ORIGINAL RESEARCH PAPER

Endocrinology

KEY WORDS: Growth, Height, Short

CLINICAL AND ETIOLOGICAL PROFILE OF CHILDREN WITH PATHOLOGICAL AND SYNDROMIC SHORT STATURE

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ABSTRACT

Background: Short stature is one of the common problem in child population with diverse aetiologies, half of them being physiological and another half due to pathological causes. The objective of this study was to study the clinical profile of the children with pathological short stature and to study the various causes for pathological stature including syndromic short stature.

Methods: Retrospective study was conducted in the Department of medicine at district hospital JLNM Srinagar and DH baramulla, where the data of Children who met the inclusion criteria were collected from the case records of 2 years period from Jan 2018 to june 2019. Data collected was tabulated and analyzed using appropriate statistical methods.

Results: Commonest age group affected was < 5 years about 40%, 38% were 5 -10 years and 22 % were > 10 years. 41.6 % were males and 58.3 % were females. Commonest causes were Hypothyroidism and genetic disorders, followed by nutritional and musculoskeletal. 54.1% were proportionate and 45.9 % were disproportionate.

Conclusions: Endocrinological and genetic syndromes are most common etiologies for pathological short stature. So detailed evaluation of short stature is very important as some of the pathological causes are treatable when diagnosed at an early stage.

INTRODUCTION:

Normal growth is considered to be an indirect indicator of overall wellbeing of the child, any alteration or disturbance in the linear growth is found to be one of the most common concerns for the family. A child is considered to be short, when his or her height is below the third centile or < 2 standard deviation on the height chart used for specific population. Approximately 3% children in any population are found to be short, amongst which half of them will be physiological (familial or constitutional) and half will be pathological.1

Short stature is not a diagnosis but a presenting symptom, sometimes the first clinical manifestation of an underlying pathological disease, like endocrinological disease eg. Growth hormone deficiency (GHD), chronic diseases eg. Chronic renal failure, disesases of bone eg. Skeletal dysplasia or clinically defined syndromes eg. Downs and Turners; thus, short stature has to be evaluated precisely.2

Short stature is a result of various diverse etiologies, which are categorized in two groups broadly as pathological causes and normal variants. Normal variant short stature includes familial short stature (FSS) and constitutional growth delay (CGD), while there are broad differential of conditions associated with pathological short stature that includes endocrine diseases, clinically defined syndromes, chronic diseases, metabolic diseases and others. 3, 4

Although after detailed evaluation, most children with short stature are assessed as being healthy careful monitoring of growth status (e.g., measurement of height, weight, and growth velocity) over time is essential for child healthcare. Most of the causes can be excluded with a thorough history, physical examination, and basic screening tests and managed accordingly as per the cause and parental counseling. Normal variation in adult height is largely due to inherited, genetic factors. Within a population, typically 80% or more of the variation in height is explained by genetic factors , although it is clear that environmental factors contribute to differences between populations and to recent increases in average height across generations.The genetic contribution to height is largely attributable to the combined effects of many different genes, meaning that height is typically a polygenic trait. At least half of the variability in adult height in a population is due to the combined effect of common genetic variants.

So, this study was undertaken to know the clinical and etiological profile of children who presented with pathological short stature as 50% of the cases are pathological, so that we can suspect and identify the pathological causes for short stature as early as possible and manage the condition appropriately as per the cause and counsel the parents regarding the same

METHODS:

A retrospective study was done. Case records of children who were admitted in these two hospitals and diagnosed as Pathological short stature.

Inclusion criteria

All Children diagnosed as short stature i.e height below 3rd centile or less than two standard deviations (SDs) below the median height for that age and sex according to the population standard, IAP growth charts were used and due to some pathological disorder being diagnosed using various investigations like, complete hemogram with erythrocyte sedimentation rate, bone age, urinalysis, stool examination, renal function test, calcium, phosphate, alkaline phosphatase, venous gas, fasting sugar, liver function tests, hormone assays like Insulin like growth factor assay/growth hormone stimulation tests, thyroid function test, karyotyping, mutation analysis(wherever possible) and neuroimaging as per the presentation and cause during 2018 January to 2019 June.

METHODOLOGY:

Retrospective study was conducted in the Department of Medicine at JLNM district hospital Srinagar and DH Baramulla where the data of children who met the inclusion criteria were collected from the case records of 1.5 years period from Jan 2018 to june 2019. Data obtained was arranged according to clinical profile and etiology and expressed as number and percentage and were analysed using the SPSS version 22

software. Institutional ethical committee clearance was taken.

