Journal of Population Therapeutics & Clinical Pharmacology

RESEARCH ARTICLE
DOI: 10.53555/r7zjdr76

ETIOLOGICAL PROFILE OF SHORT STATURE IN CHILDREN PRESENTING TO A TERTIARY CARE HOSPITAL

Mujeeb Ur Rehman[1], Irfan Ullah[2], Said Ayaz[3], Sajid Ur Rehman[4*], Muhammad Saleem[5], Aftab Alam[6], Mishkat Ullah[7]

[1,2]Assistant Professor, Department of Diabetes and Endocrinology, Lady Reading Hospital, Peshawar
[3]Consultant Pediatric & Neonatal Medicine, Polyclinic Hospital Peshawar KP Employees Social Security Institution
[4*]Children Specialist Health Department Women and Children Hospital Karak, Pakistan
[5]Trainee Registrar Children Ward, Women and Children Hospital Bannu, Pakistan
[6]Registrar Children Ward Women & Children Hospital Karak, Pakistan
[7]Combined Military Hospital, Rawalpindi Pakistan

***Corresponding author:** Sajid Ur Rehman
*Email: hafizamc1986@gmail.com

ABSTRACT

Introduction: Short stature for height complaint is prevalent in both the first and second tiers of healthcare delivery and may be attributed to physical health, endocrine, nutrition, genetic or systematic, and acquired conditions. This paper aims to establish the need for early identification of the patient's condition and administration of the right course of action to improve growth results and quality of life quality.

Objective: To establish the prevalence of short stature in children attending a tertiary care hospital and investigate the common causes of short stature.

Materials and Method: The present cross-sectional study was conducted Department of Diabetes and Endocrinology, Lady Reading Hospital, Peshawar, from April, 2024 to September, 2024. The children included in the study were 120 children of short stature aged 2 –18 years with clinical history, general examination, laboratory examination, and radiological examination.

Results: These include constitutional delay of growth 23.3%, short stature due to family history 16.7%, growth hormone deficiency 15%, nutritional causes 13.3%, and hypothyroidism 10%. The other diagnoses that were also noted included chronic illnesses, syndromes, and celiac diseases.

Conclusion: Early and structured evaluation is essential to diagnose treatable causes of short stature. Awareness and timely interventions can improve growth outcomes.

Keywords: Short stature, growth disorders, pediatric endocrinology, constitutional delay, growth hormone deficiency, hypothyroidism, nutritional deficiency.

INTRODUCTION

Growth delay in children is one of the most frequent diagnoses and implies the child's referral to an endocrinologist or another specialist. Generally, it is defined as a height that is lower than 2 Standard Deviations below the mean height, which is usually expected to be obtained from the appropriate population of the given age and sex. It has implications for both the physiologic and psychosocial

well-being of a person. Adaptable tests are crucial to diagnosis and classification and important for proper treatment and assessment (1). Diagnosis and classification are critical in this aspect in that accurate diagnosis ensures that patients receive the proper treatment and, consequently, improved prognosis (1). The rates of short stature and the contributing factors are known to vary based on geographical, socio-economic, and accessibility to health facilities. This learning indicates that some of the known causes of loss of height among children include endocrine disorders such as growth hormone deficiency, hypothyroidism, familial short stature, and constitutional delay of growth and puberty (1).

Likewise, a similar study with children from rural tertiary care hospitals in India has highlighted that most children visiting hospitals in India have idiopathic ROC and other endocrine diseases because the developed countries lack proper health care services (2). A cross-sectional study conducted in 2024 revealed that re-evaluations of children for development, dietary deficiencies, syndromes, systematic illness, and hormonal imbalances should be easily achievable by annual physical exams, although the specific cause of short stature, systematic diseases, syndromes, and hormonal disorders was not well articulated within the article (3). These findings are similar to those reported in recent studies from Bangladesh that have indicated the South Asian trend of etiologies, although it adds a special mention of malnutrition and rickets common in nutrition-deficit populations of Asia and where major deficiencies stem from poor diet (4).

