

Original Research Article

Clinical presentations of congenital hypothyroidism in pediatric age group

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ABSTRACT

Background: Hypothyroidism (HT) in pediatric age group is of prime importance because of its varied presentations. Missing a diagnosis of hypothyroidism is disastrous since it results in mental retardation (MR). Hence, it is mandatory to know the various types of clinical presentation in clinical practice.

Methods: Study includes 30 children who were diagnosed to have hypothyroidism. Detail history were taken thorough examination carried out and thyroid function tests were obtained in all the cases. Results were statistically analyzed in Microsoft office excel 2007.

Results: Majority of the children were diagnosed at the age of 1-3 month of age. Males and females were almost equally affected, 70% of the children had constipation, 26.7% of the cases presented with umbilical hernia, feeding difficulties were seen in 23.3%, abdominal distention and prolonged jaundice were seen in 20% of the cases. 10% of the cases presented with short stature and 6.6% children showed obesity.

Conclusions: Detailed history and examination of each infant will help in early diagnosis of congenital hypothyroidism (CH). High index of clinical suspicion should be made when any of clinical features of hypothyroidism are present.

Keywords: Congenital hypothyroidism, Hypothyroidism, Mental retardation

INTRODUCTION

Congenital hypothyroidism results from deficient in production of thyroid hormone, it is classified mainly as primary and central, central hypothyroidism also called as hypopituitary hypothyroidism. In primary hypothyroidism problem is in gland itself whereas in central hypothyroidism there is decreased production of thyroid stimulating hormone (TSH).¹ CH may be manifested at birth (congenital) or acquired (symptoms appear after a period of normal thyroid function), acquired HT may be due to truly acquired or result of late manifestation of congenital disorder of thyroid gland. About 80-85% of permanent CH accounts for thyroid

dysgenesis in which 33% cases are due to thyroid aplasia and 66% of cases account for hypoplasia and ectopic thyroid.^{2,3} In India it is reported to occur with incidence of 1:2640 live births. The prevalence of congenital hypothyroidism (CH) initially reported as 1 in 4000 infants worldwide, however over the last two decades the prevalence has dropped to 1 in 2000 likely the result of detection of milder cases of hypothyroidism. Newborn screening for congenital hypothyroidism in most of the centers have given a promising outcome in early diagnosis and treatment of the disease. At times we may miss the diagnosis of CH due to its clinical clues mistaken for other clinical condition like constipation of Hirschsprung's or prolonged jaundice of breast milk

jaundice. However, acquired hypothyroidism in pediatric age group stands last as far as etiology is concerned.⁴

Many times, we miss subclinical hypothyroidism, they are quite often asymptomatic and manifestations may include nonspecific complaints like memory impairment, slowness of thinking.⁵ Other complaints like muscle cramps muscle weakness, tiredness, dry skin, hoarseness of the voice, puffy eyes and constipation are not uncommon.⁶

METHODS

Hospital based prospective study conducted at tertiary care center of Mangalore for the period of 3 years from 2009 to 2011. Total number of 30 children were included in this study.

Inclusion criteria

All the freshly diagnosed cases of congenital hypothyroidism below the age of 1 year prior to starting thyroxine.

Exclusion criteria

- Babies who are having metabolic or neurodegenerative disorder.
- Babies who are having genetic chromosomal disorder.

History on poor feeding, decreased activity, constipation, abdominal distension, prolonged jaundice, developmental delay was taken. Weight, height, head circumference was recorded, presence of umbilical hernia, abdominal distention, coarse facial features, large tongue and jaundice were identified and recorded. Thyroid function test was done in all the cases. All the clinical data and lab parameters were recorded. Microsoft office excel 2007 statistical results were obtained.

RESULTS

Table 1 depicts gender distribution where in out of 30 cases 14 children were males which is equal to 46.7% and 16 children were female equal to 53.3% of the cases.

Table 1: Gender distribution.

Gender	Number of children	Percentage
Male	14	46.7
Female	16	53.3
Total	30	100

Table 2 shows, age at which children were diagnosed to have congenital hypothyroidism. It is divided into four age groups, below 1 month, 1-3 months, 3-6 months, 6-12 months. Children who are diagnosed to have CH before 1 month are 3 in number which is equal to 10% of

the total cases. Children who are diagnosed between 1-3 months are 11 in number which is equal to 36.7%, children diagnosed between 3-6 months were 8 in number is equal to 26.7% and another 8 children were diagnosed between 6-12 months.

