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Original Research Article

Clinical and genetic profile of children with delayed puberty: a tertiary care hospital experience

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ABSTRACT

Background: Delayed puberty is defined as the absence of pubertal onset by 13 years in girls and 14 years in boys. It may result from constitutional delay, hypothalamic-pituitary disorders, or primary gonadal failure. Early identification of the underlying cause is essential for timely treatment and improved psychosocial outcomes. This study aimed to evaluate the clinical and genetic profile of children presenting with delayed puberty in a tertiary care hospital.
Methods: A retrospective descriptive observational study was conducted in the Department of Paediatric Endocrinology from June 2019 to June 2026. Children aged 13-18 years with delayed puberty and complete medical records were included. Demographic details, clinical features, anthropometry, hormonal investigations, imaging, genetic testing, diagnosis, and treatment modalities were analysed.
Results: A total of 42 adolescents were included, comprising 27 females (64.3%) and 15 males (35.7%). Consanguinity was present in 25.6%, and family history of delayed puberty in 29.3%. Among girls, breast development was present in 81.5%, but menarche had occurred in only 3.7%. Among boys, testicular enlargement was present in 13.3%. Hypergonadotropic hypogonadism (HH) was the most common diagnosis (52.4%), followed by congenital hypogonadotropic hypogonadism (CHH) (28.6%) and constitutional delay of growth and puberty (CDGP) (11.9%).
Conclusions: Comprehensive clinical, hormonal and genetic evaluation is crucial for accurate diagnosis and individualized management.

Keywords: Delayed puberty, Hypergonadotropic hypogonadism, Congenital hypogonadotropic hypogonadism, Constitutional delay of growth and puberty, Turner syndrome, Pediatric endocrinology, Genetic evaluation, Pubertal induction

INTRODUCTION

Puberty is a critical developmental phase characterized by the acquisition of secondary sexual characteristics, accelerated linear growth, and attainment of reproductive maturity. It represents a complex interplay between genetic, environmental, nutritional, and neuroendocrine factors, primarily regulated by the hypothalamic-pituitary-gonadal (HPG) axis.[1,2] The onset and progression of puberty follow a predictable sequence; however, considerable inter-individual variability exists, influenced largely by genetic determinants and environmental exposures.[3,4] Delayed puberty is traditionally defined as the absence of testicular enlargement by 14 years in boys or lack of thelarche by 13 years in girls, or a prolonged interval exceeding 4 years between onset and completion of pubertal development(pubertal arrest).[5,6]

Delayed puberty is a relatively common clinical presentation in pediatric endocrinology, accounting for significant psychological stress and reduced quality of life among affected adolescents.[7] It can broadly be categorized into CDGP, CHH, HH, and functional

causes.[2,8] CDGP is the most common etiology, often familial and self-limiting, whereas pathological causes such as congenital or acquired HH and primary gonadal failure require prompt identification and intervention.[9,10] Distinguishing between CDGP and CHH remains a diagnostic challenge, often necessitating dynamic stimulation tests, bone age and newer diagnostic modalities like inhibin B.[11]

Recent advances have highlighted the role of genetic factors in the regulation of pubertal timing. Mutations in genes involved in GnRH neuronal migration, secretion, and action have been implicated in CHH, including Kallmann syndrome and normosmic idiopathic CHH.[12,13] Similarly, chromosomal abnormalities such as Turner syndrome and Klinefelter syndrome represent common causes of HH in females and males, respectively.[10,14] Emerging evidence suggests that even CDGP may have a genetic basis, with several candidate genes influencing pubertal onset.[4,12]

Despite these advancements, there remains a paucity of data from the Indian population regarding the clinical and genetic spectrum of delayed puberty. Most available literature is derived from Western cohorts, limiting the generalizability of findings to diverse ethnic populations.[2,13] Understanding the phenotypic variability and underlying molecular mechanisms in different populations is essential for accurate diagnosis, prognostication, and genetic counseling. Therefore, this study aims to evaluate the clinical presentation and genetic profile of children presenting with delayed puberty in a tertiary care setting, thereby contributing to the existing body of knowledge and improving patient care outcomes.

METHODS

A retrospective descriptive observational study conducted at the Department of Paediatric Endocrinology in a tertiary care hospital in South India. The study setting was a specialized endocrine unit catering to a large referral population, ensuring a diverse spectrum of cases with delayed puberty. The study period extended from June 2019 to June 2026.

