Section: Paediatrics

Original Research Article

ETIOLOGICAL AND CLINICAL PROFILE OF PATIENTS PRESENTING AS SHORT STATURE IN A TERTIARY CARE CENTER IN NORTH INDIA

Kajal[1], Amritesh Ranjan[2] Venkatesh Mishra[3], Manish Agrawal[4], Ankit Kashyap[5]

[1]*Junior Resident, Department of Pathology, Muzaffarnagar Medical College, India.*
[2]*Associate Professor, Department of Pediatrics, Muzaffarnagar Medical College, India.*
[3]*Senior Resident, Department of Pediatrics, Muzaffarnagar Medical College, India.*
[4]*Professor and Head, Department of Pediatrics, Muzaffarnagar Medical College, India.*
[5]*Junior Resident, Department of Pediatrics, Muzaffarnagar Medical College, India.*

Received *: 05/07/2025*
Received in revised form *: 23/08/2025*
Accepted *: 13/09/2025*

Corresponding Author:
Dr. Venkatesh Mishra,
Senior Resident, Department of Pediatrics, Muzaffarnagar Medical College, India.
Email: drvenkateshmishra@gmail.com

DOI: 10.70034/ijmedph.2025.3.592

Source of Support: Nil,
Conflict of Interest: None declared

Int J Med Pub Health
2025; 15 (3); 3234-3238

ABSTRACT

Background: Short stature is a frequent presentation in pediatric outpatient departments and is known to have multiple etiologies which include nutritional deficiencies, chronic systemic illnesses, endocrine disorders, and genetic conditions. This study aims to evaluate the clinical and etiological profile of patients presenting with short stature at a tertiary care center in North India.

Materials and Methods: A cross-sectional study was conducted over one year (January 2024 – December 2024) at MMCH, involving children aged 2–12 years who attended the outpatient and inpatient departments. Anthropometry was done and plotted on appropriate growth charts to identify cases of short stature. A detailed history, clinical examination, and appropriate investigations were conducted to determine the etiological factor for the presentation. The etiological factors were categorized and analyzed.

Results: A total of 2500 pediatric patients were screened, out of which 236 (9.4%) were diagnosed with short stature. After applying exclusion criteria, 124 children were included in the study. Among them, 88% (110 cases) had proportionate short stature, and 12% (14 cases) had disproportionate short stature. The male-to-female ratio was 1.58:1. Nutritional deficiencies were the most common cause (29.8%), followed by chronic systemic illnesses (25.8%, primarily Hemoglobinopathies), idiopathic short stature (16.9%), endocrine disorders (11.2%, mainly hypothyroidism), genetic conditions (8.0%), and skeletal dysplasias (8.0%).

Conclusion: Nutritional deficiency is seen as most common cause of short stature in this population, prompting requirement of early intervention through community-based nutritional support and awareness campaigns. The high prevalence of Hemoglobinopathies and celiac disease suggests the need for increased screening in short children. Endocrine causes, although less frequent, require universal thyroid screening in primary healthcare settings for early detection and treatment. Addressing these factors can significantly reduce the incidence of short stature and improve overall child growth outcomes in rural India.

Keywords: Short stature, Growth disorders, Nutritional deficiencies, Hemoglobinopathies, Celiac disease, Endocrine disorders, Hypothyroidism, Genetic conditions, Skeletal dysplasias, Idiopathic short stature, Pediatric growth, North India.

INTRODUCTION

The process of growth is complex, shaped by an individual's genetic makeup, hormones, adequate nutrition, chronic illnesses, exercise and sleep.[1] One way to assess growth is through consistent height measurements. Being shorter than average may be the initial indication of underlying pathological conditions. Identifying short stature early enables early intervention, which is very crucial for achievement of normal adult height. [2]

Short stature is defined by an individual's height less than 3rd percentile or more than two standard deviations beneath the average height for their specific age and gender.[3] In any given population, about 3% of children will present with short stature; and of these, almost 50% are due to physiological causes and rest 50% due to pathological causes.[4] Physiological causes are categorized as familial short stature and a constitutional delay in growth and puberty. These are normal growth variations and do not require any intervention.[5] Pathological causes include malnutrition, chronic disorders (eg-chronic kidney disease, anemia), genetic and endocrine causes.[6] If a child exhibits short stature without an identifiable specific cause, they may be diagnosed with idiopathic short stature.[7]

Prompt diagnosis and management of underlying etiology can help to have a typical growth pattern and avert psychological and other difficulties linked to pathological causes of short stature.[10] The range of causes and their frequency will differ between community-level hospitals and tertiary care centers. A considerable number of children in rural areas and lower socioeconomic groups may remain undiagnosed.[11] This is because they are less likely to seek healthcare visit for the same.[10] This current research intends to screen patients visiting OPD in a tertiary medical college and investigate their etiological profiles.