Table 2: Age distribution.

Age in years	Number of children	Percentage (%)
<1 month	3	10.0
1-3 months	11	36.7
3-6 months	8	26.7
6-12 months	8	26.7
Total	30	100%

Table 3: Clinical features.

Clinical features	Number of patients	Percentage (%)
Speech delay	7	23.3
Constipation	21	70
Umbilical hernia	8	26.7
Feeding difficulties	7	23.3
Abdominal distention	6	20.0
Prolonged jaundice	4	13.3

Table 3 depicts children presenting with various clinical presentations of congenital hypothyroidism. Seven children were presented with speech delay as initial symptoms of CH which is equal to 23.3% of the total cases. Constipation was the initial symptom in 21 cases which is equal to 70% of the total number of the cases. Umbilical hernia due to hypothyroidism in our study is seen as initial symptom in 8 cases which is equal to 26.7%. Seven cases (23.3%) presented with feeding difficulties in the form of difficulty in swallowing and regurgitations. Abdominal distention mostly contributed to hypotonia of the abdominal muscle seen in 6 cases which is equal to 20.0%. Prolonged jaundice in pediatric age group are due to various etiological factors, however in our study 4 cases (13.3%) of CH are presented with prolonged jaundice as initial symptoms.

Table 4: Anthropometry.

Anthropometry variable	Number of children	Percentage (%)
Weight for age		
Appropriate	28	93.3
Obesity	2	6.7
Height for age		
Appropriate	27	90
Stunting	3	10

Table 4 shows among 30 Patients, 28 patients had normal weight for age is equal to 93.3% and 2 pts had obesity. Out of 30 patients 27 patients had normal height for age

and 3 patients were diagnosed to have short stature is equal to 10% of the total cases.

Table 5: Thyroid function test.

Thyroid profile of patients studied	No. of children studied (N=30)	Percentage (%)
T3		
Decreased	23	76.7
Normal	7	23.3
T4		
Decreased	25	83.3
Normal	5	16.7
TSH		
Increased	30	100
Normal	0	0

Table 5 depicts thyroid profile of patients studied. Out of 30 patients 23 patients had decreased T3 level is equal to 76.7% and 25 patients (83.3%) had decreased T4 level and 5 patients had normal T4 level. All the 30 patients (100%) had increased level of thyroid stimulating hormone.

DISCUSSION

Males and females are almost equally affected with ratio of 1:1.3, whereas most of the other studies showed gross difference between gender distribution. Study done by Waller DK et al, depicts male: female ratio is 1:2.⁷

In present study, majority of the patients presented with constipation which accounts for 70 % of the cases where as study done by Pezzuti I et al, showed constipation in 40% cases.⁸ Whereas 56.9% of the cases had constipation in a study done by Karki et al, in Nepal.⁹

Total 26.7% of the cases had umbilical hernia in our study and Waller DK et al, showed similar result. According to Pezzuti I et al, 51% presented with umbilical hernia.⁸ Karki et al, in their study showed only 36% cases of umbilical hernia.⁹ Another study done by Grant DB et al, revealed 32% cases had umbilical hernia.¹⁰

Speech delay and feeding difficulties were identified in 23.3% (7) cases in our study. Study done by Singh A et al, showed speech delay in 37.7% and feeding difficulty in 19.6% of the cases.¹¹ As per data provided by Pezzuti I et al, 1.4% children had neuropsychomotor development and 5.6% cases had feeding difficulties. Study done by Karki et al, depicted 26.3% children had feeding difficulties.⁹ Abdominal distention in a case of hypothyroidism is mainly attributed to hypotonia of the abdominal muscle. In present study 20% (6) cases had abdominal distention and it is par with many other

studies. Present study showed 20% cases had prolonged neonatal jaundice whereas study done by Singh et al, presented with prolonged neonatal jaundice in 14.7% cases.¹¹

CONCLUSION

Congenital Hypothyroidism has taken a new turn in many medical care centers due to routine new born screening which helps in early detection of the disease and prevents complication. However, it is very important in day to day clinical practice to concentrate on early symptoms and signs of hypothyroidism which helps to pick up missed cases of CH during neonatal screening.

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