The study population included children and adolescents aged between 13 and 18 years who presented with clinical features suggestive of delayed puberty. Patients fulfilling the above defined criteria and who had complete clinical and laboratory records were included in the study.

Children with known chronic systemic illnesses that could result in transient delay in puberty (FHH-Functional Hypogonadotrophic hypogonadism) were excluded from the study. These included conditions such as thalassemia, cystic fibrosis, coeliac disease, chronic kidney disease, diabetes mellitus, and hypothyroidism. Patients with incomplete records/follow-up data were also excluded.

Data were collected retrospectively from hospital medical records using a structured proforma. The variables recorded included demographic details (age, sex), clinical history (family history of delayed puberty, consanguinity, birth and developmental history), and presenting complaints. Detailed clinical examination findings such as anthropometric measurements (height, weight, body mass index), sexual maturity rating (SMR), and presence of dysmorphic features or midline defects were documented.

Biochemical evaluation included measurement of serum luteinizing hormone (LH), follicle-stimulating hormone (FSH), and sex steroids (testosterone in males and estradiol in females). Additional hormonal investigations such as anti-Müllerian hormone (AMH), inhibin B, thyroid function tests, prolactin levels, and other relevant investigations were recorded wherever available. Dynamic testing, including gonadotropin-releasing hormone (GnRH) stimulation test, was analyzed to differentiate between CDGP and hypogonadotropic hypogonadism in 2 patients.

Radiological investigations such as bone age assessment using wrist X-ray by Tanner Whitehouse 3, pelvic ultrasonography in girls, and magnetic resonance imaging (MRI) of the brain for evaluation of hypothalamic-pituitary abnormalities for CHH were included. Genetic analysis, including karyotyping and targeted genetic testing, was reviewed to identify chromosomal abnormalities and specific gene mutations associated.

All collected data were entered into a structured database and analyzed systematically. Continuous variables were expressed as mean±standard deviation or median with interquartile range, depending on distribution. Categorical variables were presented as frequencies and percentages.

RESULTS

A total of 42 participants were included in the study, comprising 27 females (64.3%) and 15 males (35.7%). Consanguinity was present in 25.64% of cases. Developmental delay was noted in a minority (10.26%).

Clinical assessment of pubertal characteristics (Table 1) revealed varying degrees of pubertal development among the participants. Among females (n=27), breast enlargement was present in 22 individuals (81.48%). Pubic or axillary hair was observed in 18 females (66.67%), whereas none had menarche. This showed pubertal arrest.

Among males (n=15), testicular enlargement was present in only 2 participants (13.33%). Voice change was observed in 1 participant (6.67%). Similarly, adrenarche

was present in 1 participant (6.67%). Overall, the findings suggest that most male participants had minimal pubertal progression at presentation.

Growth-related parameters (Table 2) showed that among participants with available data (n=34), only 4 individuals (11.76%) had experienced a height spurt. A history of being small for gestational age (SGA) was present in 3 out of 41 participants (7.32%), suggesting that intrauterine growth restriction was uncommon in this cohort. Family history of delayed puberty was reported in 12 out of 41 participants (29.27%), indicating a possible familial or genetic contribution in nearly one-third of cases. Family history of short stature was present in 3 out of 40 participants (7.50%) suggesting that familial short stature was relatively uncommon among the study population.

Hormonal profiling (Table 3) demonstrated that the majority, 33 individuals (78.57%) had LH levels >0.6 IU/L. A smaller proportion had LH between 0.3-0.6 IU/L (3 participants; 7.14%), while 6 participants (14.29%) had LH levels <0.3 IU/L.

For FSH (n=42), 22 participants (52.4%) had levels ≥10 IU/L, whereas 10 (23.81%) had levels <3 IU/L, and between 3-10 each indicating that most individuals had elevated gonadotropin levels. The 3-10 was considered normal pubertal range and above 10 IU/L were considered HH.

Among those tested for AMH (n=9), 5 participants (55.56%) had levels ≥20 ng/mL, while 4 (44.44%) had levels <20 ng/mL. AMH done in HH patients is useful for fertility options whereas it is done in hypogonadotrophic hypogonadism patients to differentiate between CDGP (>20) and CHH (<20).[17]

Similarly, for inhibin B (n=9), 7 participants (77.78%) had levels ≥28.5 pg/ml suggestive of CDGP, whereas 2 participants (22.22%) had levels <28.5.

