Section: Paediatrics

Original Research Article

ETIOLOGICAL PROFILE OF SHORT STATURE IN CHILDREN: A CROSS-SECTIONAL STUDY

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Abstract

Background: Short stature is a significant concern in pediatric endocrinology, requiring detailed evaluations to determine its etiology. This study aimed to identify the causes of short stature in children and adolescents and assess the impact of hormonal and systemic factors on growth impairment. **Material and Methods:** A cross-sectional study was conducted in the Department of Pediatrics, World College of Medical Sciences Research and Hospital, Jhajjar. A total of 46 children aged 1-14 years with short stature (height more than 2 SD below the mean for their age and sex or height <3rd percentile) were included. Anthropometric measurements, biochemical tests, growth hormone stimulation tests, and imaging studies were conducted to assess the underlying causes. **Results:** The most common cause of short stature was constitutional delay of growth and puberty (28.2%), followed by systemic illness (19.5%) and familial short stature (17.3%). Endocrine causes, including growth hormone deficiency (8.6%) and hypothyroidism (4.3%), were less frequent. Rickets (10.8%) and celiac disease (6.5%) were also contributing factors. Skeletal dysplasia was observed in 4.3% of cases. The prevalence of CDGP was higher in males, while systemic illnesses were more common in females. **Conclusion:** Most cases of short stature in this study were due to non-endocrine causes, emphasizing the importance of a structured diagnostic approach, including screening for systemic illnesses and nutritional deficiencies before hormonal testing. Early diagnosis and intervention are critical for optimizing growth outcomes and minimizing psychosocial consequences.

INTRODUCTION

The primary symptom that requires a careful and comprehensive evaluation by the medical professionals working in pediatric practice is short stature, which bears physical, emotional, and social stigma for both the kids and the parents. The usage of evaluation standards is one of the major problems that individuals involved in the assessment of short stature in India face. If national standards are available, Goldstein and Tanner contend that they should be used as they are derived from representative samples of the population.[1] Clinical characteristics and hormonal testing are used to diagnose growth hormone insufficiency in children who are short in stature. A development velocity of less than 4 cm/year and extreme short height are typical symptoms of growth hormone insufficiency in children.[2]The dynamics of the growth process, which determine body height and growth spurt, are influenced by a wide range of internal and external stimuli. Growth process regulation is influenced by a number of endocrine factors like thyroid hormones andgrowth hormone.[3] When a person's height is ≤ 2standard deviations (SD) below the average for their age and sex, they are considered short stature.Growth hormone deficiency and hypothyroidism are examples of endocrine causes of short stature, while familial, chronic, and "constitutional delay of growth and puberty" (CDGP),[4] are examples of non-endocrine causes. In CDGP, there are unrecognized variations within the pituitary-gonad in addition to the growth hormone-IGF-1 axis, which causes delayed skeletal maturity and delayed puberty spurt.[5] The final adult height is determined by a child's genetic makeup. A child whose height falls within the range of its "parents" is familial short stature.[6]Auxology, bone age, biochemical tests, and magnetic resonance imaging are all helpful in the diagnosis of growth hormone deficiency.[7]The aim of study was to identify the causes of short stature in children and adolescents.