MATERIALS AND METHODS

Patients visiting OPD and IPD of MMCH from January 2024 to December 2024 between age group of 2-12 years were enrolled in study after taking informed consent. Short stature was identified after taking anthropometry and plotting them on specified growth charts. Along with this parental height was taken to identify physiological causes using mid parental height.

Study type- Cross sectional study

Inclusion Criteria: All patients visiting OPD and IPD in given time and giving consent for enrollment in study

Exclusion Criteria: Refusal of consent or further investigations

Outcome measures: Prevalence of short stature as well as their clinical and underlying etiological factors evaluated.

RESULTS

The study was done in period of 1 year from Jan 1,2024 to Dec 31, 2024. 2500 patients presented in OPD and IPD, who were screened for short stature. Out of a total of 2500 patients, 236 patients were diagnosed with short stature. This represents 9.4% of the total population. Among 236 patients diagnosed with short stature, 112 were excluded as per exclusion criteria. 124 children were recruited for study. 88% (110 cases) had proportionate short stature and only 12% (14 cases) were found to have disproportionate short stature. Out of 124 cases 70(56.4%) were males and 54(43.5%) were female. Gender was not statistically significant. Table 1 summarizes auxological parameters of study participants showing weight and BMI being statistically significant findings. Table 2 and 3 shows age and sex wise distribution of the population. Participants had an average height of 107.28+/-13.34 cm showing significant p value(p=0.04), while BMI had mean of 17.44 also showing significant p value(p=0.03). Other parameters like age, weight, mid-parental height etc. were measured but without statistically significant differences in most cases.

Table 1: Baseline demographics

Parameter	Mean (SD)	Median (IQR)	Min-max	p-value
Chronological age	07.82+/-2.32	07.58	3.9-11.8	0.06
Height	107.28+/-13.34	110	85.7-1435	0.04
Weight	15.75+/-5.38	16.5	10.1-35	0.09
BMI	17.44+/-1.98	17.3	15.3-25.2	0.03
Birth weight	2.14+/-0.54	2.1	1.4-3.2	0.11
Height age	6.45+/-2.58	6.5	2.7-10.8	0.09
Bone age	7.68+/-1.73	9.6	4-11.5	0.13
Mid-parental height	152.88+/-5.36	150	134.8-157.6	0.07

Table 2: Age wise distribution in study

Age Group (years)	No. of cases	% of cases
2 – 5	35	28.2
6 – 9	60	48.4
10 – 13	29	23.4

Total	**124**	**100.0**

Table 3: Sex distribution in study

Sex	No. of cases	% of cases
Male	70	56.4
Female	54	43.5
Total	**124**	**100.0**

Table 4: Etiological distribution of cases

	Number	Total	Percentage
CSI	32	124	25.8
ENDOCRINE	14	124	11.29
GENETIC	10	124	8.06
NUTRITIONAL	37	124	29.83
Idiopathic	21	124	16.93
SKELETAL	10	124	8.064
	124		

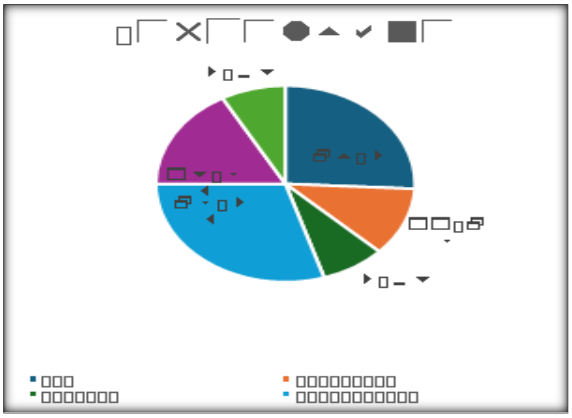

Figure 1: Etiological distribution of cases

Figure 1 and table 4 categorize causes of the condition with majority being Nutritional (29.8%) followed by chronic systemic illness (25.8%) and idiopathic (16.9%). Incidence of endocrine causes were (11.2%), majority of them being hypothyroidism. Genetic causes and skeletal dysplasia were least common.