GnRH Stimulation test was done in 2 patients to differentiate between CHH and CDGP. In 1 male patient, Inhibin B was suggestive of CDGP (>35) but GnRH Stimulation test done with triptorelin (LH peak<8) and genetic testing were suggestive of CHH-Xp21 continuous gene deletion syndrome.[15,16] Another male patient-GnRH Stimulation test with Leuprolide was suggestive of CDGP (LH peak>5) but genetic testing came suggestive of CHH-IL17RD.

The distribution of primary diagnoses and their underlying genetic etiologies are summarized in Tables 4 and 5. HH emerged as the most common cause, accounting for 23 of the 42 participants (54.76%). This condition showed a marked female predominance, affecting 18 of 27 females (66.67%) compared with 5 of 15 males (33.33%). CHH was the second most common diagnosis, identified in 12 participants (28.57%), including 8 females (29.63%) and 4 males (26.67%). CDGP was observed exclusively among males, affecting 5 participants (33.33% of male cases. CHH associated with adrenal insufficiency (AI) was identified in two males (13.3%), while a single female participant (3.70%) was diagnosed with an anatomical defect-Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH).

MRI brain was done in the 11 CHH patients, 8 of which were normal and 3 showed pituitary hypoplasia consistent with IGHD/CPHD.

Cytogenetics(karyotype) differentiated the Turner syndromes into 4 monosomy, 4 mosaic and 1 isochromosome.

Whole exome sequencing further demonstrated the heterogeneous nature of delayed puberty (Table 5).

Parental segregation studies were offered to the patients with VUS but affordability was an issue.

The distribution of treatment modalities is presented in Table 6. Estradiol valerate-based replacement therapy was the most frequently administered treatment, prescribed to 19 participants (31.7%), for pubertal induction in females. GH therapy was administered to 11 participants (18.3%), (9- Turner syndrome, 2- GHD). Testosterone therapy was used in 9 participants (15.0%) for induction of puberty in males. All the patients demonstrated progression in pubertal status after initiation of treatment with some females starting menstrual cycles on their own while some had to be initiated on progesterone.

Progesterone therapy was prescribed to 7 participants (11.7%) following approximately two years of estrogen therapy to induce withdrawal bleeding and establish cyclical hormonal replacement. Conservative management was adopted in 4 participants (6.7%), representing cases of CDGP in whom observation and regular follow-up were considered appropriate. Short course of Testosterone therapy was offered to them, which only 1 patient opted for.

Additional treatment modalities included thyroxine replacement in 5 participants (12.0%) with associated thyroid dysfunction and transdermal estrogen therapy in 2 participants (3.3%) who developed transaminitis on oral estrogen. Hydrocortisone, fludrocortisone were started in the 2 patients with AI and human chorionic gonadotropin (HCG) was started in 1 participant (1.7%) with FGFR1 mutation. Risedronate sodium was initiated in one patient because of reduced bone mineral density documented on dual-energy X-ray absorptiometry (DEXA) scan and history of fractures.

In addition to medical management, one female participant with mosaic Turner syndrome [46,XY (44 metaphases)/45,XO (6 metaphases)] and an associated

DHX31 mutation causing 46,XY sex reversal syndrome underwent gonadal biopsy which demonstrated dysgenetic gonads, following which prophylactic laparoscopic gonadectomy was performed because of the increased risk of gonadoblastoma associated with Y-chromosome material in dysgenetic gonads. Six boys had undergone orchidopexy in infancy period, 4 of whom developed HH later and 2 CHH.

Table 1: Clinical pubertal characteristics.

Parameters		Category	N	Percentages (%)
Females, (n=27)	Breast enlargement	Present	22	81.48
		Absent	5	18.52
	Pubic/axillary hair	Present	18	66.67
		Absent	9	33.33
	Menarche	Present	0	0
		Absent	27	100
Males, (n=15)	Testicular enlargement	Present	2	13.33
		Absent	13	86.67
	Voice change	Present	1	6.67
		Absent	14	93.33
	Adrenarche	Present	1	6.67
		Absent	13	93.33

Table 2: Growth and family history.

Variables	Category	N	Percentages (%)
Height spurt, (n=34)	Present	4	11.76
	Absent	30	88.24
SGA at birth, (n=41)	Yes	3	7.32
	No	38	92.68
Family history-delayed puberty, (n=41)	Present	12	29.27
	Absent	29	70.73
Family history-short stature, (n=40)	Present	3	7.50
	Absent	37	92.50

Table 3: Hormonal and biochemical profile.