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MATERIALS AND METHODS

This present study was conducted in the Department of Paediatrics, World College of Medical Sciences Research and Hospital, Jhajjar. The study was approved by the institute's ethical board. It was a cross-sectional study conducted from January, 2023 and October, 2023. Children aged 1-14 years with height more than 2 standard deviations (SD) below the mean for specific age and sex or height <3rd percentile. Written informed consent was obtained from the parents/guardians. 46 patients, all fulfilling the criteria of short stature, made up the study population. A detailed history, including developmental history, history of systemic illness, and family history of short stature was taken. Children underwent complete anthropometric examination, including height, weight, body mass index (BMI), upper segment-lower segment ratio, and MPH and plotted on growth charts. A thorough clinical examination was done and first-line tests including complete blood count, erythrocyte sedimentation rate, serum creatinine, serum alanine aminotransferase, free T4, thyroid stimulating hormone and insulin-like growth factor-1 levels were done in all children. Bone age estimation was done by doing x ray of the left hand with the wrist by Greulich and Pyle method.Second-level investigations like screening for celiac disease, arterial blood gas, follicle-stimulating hormone, karyotyping and further testing based on history and clinical examination were done on a case-to-case basis.In children with suspected GHD, GH stimulation tests with clonidine were done. Testing the patients' basal GH level and its level following clonidine stimulation at a dose of 0.15 mg/m2 was the first step in the GH screening process. Blood samples were taken at zero, thirty minutes, one hour, one and a half, and two hours. GH estimation was done by chemiluminescent immunoassay(CLIA).GH measurement had a lower limit of 0.1 ng/ml. A sufficient secretory response of GH was defined as a peak GH level of 7 ng/ml or higher.[8] In the appropriate clinical context, people are considered to be GH deficient if they fall below this cut-off.[9]If child was diagnosed to have GH deficiency with clonidine stimulation test then GH stimulation test was done with glucagon. MRI of the brain and sellawas done to confirm GHD.Data entry and analysis were conducted using SPSS version 20.

RESULTS

For the study, 46 patients with shortstature between the ages of 1 and 14 were included. Out of which 26 were male and 20 female children (Table 2). Constitutional Delay of Growth and Puberty (CDGP) is the most common cause, accounting for 28.2% of cases, with a higher prevalence in males (30.7%) compared to females (25%). This suggests that delayed puberty plays a significant role in male short stature. Systemic illness is the second most frequent cause (19.5%), more commonly affecting females (25%) than males (14%), indicating that chronic health conditions may contribute more to growth impairment in girls. Familial Short Stature (FSS) is the third leading cause (17.3%), with a slightly higher prevalence in males (19.2%) than in females (15%), emphasizing the genetic influence on height.

Sex differences are evident in the data, with males being more affected by CDGP and FSS, while females show a higher prevalence of Systemic illness and rickets (15% vs. 7% in males). Growth hormone deficiency (8.6%) and rickets (10.8%) were observed in a smaller proportion of cases, while less frequent causes included hypothyroidism (4.3%), celiac Disease (6.5%), and skeletal Dysplasia (4.3%).

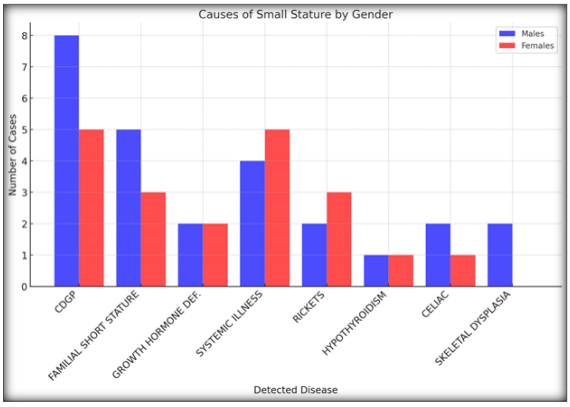

Figure1: Shows the Causes for small stature among study groups

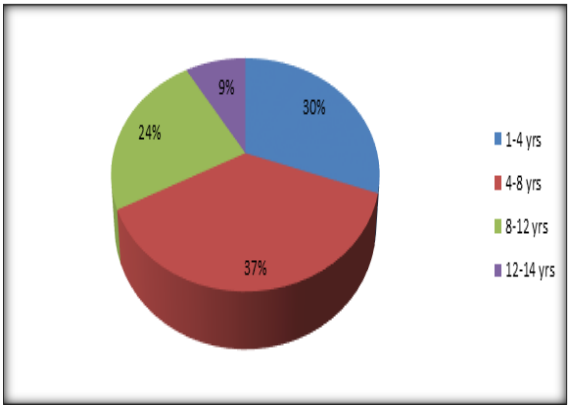

Figure 2: Shows the percentage of participants according age group.