In other studies, populations variously attributed to pathological and syndromic causes, including Turner syndrome and Noonan syndrome, and chronic disease, including chronic kidney disease and celiac disease, among the various causes of AES among these groups of patients (5). The initial assessment may involve a review of past history and general physical examination that may indicate grounds for discriminating primary growth disorders from those of systemic aetiologies (6). Consequently, it is highly relevant in helping detect the root cause early enough to inform treatment approaches since it directly influences the outcome. Early intervention in children with growth hormone deficiency in etiology and treatment showed improvement in growth results, emphasizing the importance of diagnosis and treatment intentions. Research from rural and low-resource environments sheds more light on how public health interventions aid in diagnosing and addressing short stature.

Another study carried out in an extensive study at a rural tertiary center showed that there is a delay in diagnosis and malnutrition, which are some causes that have not ceased to be the causes of growth failure in children. Constitutional delay and small family height, especially in states like Rajasthan, due to low income and poor health facilities, were reported more with nutritional and endocrine factors (9). The same picture was found in the study of another developing country, Bangladesh, where endocrine disorders such as hypothyroidism, nutritional rickets, and chronic system diseases in tertiary care centers were common, supporting the relation to healthcare availability and nutrition (10). Furthermore, recent work in molecular genetics on growth failure has pointed to several genetic disorders that may not otherwise be apparent clinically without assays.

In a study conducted on patients from north India, authors provided various cases that were previously diagnosable only via MGA, including genetic clinics and short stature of an unspecified etiology (11). However, there are other diseases, such as precocious puberty, that, except for short stature in young children, affect normal bone growth by closing off the epiphyseal plates and thus result in reduced height if not treated early (12). Another prevalent but less realized factor is age-related celiac disease and, especially, children presenting entirely different complaints from those of a gastrointestinal nature. In a study conducted in Peshawar, several children with growth failure were found to have celiac disease, and this has established the fact that there is a need to screen children in general for the disease if they present symptoms that cannot be explained (13).

Vitamin D deficiency leading to rickets is one of the major causes of dwarfism in the state of Eastern India. Many children who enrolled for examination had clinical and biochemical manifestations of rickets, although they hailed from sunny areas (14). A number of definitions characterize idiopathic short stature as a diagnosis that can only be made after one has excluded all other documented causal factors to short stature. A study in Lahore conducted in the pediatric endocrine clinics identified 36%

of children having ISS, and this perhaps justified the dearth of facilities for diagnosing or any other unidentified reasons (15). Altogether, these papers demonstrate how short stature in children can be caused by multiple factors and why the evaluation should follow a step-by-step fashion and be performed by a team of specialists. In the case of children who are failing to grow as expected, early referral, specific investigations, and management are likely to have a positive impact on the growth and quality of life of such children.

Objective: To determine the etiological profile of short stature in children presenting to a tertiary care hospital, aiming to identify the most common underlying causes and guide appropriate clinical management.

MATERIALS AND METHODS

Design: Descriptive Cross-sectional Study.

Study setting: This study was done at the Department of Diabetes and Endocrinology, Lady Reading Hospital, Peshawar.

Duration: The study was carried out over a six-month period, from April, 2024 to September, 2024.

Inclusion Criteria

Children with a height below the third percentile or below two standard deviations in age and sex were recruited. Interviews were conducted with patients who came to the hospital's outpatient department and the in-patient pediatric section. The sample was comprised of children who had never had any form of treatment for their growth and only those who had parental consent to take part in the research. All the categories mentioned above included males and females. The other aspects captured regarding the candidate include age and socioeconomic status.

Exclusion Criteria

Consequently, children with acute or chronic diseases that could affect the current height assessment (like the current infection or severe dehydration) or with no prior medical records were excluded. Children with any previous diagnosis and treatment for endocrine or systemic diseases related to short stature were also excluded.

Methods

All the children falling under the inclusion criteria were attained successively from the pediatric outpatient and inpatient Clinic during the consecutive period of study. Other demographic characteristics captured include age, gender, socioeconomic status, and for the child their parents' height. These were the patient's birth history, nutritional history, developmental history, and any chronic illness history. Another assessment was done through anthropometric measurements, which consist of height, weight, BMI, and upper-to-lower segment ratio. Centile graphs and mid-parental height were utilized to assess the growth profiles of the children. Appropriate serological and biochemical investigations performed were CBC, TSH, serum calcium, phosphate, alkaline phosphatase, tissue transglutaminase antibodies, and celiac serology as and when required growth hormone level. Any necessary X-rays, for example, bone age determination and karyotyping in special situations like Turner syndrome, were performed. All data were recorded on a structured proforma, and data analysis was performed to determine the causes of short stature in the studied population.