Parameters	Category	N	Percentages (%)
LH (IU/L), (n=42)	<0.3	6	14.29
	0.3-0.6	3	7.14
	>0.6	33	78.27
FSH (IU/L), (n=42)	<3	10	23.81
	≥3-10	10	23.8
	>10	22	52.4
AMH (ng/mL), (n=9)	<20	4	44.44
	≥20	5	55.56
Inhibin B, (n=9)	<28.5	2	22.22
	≥28.5	7	77.78

Table 4: Primary diagnosis by gender.

Diagnosis	Female		Male		Total
	N	%	N	%	
Anatomical defect	1	3.7	0	0	1
CDGP	0	0	5	33.3	5
CHH	8	29.6	3	20	11
HH	18	66.6	5	33.3	23
CHH with AI	0	0	2	13.3	2
Total	27	100	15	100	42

Table 5: Genetic and etiological distribution of delayed puberty.

HH	N	Genetic diagnosis	N	Zygosity	Variant ACMG classification	Inheritance
	23	Turner syndrome	9			
		Klinefelter syndrome	2			
		TWNK	1	Compound heterozygous	LP	AR
		FSHR	1	Heterozygous	VUS	AR
		DIAPH2	1	Heterozygous	VUS	XLD
		ERCC6/FANCM	1	Heterozygous	VUS	AD/AR
		DHX31	1	Heterozygous	VUS	AD
		MAP3K1	1			
		AMH	1	Homozygous	VUS	AR
		Swyer syndrome	1			
		Idiopathic	4			
CHH	**N**	**Genetic diagnosis**	**N**	**Zygosity**	**Variant ACMG classification**	**Inheritance**
	11	FGFR1	2	Heterozygous	LP/P	AD
		IL17RD	1	Heterozygous	VUS	AD/AR
		NSMF	1	Heterozygous	VUS	AD
		PROP1	1	Homozygous	LP	AR
		TRMT10A/TCAP	1	Homozygous/Heterozygous	P/LP	AR/AD
		POL3RA	1	Homozygous	VUS	AR
		Idiopathic	2			
		IGHD	2			
HH with AI		Xp21 deletion/ DAX1-associated adrenal hypoplasia congenita	1	Hemizygous		XLR
		DAX1 mutation	1	Hemizygous		XLR
CDGP	**N**					
	5					
Anatomical defect	**N**	**Diagnosis**				
	1	MRKH syndrome				

*LP-Likely pathogenic, P. Pathogenic, VUS-Variant of uncertain significance.

Table 6: Treatment modalities.

Treatments	N	Percentages (%)
Estradiol valerate	19	31.7
Testosterone therapy	9	15.0
Growth hormone therapy	11	18.3
Conservative management	4	6.7
Progesterone	7	11.7
Risedronate sodium	1	1.7
Thyroxine	5	12
Hydrocortisone	2	3.3
HCG	1	1.7
Fludricortisone	2	3.3
Estraderm patch	2	3.3

DISCUSSION

Delayed puberty is a common pediatric endocrine concern and includes a broad spectrum of conditions ranging from CDGP to permanent disorders of the HPG axis. In the present study, 42 adolescents with delayed puberty were evaluated, of whom 27 (64.3%) were females and 15 (35.7%) were males. This female predominance differs from most published studies, where boys predominate because CDGP is more common in

males. Sedlmeyer et al in a large series of 232 adolescents, reported that approximately two-thirds were males, and CDGP was the most frequent diagnosis.[6] Similarly, Varimo et al observed a male predominance of nearly 70%.[7] The higher proportion of females in our study likely reflects referral bias, as girls with primary amenorrhea and gonadal dysgenesis are more likely to be referred to tertiary endocrine centers.

Howard et al emphasized that consanguinity increases the likelihood of autosomal recessive forms of CHH, which may explain the relatively high prevalence of CHH in our cohort (25.64 %).[2]

Sedlmeyer et al similarly noted that girls often present with thelarche without menarche, whereas boys typically present with absent testicular enlargement.[6,10]

They also reported a positive family history in 50-75% of adolescents with CDGP. The lower proportion in our study (29.27%) is likely due to the predominance of pathological causes due to referral bias. Small for gestational age was documented in 7.32%.[10]

They reported CDGP in nearly 53% of cases, while permanent hypogonadism constituted a smaller proportion. The etiological distribution in our findings differ from several Western studies where CDGP has been reported as the leading etiology.