Table 4: Causes of short stature among study groups

Detected Disease	Males	Females	Total No. (%)
Cdgp	8 (30.7)	5 (25)	13 (28.2)
Familial Short Stature	5 (19.2)	3 (15)	8 (17.3)
Growth Hormone Deficiency	2 (7)	2 (10)	4 (8.6)

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Systemic Illness	4 (14)	5 (25)	9 (19.5)
Rickets	2 (7)	3 (15)	5 (10.8)
Hypothyroidism	1 (3.8)	1 (5)	2 (4.3)
Celiac	2 (7)	1 (5)	3 (6.5)
Skeletal Dysplasia	2 (7)	0 (0)	2 (4.3)
Total	26	20	46

Table 2: Patient's height (mean ±standard deviation) according to age group

Age in years	No. (%)	Height in cm (Mean ±SD)
1-4	14 (30.43%)	82.12±14.26
4-8	17 (36.95%)	97.32±15.14
8-12	11 (23.91%)	110.42±16.54
12-14	04 (8.7%)	123.02±15.16

DISCUSSION

Short stature is a common concern in pediatric endocrinology, often leading to extensive investigations to determine its etiology. This study aimed to identify the causes of short stature in children and adolescents and to assess the role of hormonal and systemic factors in growth impairment. Our findings suggest that constitutional delay of growth and puberty (CDGP) is the most frequent cause of short stature, with a higher prevalence in males compared to females. This observation aligns with previous studies, which indicate that CDGP is a predominant cause of short stature, particularly among adolescent boys.[8,9] CDGP is a normal variant of growth and typically does not require medical intervention beyond reassurance and monitoring.[10]Familial short stature accounted for 17.3% of cases, supporting the notion that genetic factors significantly influence growth patterns.[11]The importance of parental height in predicting a child's final height is well established, and children with FSS often exhibit normal growth velocity and bone age that is consistent with their chronological age.[12]Systemic illness was more prevalent among females than males, suggesting that chronic health conditions such as malnutrition, renal disorders, and chronic infections may disproportionately impact female growth.[13] Studies have reported a similar trend, with chronic diseases contributing to a substantial proportion of pathological short stature cases.[14] Endocrine causes of short stature, such as growth hormone deficiency (GHD) and hypothyroidism were relatively uncommon in our study population compared to findings from other studies, which have reported higher rates of GHD.[15]This difference may be due to variations in referral patterns, diagnostic criteria, and genetic factors influencing growth hormone secretion.[16] It is noteworthy that hypothyroidism contributed to significant bone age retardation in affected children, reinforcing the need for early thyroid screening in short children.[17] Rickets and celiac disease were notable contributors to short stature in this cohort. Rickets, caused by vitamin D deficiency, was more prevalent in females, consistent with previous reports highlighting the higher susceptibility of girls to nutritional deficiencies.[18]Celiac disease, though less common, should be considered in children with unexplained growth failure, especially in regions with a high prevalence of gluten intolerance.[19]Skeletal dysplasia, though rare remains a crucial differential diagnosis in children with disproportionate short stature. Advanced imaging and genetic testing are essential for confirming the diagnosis.[20]Our study underscores the importance of a structured diagnostic approach to short stature, incorporating clinical, anthropometric, biochemical, and radiological assessments. Given that the majority of cases were due to non-endocrine causes, targeted screening for systemic illnesses and nutritional deficiencies is recommended before resorting to growth hormone testing.[21]Early intervention and appropriate management can significantly improve growth outcomes and reduce the psychosocial impact of short stature on affected children and their families.[22]Future research should focus on longitudinal follow-ups to assess the impact of various interventions on final adult height, as well as the role of genetic and environmental factors in growth regulation.[23]

CONCLUSION

This study highlights the multifactorial etiology of short stature, with the majority of cases attributed to non-endocrine causes such as constitutional delay of growth and puberty, familial short stature, and systemic illnesses. Although endocrine disorders, including growth hormone deficiency and hypothyroidism, were observed, they were relatively uncommon. The findings emphasize the importance of a comprehensive diagnostic approach, prioritizing systemic and nutritional assessments before considering hormonal testing.

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