Among 37 patients presenting with short stature due to nutritional causes 24 patients (64.8%) presented with malnutrition, constituting the majority. 7 patients (18%) presented with rickets, and 3 patients presented with severe anemia. There was no statistically significant gender difference.

In the patient group presenting chronic systemic disease majority were due to Hemoglobinopathies (n=14,43.7%), celiac disease (n=10,31.2%) followed by 3 cases of chronic kidney disease (9.3%), severe asthma (n=2,6.25%) and 1 case with chronic heart disease (3.1%).

Out of total 14 patients presenting with endocrine causes,10 was due to hypothyroidism, and 4 due to growth hormone deficiency.

DISCUSSION

Short stature is a common condition seen in pediatric practice. Its prevalence and etiological spectrum can vary widely depending on geographic, socioeconomic, and nutritional factors. This study aimed to evaluate the prevalence and etiological profile of short stature among children presenting to a tertiary care center in North India, and to compare these findings with previously published studies.

Prevalence of Short Stature

In our study, the prevalence of short stature was found to be 9.4%, which is higher than several earlier Indian studies but lower than some others. For instance, Garg P et al,[13] reported a prevalence of 13.8%, while Velayudhan K et al,[14] and Khadgawat R et al,[15] found lower rates of 2.86% and 7%, respectively. Studies by Colaco P et al. and De Mel T et al. reported prevalences of 5.6% and 5.3% and study by Dubey B et al,[9] reported prevalence of 4.2%. The variability in prevalence rates across studies may reflect differences in the study populations, referral patterns, diagnostic criteria, and regional factors such as nutrition and access to healthcare. The relatively higher prevalence observed in our study could be attributed to the tertiary care setting, where more complex and referred cases are likely to be seen.

Table 5: Comparison of prevalence with other studies

Study name	Prevalence
Present study	9.40%
Garg P et al.	13.80%
Velayutham K et al.	2.86%
Khadgawat R et al.	7%
Colaco P et al.	5.60%
De Mel T et al.	5.30%
Dubey B et al	4.7%

Etiological Spectrum

The etiological analysis in our cohort revealed that the most common cause of short stature was chronic malnutrition with iron deficiency anemia (IDA), accounting for 29.83% of cases. This finding underscores the ongoing burden of nutritional deficiencies in the region, despite advances in public health and nutrition programs. Chronic malnutrition as a leading cause is consistent with the findings of Singh A et al,[16] who reported a similar prevalence (29.25%) for this etiology, highlighting that nutritional factors continue to play a significant role in the pathogenesis of short stature in Indian children. Constitutional delay of growth and puberty was the second most common cause in our study (25.8%), followed by idiopathic short stature (16.93%), hypothyroidism and other endocrine diseases (11.29%), familial short stature (8.06%), and chronic systemic diseases such as uncontrolled asthma (4%). These findings are in line with previous studies, though the proportions vary. For example, Sadiq A et al,[17] reported a much higher prevalence of familial short stature (36%) and a lower prevalence of constitutional delay (10%) and Dubey B et al reported idiopathic short stature as most common cause(29.8%),followed by chronic systemic illness(25.8%) and endocrine causes(11.3%) The differences may be due to genetic, environmental, and methodological factors, including the criteria used for categorizing etiologies.

Comparison with Previous Studies

Comparative analysis with other studies reveals both similarities and differences in etiological patterns. While chronic malnutrition remains a significant cause across studies, its proportion varies widely. The higher prevalence of familial short stature in Sadiq A et al. (36%) compared to our study (8.06%) may reflect differences in genetic backgrounds or family history ascertainment. Similarly, the prevalence of idiopathic short stature was higher in our study (16.93%) compared to Singh A et al. (8%) and Essaddam L et al. (8%),[18] which may be due to differences in the thoroughness of diagnostic workup or the inclusion criteria for idiopathic cases.

Endocrine causes, particularly hypothyroidism, accounted for 11.29% of cases in our study, which is higher than the 4–4.75% reported in other studies. This could be due to increased awareness and improved diagnostic facilities for thyroid disorders in our center. Chronic systemic diseases as a cause of short stature were less common, but their presence highlights the need for a comprehensive clinical evaluation in all children presenting with growth concerns.

Implications and Recommendations

The findings of this study have important clinical and public health implications. The high prevalence of nutritional causes indicates the need for ongoing efforts to improve child nutrition and address macro and micronutrient deficiencies. At the same time, the significant proportion of cases due to constitutional and familial factors emphasizes the importance of detailed history-taking and growth monitoring to differentiate between pathological and non-pathological short stature.