Similarly, Varimo et al observed CDGP in approximately 45% of patients and CHH in 23% of cases.[7]

The marked female predominance of HH observed in the present study is consistent with the established association between delayed puberty in girls and gonadal dysgenesis. Turner syndrome and POF are among the most common causes of HH in adolescent females.[10,14]

Genetic analysis in the present study identified several important molecular etiologies associated with HH, including mutations involving TWNK, FSHR, DIAPH2, ERCC6/FANCM, DHX31, MAP3K1, and AMH genes. However, few were heterozygous mutations in an autosomal recessive condition with ACMG classification of VUS, meaning they were not disease-causative like FSHR mutation.

Mutations in the TWNK gene are associated with Perrault syndrome, characterized by ovarian dysgenesis and sensorineural hearing loss. Defects affecting mitochondrial function can impair gonadal development and contribute to pubertal failure.[2] Similarly, mutations in the FSHR gene result in ovarian resistance to FSH, leading to impaired folliculogenesis and HH.[12]

POF associated with DIAPH2 and ERCC6/FANCM mutations further supports the growing evidence linking defective DNA repair pathways with gonadal insufficiency. Genes involved in ovarian follicular maintenance and genomic stability are critical for preservation of reproductive function.[9] These mutations then accelerate follicular depletion, resulting in early ovarian failure.

The identification of 46 XY sex reversal syndrome associated with DHX31 and MAP3K1 mutations also highlights the importance of genes regulating testicular differentiation. MAP3K1 mutations are recognized causes of differences in sex development (DSD) and gonadal dysgenesis. Abnormalities in genes involved in testicular determination frequently present with delayed puberty and impaired virilization.[2] Very few mutations in DHX31 have been reported.

The AMH gene plays a central role in Müllerian duct regression during male fetal development. Boehm et al emphasized that mutations affecting AMH signaling pathways can result in DSD and pubertal abnormalities-PMDS.[8]

CHH results from deficient neuronal migration, secretion or action of GnRH, leading to impaired activation of the hypothalamic-pituitary-gonadal axis.

Several pathogenic variants associated with CHH were identified in the present study, including DAX1, FGFR1, IL17RD, NSMF, PROP1, TRMT10A/TCAP and POLR3A mutations.

DAX1 (NR0B1) mutations/deletions are classically associated with X-linked adrenal hypoplasia congenita and CHH.[8]

IL17RD mutations have been implicated in Kallmann syndrome and normosmic CHH due to defective fibroblast growth factor (FGF) signaling involved in GnRH neuronal migration.[2] FGFR1 mutations can cause normosmic or anosmic CHH supporting our finding of 1 patient with anosmia and 1 with normosmia.

Mutations in the NSMF gene are also known to impair migration of GnRH neurons during embryogenesis. Similarly, PROP1 mutations affect pituitary organogenesis and may result in combined pituitary hormone deficiency (CPHD). Harrington and Palmert et al highlighted the importance of evaluating other anterior pituitary hormone deficiencies in adolescents presenting with delayed puberty and growth failure.[11]

The tRNA methyltransferase 10 homologue A (TRMT10A) mutation causes short stature (may be growth hormone deficiency), intellectual disability, microcephaly, diabetes and POF.[17]

POL3RA (RNA polymerase III subunit A) gene mutations are associated with 4H Syndrome (hypomyelination, hypodontia, CHH).[18]

Two cases of growth hormone deficiency (GHD) were associated with CHH, both of whom demonstrated progression in SMR following GH therapy.

The presence of anatomical defects in one female participant, diagnosed as Mayer-Rokitansky-Küster-Hauser syndrome, further emphasizes the necessity of evaluating structural reproductive tract anomalies.[10]

Dunkel et al recommended gradual induction of puberty with low-dose estrogen or testosterone to mimic normal physiological progression, which was reflected in our management approach.[13]

This study has a few limitations. The retrospective design relied on medical records, which led to missing data for some variables such as AMH and inhibin B. The sample size was relatively small and drawn from a single tertiary care center, limiting the generalizability of the findings. Referral bias likely resulted in overrepresentation of pathological causes. In addition, genetic testing was not uniformly available for all participants, which may have led to underdiagnosis of specific molecular etiologies. Long-term fertility data was not available.

CONCLUSION

The present study highlights the heterogeneous clinical and etiological spectrum of delayed puberty in children and adolescents attending a tertiary care pediatric endocrine center. A systematic multidisciplinary approach is essential for early diagnosis, timely intervention, and improved long-term reproductive and psychosocial outcomes.

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