RESULTS

A total of 120 children presenting with short stature were enrolled in the study. Among them, 68 (56.7%) were males and 52 (43.3%) were females, with a male-to-female ratio of 1.3:1. The mean age of the children was 10.2 ± 3.5 years. Most patients (60%) belonged to lower socioeconomic backgrounds, followed by 30% from middle income and 10% from higher income families. The distribution of etiologies among the participants showed that normal variants of growth were the most

common cause, followed by endocrine, nutritional, and systemic causes. The frequency and percentage of various etiologies are shown in Table 1.

Table 1: Etiological Distribution of Short Stature (n=120)

Etiology	Frequency	Percentage (%)
Constitutional Delay of Growth	28	23.3%
Familial Short Stature	20	16.7%
Growth Hormone Deficiency	18	15.0%
Hypothyroidism	12	10.0%
Nutritional Deficiency	16	13.3%
Chronic Systemic Diseases	10	8.3%
Syndromic Causes (e.g., Turner)	8	6.7%
Celiac Disease	5	4.2%
Idiopathic Short Stature	3	2.5%

Age-wise distribution of etiologies revealed that constitutional delay and familial short stature were more prevalent in older children (10–14 years), while endocrine and nutritional causes were more common in younger children. Table 2 shows the age distribution of major etiological categories.

Table 2: Age-wise Distribution of Major Etiologies

Age Group (Years)	Normal Variants	Endocrine	Nutritional	Others
2–5	5	4	6	3
6–9	12	10	7	6
10–14	25	16	3	9
15–18	6	5	0	2

Gender-wise distribution showed a slight male predominance across all etiologies, with growth hormone deficiency and constitutional delay more common in males, whereas hypothyroidism and syndromic causes showed a female preponderance. Table 3 provides the gender-wise breakdown.

Table 3: Gender-wise Distribution of Common Etiologies

Etiology	Males (n=68)	Females (n=52)
Constitutional Delay	20	8
Familial Short Stature	11	9
Growth Hormone Def.	12	6
Hypothyroidism	4	8
Nutritional Deficiency	9	7
Syndromic Causes	2	6

These findings highlight the diverse etiological spectrum of short stature and underscore the need for a structured diagnostic approach tailored to age and gender presentations.

DISCUSSION

This makes it present in any age and social class of children because there are several factors that lead to short height in kids. It is often stated as the outcome of functional disorders of the physiological, endocrine, nutritional, hereditary, and systemic body functions. This cross-sectional study was

conducted in a tertiary care teaching hospital in Islamabad, Pakistan. The aim of the study was to identify the causes of short height in children. These results are compatible with other studies that have been conducted with similar subjects in South Asia but also show differences depending on the context. In the present study, constitutional delay of growth and development (CDGD) had the highest percentage of normal variants, including constitutional delay of growth and development and familial short stature. Constitutional delay was seen in 23.3 percent of children, similar to the study by Rajput et al. This used the description from one of the sources claiming that constitutional delay of growth and development (CDGD) remains one of the most common causes of short stature without a known medical explanation.

Similarly, in the observational study conducted by Shah et al. from a rural tertiary center, CDGD, and FSS were identified as the most common causes of short stature, so the author found it important to categorically distinguish between normal short stature and other forms of reduced height in order to reduce on unnecessary investigations. The prevalence of GHD was found to be 15% in the children in the present study, which is similar to 14%, as reported by Jain and Gupta (3). gonadal evaluation is of importance in the attempts to screen short stature, particularly where height is substandard, and there is also retardation of bone age. Danda et al. (7) assessed recombinant growth hormone in this study, which showed benefits to children with GHD who are willing to seek early diagnosis.

Analysis of the results established that children in this study had a nutritional deficiency-related short stature of 13.3%. This shows the high prevalence of undernutrition in the region, even with the awareness of various health issues. Jasim et al. (4) corroborated these findings in a study carried out on children in Bangladesh, especially noting that poverty and food insecurity lead to stunting. Nutritional interventions and public health campaigns to curtail this preventable cause are still relevant in areas where there are scarce resources. This is a result of other endocrine disorders apart from GHD, especially hypothyroidism, which comprised 10% of the subjects. Baigh et al. (5) pointed out hypothyroidism among the root endocrine causes of short stature in children. It is essential for such patients to be diagnosed and treated at an early age while using hormonal therapies to avoid a severe rise in stature and intellect losses. Angadi et al. (6) also noted the importance of thyroid screening in children with unexplained growth retardation.