RESULTS:

Records of 24 children who were diagnosed as pathological short stature showed that the commonest age group being affected was < 5 years about 41.6%, then 33.3% were in the age group of 5-10 years and 25% were > 10 years (Table 1).

Among the 24 children 41.6 % were males and 58.3% were females (Table 2). Analysis of various etiologies (Table 3) for the pathological short stature showed that the commonest causes were endocrinological like hypothyroidism and genetic disorders including various syndromes like Downs, Turners which constituted about 24% each, followed by nutritional causes and musculoskeletal causes like skeletal dysplasia, vertebral defect, were about 20% each and the remaining were chronic diseases like Chronic renal failure, cyanotic congenital heart disease, caries spine and based on body proportions ,clinical profile showed 54.1% were in the Proportionate group and 44.9 % were in the Disproportionate group (Table 4).

And short children with presence of clinical features like skeletal abnormalities were 31% ,with dysmorphism were 24%, with coarse, dry skin and goitre were 20%, with features of micronutrient deficiency like pallor, dermatoses, chelitis were 13% and with neonatal hypoglycaemia 3% ,with hypertension 3% ,with murmur 3% (Table 5). Among the males, nutritional, endocrinological and chronic diseases were the common etiologies and among the females skeletal causes were common though not statistically significant. Although after detailed evaluation, most children with short stature are assessed as being healthy careful monitoring of growth status (e.g., measurement of height, weight, and growth velocity) over time is essential for child healthcare. Most of the causes can be excluded with a thorough history, physical examination, and basic screening tests and managed accordingly as per the cause and parental counseling.

So, this study was undertaken to know the clinical and etiological profile of children who presented with pathol ogical short stature as 50% of the cases are pathological, so that we can suspect and identify the pathological causes for short stature as early as possible and manage the

Table 1: Age wise distribution (years)	No (%)
1-5	10 (41.6%)
6-10	8 (33.3%)
11-15	06 (25%)
Total	24 (100%)

Table 2:Gender distribution	No (%)
Male	10 (41.6 %)
Female	14 (58.3 %)

Table 3: Etiology of pathologial short stature.

Cause	Male	Female	Total
Rickets/hypophosphatemic	1	1	2
RTA/Amelogenesis imperfect	1	1	2
Osteogenesis imperfecta	1	-	1
MAS	1	-	1
CHF	1	-	1
CRF	-	1	1
Chronic diseases/T.B/Diabetes	-	1	1
Hypothyroidism	2	3	5
GHD		1	1
Downs		1	1
Turners		1	1
PradderWilli		1	1
Seckel		1	1
Rubinstein Taybi syndrome	1		1
Ellis van crevald syndrome	1		1
Skeletal dysplasia	1	2	3

Table 4: Proportionate versus disproportionate

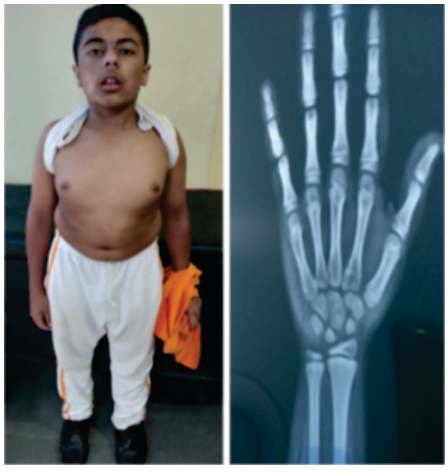
Proportionate: no (%)	Disproportionate: no (%)
13 (54.1)	11 (45.8)
PEM: 4 Endocrine/Genetic causes 7 CRF: 1 CHD: 1	Hypothyroidism: 5 Skeletal dysplasia: 1 Rickets: 4 Osteogenesis imperfect 1

Table 5: Table 5: Clinical profile of children with pathol ogical short stature.

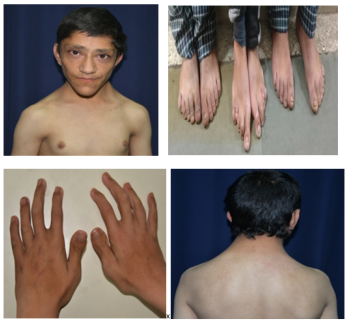
Clinical features	Etiology	Frequency of cases
With dysmorphism	Genetic syndromes	7
With skeletal abnormalities	Skeletal dysplasia/ mucopolysaccharid osis/ caries spine/rickets	1
With dry skin/goitre	Hypothyroidism	5
With hypertension	Chronic renal failure	01
With murmur	Congenital heart disease	01
With pallor/ platynychia /dermatosis/chelitis/stom atis/phrynoderma	Protein energy malnutrition	04
With neonatal hypoglycemia/micropenis	Growth hormone defeciency	01

Table 6: Mean of chronological age and HT age for 2 most common causes.