Furthermore, the identification of endocrine and systemic causes underscores the necessity of a systematic approach to the evaluation of short stature, including appropriate laboratory investigations and specialist referrals when indicated. Early identification and intervention can prevent long-term sequelae and improve quality of life for affected children.

CONCLUSION

In summary, this study highlights that short stature remains a prevalent clinical problem in North India, with a multifactorial etiology dominated by nutritional, constitutional, and idiopathic causes. The etiological spectrum observed is broadly consistent with previous Indian and international studies, though regional variations persist. Nutritional deficiency was identified as the most prevalent cause of short stature in this population, highlighting the need for early intervention through awareness programs, improved dietary practices, and community-based nutritional support. The high prevalence of Hemoglobinopathies and celiac disease suggests the need for increased screening in short children. Endocrine causes, although less frequent, require universal thyroid screening in primary healthcare settings for early detection and treatment. Addressing these factors can significantly reduce the incidence of short stature and improve overall child growth outcomes in rural India. Addressing nutritional deficiencies, raising awareness about normal growth patterns, and ensuring timely referral and evaluation are key strategies to improve outcomes for children with short stature in this setting.

REFERENCES

1. Kamboj M. Short stature and growth hormone. IJP. 2005;72(2):149-57.
2. Haymond M, Kappelgaard AM, Czernichow P, Biller BM, Takano K, Kiess W.Early recognition of growth abnormalities permitting early intervention. Acta Paediatrica. 2013;102(8):787-96.
3. Cause of short stature in our population. J Ayub Med Coll Abbottabad. 2016;28(1):135-40.
4. Nath P, Kumar J, Rahaman SK, Rai M. Short stature: Evaluation and management [of short stature [Internet]. 2008 [cited 15 June 2017]. Available from: http://www.apiindia.org/medicine_update_2013/chap137.pdf .
5. Sultan M, Afzal M, Qureshi SM, Aziz S, Lutfullah M, Khan SA, et al. Etiology of short stature in children. J Coll Physicians Surg Pak. 2008;18(8):493-97.
6. Ray J. Childhood short stature. Al Ameen Journal of Medical Sciences. 2012;1(5):212-15.
7. Wit JM, Reiter EO, Ross JL, Saenger PH, Savage MO, Rogol AD, et al. Idiopathic short stature: Management and growth hormone treatment. Growth Horm. 2008;18(2):111-35.
8. Ghai OP, Paul VK, Bagga A. Ghai Essential Pediatrics. 10th ed. New Delhi: CBS Publishers & Distributors; 2023. p. 17.

9. Dubey B, Raipurkar S, Doharey N C, Neha, Sagar U, Evaluation of short stature in paediatric age group in rural medical college. IP Int J Med Paediatr Oncol 2018;4(4):161-166
10. Mohammadian S, Khoddam H. An etiologic evaluation of children with short stature in Gorgan (Northeast Iran), 2005. J Med Sci. 2007;7(7):1206-09.
11. Garg P. Short stature in Indian children: Experience from a community level hospital. Srilanka J Child Health. 2004;34:84-88.
12. Velayutham K, Selvan SS, Jeyabalaji RV, Balaji S. Prevalence and etiological profile of short stature among school children in a South Indian population. Indian J Endocrinol Metab. 2017;21(6):820.
13. Garg P. Short stature in Indian children: Experience from a community level hospital. Srilanka J Child Health. 2004;34:84-88.
14. Velayutham K, Selvan SS, Jeyabalaji RV, Balaji S. Prevalence and etiological profile of short stature among school children in a South Indian population. Indian J Endocrinol Metab. 2017;21(6):820.
15. Khadgawat R, Dabadghao P, Mehrotra RN, Bhatia V. Growth charts suitable for evaluation of Indian children. Indian Pediatr.1998;35:859-65.
16. Singh A, Masand R, Verma CR, Khinchi Y. Etiological profile of short stature in rural Rajasthan. IJCH. 2021:297-301.
17. Sadiq AM, Hassan QF, Mutlag YA. The prevalence and etiological profile of short stature in children. Eurasian J Med Oncol. 2022;6:17-23.
18. Essaddam L, Kallali W, Cherifi E, Guedri R, Mattoussi N, Fitouri Z, et al. Characteristics and etiologies of short stature in children: Experience of an endocrine clinic in a Tunisian tertiary care hospital. Int J PediatrAdolesc Med. 2020;7(2):74-77.