEPENDING ON THE CAUSE OF PRIMARY AMENORRHEA, VARYING CLINICAL SEQUALAE ARE POSSIBLE. TIMELY EVALUATION AND CONSIDERATION OF A BROAD DIFFERENTIAL DIAGNOSIS HAVE IMPORTANT IMPLICATIONS FOR EMOTIONAL, PHYSICAL AND REPRODUCTIVE HEALTH IN YOUNG WOMEN.
- TREATMENT IS PRIMARILY SURGICAL IN CONGENITAL ANOMALIES AND OUTFLOW TRACT OBSTRUCTION. GONAECTOMY IS CONSIDERED IN GONADAL DYSGENESIS CASES. CYCLICAL HORMONAL THERAPY IS INSTITUTED IN PREMATURE OVARIAN FAILURE CASES. COUNSELLING ABOUT GENDER IDENTITY IS DONE EMPATHICALLY.

majority of cases of primary amenorrhea are caused by anatomical defects, elevated follicle-stimulating hormone (FSH) levels, hyperprolactinemia, hypothalamic amenorrhea, or polycystic ovary syndrome (PCOS).[2] Gonadal dysgenesis includes most commonly Turner syndrome, which makes up to 43% of primary amenorrhea cases. Anatomical Defects When the uterus and vagina are partially or completely absent in the presence of otherwise normal female sexual characteristics, the diagnosis is typically Mullerian agenesis, which accounts for approximately 10% of primary amenorrhea cases. Other anatomical defects include imperforate hymen or transverse vaginal septum, both of which create a partial or complete vaginal blockage, and isolated absence of the vagina or cervix.[3] Elevated FSH Levels Elevated follicle- Stimulating hormone levels can indicate gonadal dysfunction. In individuals with XX chromosomes, gonadal failure refers to ovarian failure.[3] Hyperprolactinemia Prolactin is a pituitary hormone that is involved in many reproductive functions. The secretion of prolactin causes an inhibition of the secretion of gonadotropin-releasing hormone, which negatively modulates the secretion of pituitary hormones responsible for the gonadal function.[4] Hypothalamic Amenorrhea This condition causes cessation or absence of menses due to a functional disorder of the hypothalamus. Polycystic Ovary Syndrome PCOS is the most common cause of amenorrhea in females with evidence of androgen excess. It is a common reproductive and endocrinologic disorder. The three main characteristics of this syndrome are hyperandrogenism, polycystic ovaries, and ovulatory dysfunction.[5] Hypopituitarism, weight loss, anorexia nervosa, and isolated gonadotropin-releasing hormone (GnRH) deficiency can also cause amenorrhea. Constitutional delay of puberty and chronic systemic disease or acute illness can also lead to amenorrhea. The World Health Organization (WHO) categorized amenorrhea into three groups. WHO group I includes females with no evidence of endogenous estrogen production, normal or low follicle-stimulating hormone (FSH) levels, normal prolactin levels, and no evidence of lesions in the hypothalamic- pituitary region. WHO group II includes females who can produce estrogen and have normal levels of prolactin and FSH. WHO group III includes females with increased serum FSH, which indicates gonadal insufficiency or failure.[6][7]

## VIII. DIFFERENTIAL DIAGNOSIS OF PRIMARY AMENORRHEA

- Primary hypothalamic amenorrhea
- Emotional/physical stress
- Female athlete triad
- Hypogonadotropic hypogonadism (Kallman syndrome)
- Malnutrition
- Chronic disease state
- Constitutional delay
- PCOS
- Outflow tract obstruction

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