The specific associated syndromes, such as Turner syndrome among the cohort, were found in 6.7 percent. This is consistent with the findings made by Singh et al. (11), who investigated the genetic etiology of growth failure and concluded that Turner syndrome is one of the most prevalent genetic disorders among girls with such clinical presentation. Karyotyping should be performed in all females with unexplained short stature since early diagnosis can also initiate the treatment by administering growth hormones and monitoring for comorbid conditions. Chronic systemic disease accounted for 8.3% of cases in the study. These were patients with kidney disease, congenital heart ailments, and anemia that was persistent in nature. Similar observations were made by Shah et al. (8), who wrote extensively on the effects of chronic illness on linear growth. According to Singh et al. (9), such systemic diseases are left out in the early evaluation of children and should be among differential diagnoses of growth failure in children who complain of fatigue, poor appetite, or frequent infections. Celiac disease prevalence has been estimated in children at 4.2 percent. This figure is in concordance with the study by Muhammad et al. (13), who conducted a study in Peshawar and found that growth failure may be the only presenting feature of celiac disease in children. Since symptoms may not necessarily appear at an early progress of the disorder, screening with anti-tTG antibodies should be carried out in children with growth delay. Notably, primary growth failure or idiopathic short stature (ISS) was identified in just 2.5% of subjects enrolled in the study. This may be due to enhanced diagnostic accuracy, where more cases are accurately identifiable under certain causes. Nonetheless, as pointed out by Usman et al. (15), In the present study, ISS continues to be as prevalent in many pediatric Endocrine clinics in Pakistan, which may partly be attributed to the restricted availability of sophisticated diagnostic facilities in some centers.

Although rickets was not amenable as a primary diagnosis in this group of patients, its role in the differential of short stature in such children cannot be overemphasized. Bhattacharjee et al. (14) explored the etiology of rickets in tertiary hospitals and pointed out that rickets are still common in

Eastern India. Clinical suspicion and biochemical, molecular, and imaging biomarkers still hold a pivotal role in diagnosis and management. Males were also found to be more affected in this study than the female population. Females had a comparatively higher prevalence of hypothyroidism and syndromic disorders such as Turner syndrome. These findings are in line with studies by Shah et al. (12) and Singh et al. (11), who acknowledge sexual dimorphisms in homosexual maturation and inheritance of short stature. This underlines the necessity of focusing on sex-related clinical differences during assessment.

Age-specific patterns also emerged as diagnostic clues. Normal variants were recently seen to be more prevalent in the adolescent group, while nutritional and endocrine causes were seen more in the young children group. Biswas et al. (10) gathered similar results as Shah et al. (8), which supported the use of age-related screening and/ or clinical decision-making. This aligns with regional and international reports indicating that short stature in children is not a disease with a single cause. The first-time and right mode of evaluation is usually critical to ascertain the most definitive diagnosis and subsequently manage the illness. Clinical, hormonal, nutritional, radiological, and genetic models must be implemented in a structured format to aid different diagnostics and therapies in performing better. However, the avenues for general practitioners and parents should be provided regularly so that such delays are not occasioned.

CONCLUSION

The purpose of this research is to identify the various causes of short stature in children who are attending a tertiary-care-teaching hospital in Islamabad. The factors mentioned by the respondents as the causes of short stature included constitutional delay of growth and development, familial short stature, and growth hormone deficiency. This includes diet deficiencies, hypothyroidism, systemic diseases, syndromes, and coeliac disease were also other causes. For given patients, isolated and combined etiologies have gender and age dependency, which indicates the need to focus on a further detailed assessment of the patients. A thorough history and clinical examination form a sound base for diagnosis and the institution of an appropriate management plan that is a teamwork approach because it involves other specialties. Since most of these cases are preventable or at least easily manageable, growth can be enhanced if such causes as endocrine and nutritional imbalances are detected in the early stages. Therefore, the study features the enhancement of practitioners' and parents' knowledge of early detection of children's growth issues. Therefore, future research to extend the presented results and to build the diagnostic clinical algorithm should be based on multi-center studies.