Mean HT age		Mean chronological age		
Chromosomal disorders	3.1		6.5 Years	
Hypothyroidism		5.8 Years		10 Years



Specific short syndromic cases a: spondyloepiphyseal dysplasia b:Ellis van crevald syndrome with polydactyly, ectodermal dysplasia and enamel hypoplasia c:Rabson mendalhal syndrome with enamel hypoplasia and acanthosis with severe insulin resistance.



Rubinstein-Taybi syndrome : Coarse facial features, asymmetric shoulders, broad distal phalanx.

DISCUSSION:

In our study, the evaluation of records of 29 Children who were diagnosed as pathological short stature showed that the commonest age group being affected was < 5 years about 48%, then 34% were in the age group of 5-10 years and 17% were > 10 years. Similarly Pankaj garg in his study identified 2-5 years was most commonly affected. In this study, we included only those with pathological short stature, and majority of them in our study were due to endocrinological and genetic causes that presents early in life and detected early so < 5 years were more but in other two studies they included both physiological and pathological short staure. Among the 29 children 48% were males and 52% were females, this was similar to Siamak Shiva study where as various other studies showed males were commonly affected.9 But again most of the studies included both physiological and pathological short stature.5

Analysis of various etiologies for the pathological short stature showed that the commonest causes were endocrin ological like hypothyroidism and genetic disorders including various syndromes like Downs, Seckel, Turners which constituted about 24% each, followed by nutritional causes and musculoskeletal causes like skeletal dysplasia, vertebral defect, MPS were about 20% each and the remaining were chronic diseases like chronic renal failure, cyanotic congenital heart disease, caries spine and based on body proportions, 58% were in the proportionate group and 42% were in the disproportionate group. Among the males, nutritional, endocrinological and chronic diseases were the common etiologies and among the females skeletal causes were common though not statistically significant.

CONCLUSION:

The findings of the present study help us in developing an insight into, age-sex distribution, clinical profile (proportionate versus disproportionate), (with dysmorphic features, skeletal abnormalities, etc.) and aetiological profile of children presenting with pathological short stature.

REFERENCES:

1. Short stature: evaluation and management. Available at www. apiindia. org/ medicine update. Accessed on 12 July 2016.
2. Khadilkar VV, Khadilkar AV. Practical approach to short stature. IJPP. 2011;13(4)396-401.
3. Ray J. Childhood short stature. Al Ameen J Med Sci. 2012;5(3):212-5.
4. Song KC, Jin SL, Kwon AR, Chae HW, Ahn JM, Kim DH. Etiologies of short stature. Ann Pediatr Endocrinol Metab. 2015;20:34-9.
5. Revised IAP growth charts, 2015. Available at iapindia.org/ Revised-IAP-Growth-Charts-2015.php. Accessed on 12 July 2016
6. Garg P. Short stature in Indian children: experience from a community level hospital. Sri Lanka J Child Health. 2004;34:84-8.
7. Ullah F. Short stature: what is the cause in our population. J Ayub Med Coll Abbottabad. 2016;28(1):135-40.
8. Rabbani MW, Khan WI, Afzal AB, Rabbani W. Causes of short stature identified in children presenting at a tertiary care hospital in Multan Pakistan. Pak J Med Sci. 2013;29(1):53-7.
9. Shiva S, Nikzad A. Etiology of short stature in East Azerbaijan, Iran. Iran J Pediatr. 2009;19(1):35-40.
10. Sultan M, Afzal M, Qureshi SM, Aziz S, Lutfullah M, Khan SA, et al. Etiology of short stature in children. J Coll Physicians Surg Pakistan. 2008;18(8):493-7.
11. Lashari SK, Korejo HB, Memon YM. To determine frequency of etiological factors in short statured patients presenting at an endocrine clinic of a tertiary care hospital. Pak J Med Sci. 2014;30(4):858-61.
12. Chowdhury SP, Sarkar TK, Haldar D, Taraphdar P, Naskar TK, Sarkar GN. Short stature in children: Experience from a tertiary care hospital in Kolkata, India. The Health. 2011;2(4):139-42.