REFERENCES

1- Rajput, R., Rani, M., Rajput, M. and Garg, R., 2021. Etiological profile of short stature in children and adolescents. Indian Journal of Endocrinology and Metabolism, 25(3), pp.247-251.
2- Shah, D., Pathak, S., Rathore, A.S., Modi, P., Sheth, D. and Shah, M., 2023. An analysis of demographic and etiological factors of children and adolescents with short stature in the rural tertiary center-An observational study.
3- Jain, S.C. and kumar Gupta, A., 2024. ETIOLOGICAL PROFILE OF SHORT STATURE IN CHILDREN: A CROSS-SECTIONAL STUDY. Int J Acad Med Pharm, 6(1), pp.2169-2172.
4- Jasim, S., Mohsin, F., Islam, N., Halder, A.L. and Mollah, M.A.H., 2022. Etiology of short stature in children and adolescents: experience at a tertiary care hospital in Bangladesh. BIRDEM Medical Journal, 12(2), pp.117-123.
5- Baigh, Z.H., Sudhakar, D., Fayaz, I. and Ali, I., 2024. Pathological and Syndromic Short Stature in Children: Clinical and Etiological Profile. Res. J. Med. Sci, 18, pp.71-76.
6- Angadi, K., Ahmed, M.M., Bhimalli, S.S. and Patil, S., 2021. A study of clinical profile of children with short stature. Indian Journal of Child Health, 8(9), pp.328-330.
7- Danda, V.S.R., Verpula, M., Paidipally, S.R., Thaduri, K.R., Lodha, P. and Devireddy, S.R., 2022. A prospective study of etiology and clinical characteristics of children with short stature

and response to treatment. Indian Journal of Health Sciences and Biomedical Research kleu, 15(2), pp.137-142.

8- Shah, D., Pathak, S., Muley, P., Modi, P., Ahmed, N.M. and Bhatt, V., 2021. A study of short stature among adolescents in the rural tertiary center-a prospective observational study.

9- Singh, A., Masand, R., Verma, C.R. and Khinchi, Y., 2021. Etiological profile of short stature in rural Rajasthan. Indian Journal of Child Health, 8(8), pp.297-301.

10- Biswas, R., Farzana, N. and Rahman, S.N., 2022. Clinicoepidemiological Profile of Short Stature at A Tertiary Care Centre in Bangladesh. Dhaka Shishu (Children) Hospital Journal, 38(2), pp.79-83.

11- Singh, K., Puri, R.D., Bijarnia-Mahay, S., Lall, M., Verma, J., Saxena, R., Kohli, S., Thomas, D., Saviour, P. and Verma, I.C., 2022. Clinical and genetic profile of children with short stature presenting to a genetic clinic in Northern India. Indian Pediatrics, 59(6), pp.463-466.

12- Aftab, S., Manzoor, J., Mahmood, Q. and Shaheen, T., 2022. Precocious puberty: The clinical profile and the etiological classification of children presented at a tertiary care children's hospital. Pakistan Journal of Medical Sciences, 38(4Part-II), p.955.

13- Muhammad II, A., Arif, N., Wajid, K.K., Rehman, K., Sardar, N., Khan, P., Hussain, U. and kamran Wajid, K., 2022. Short stature and celiac disease in children (5 to 16 years) presenting at a tertiary care hospital in peshawar. Cureus, 14(6).

14- Bhattacharjee, R., Chakraborty, P.P., Agrawal, N., Roy, A., Maiti, A. and Chowdhury, S., 2024. Etiology and biochemical profile of rickets in tertiary care centres in Eastern India: a retrospective cross-sectional study. Indian Journal of Endocrinology and Metabolism, 28(2), pp.184-191.

15- Usman, M., Aftab, S., Chaudhry, A.R., Naseer, A., Gul, R. and Butt, T.A., 2024. FREQUENCY OF IDIOPATHIC SHORT STATURE AT PEDIATRIC ENDOCRINE CLINIC IN A TERTIARY CARE HOSPITAL AT LAHORE. Pakistan Postgraduate Medical Journal, 35(03), pp.96-100.