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Case Report

OUTCOME OF TWO CASES WITH LATE DIAGNOSIS OF CONGENITAL HYPOTHYROIDISM – A CASE REPORT

***Joshi GA and Joshi PG**

[1] *Composite Regional Centre for Persons With Disabilities, Department of Disability Affairs, Ministry of Social Justice and Empowerment, Government of India, Punarvas Bhavan, Khajuri Kalan, Piplani, Bhopal 462022*

**Author for Correspondence*

ABSTRACT

Two girls with congenital hypothyroidism followed up at our centre are presented. The time of diagnosis, institution of medical management and therapeutic inputs are compared with the outcome in their growth and development. While early identification and thyroxine supplementation form the core management strategy, this case study emphasises how therapeutic inputs of composite nature improve the outcome in cases of congenital hypothyroidism.

Keywords: *Congenital Hypothyroidism, Thyroxine, Child Development, Early Diagnosis*

INTRODUCTION

Early diagnosis and treatment can prevent disability arising due to congenital hypothyroidism. Health policies in developing countries have taken successful steps to prevent hypothyroidism through iodization of edible salt. However, they are lagging in screening for congenital hypothyroidism, which should be given equal importance, just like universal immunization for any child.

CASES

Presenting Concerns

Two girls of different ages reported at Composite Regional Centre for persons with disabilities (CRC) - Bhopal at different times. The presenting concerns were stunted growth and delay in speech.

Clinical Findings

Case 1: A 1½ year old girl was brought to CRC-Bhopal in year 2004 by her mother with presenting complaint of delayed speech. She was from lower socioeconomic status with 3 elder sibs having normal developmental history and was staying in a nuclear family. Her height was 67 cm (below 10 percentile for age), had rough skin and lanugo hair. Her serum TSH level was 87 IU/ml.

Case 2: A 5 years old girl was seen in year 2010 at her residence with presenting complaints of stunted growth and delayed development. She stayed in a residential institution caring for abandoned children having disabilities. Her height was 75 cm (below 3 percentile for age), had moon face with puffy eyes, unerupted teeth, pot belly and sluggish activities. Her serum TSH level was 135 IU/ml. She was referred to CRC-Bhopal for management.

Timeline

The comparison of the two cases from identification to Dec. 2013 is given in Table 1.

Table 1: Comparison of the Two Cases

Heading	Case 1	Case 2
Year of presentation	2004	2010
Age of starting thyroxine	1.5 years	5 years
Frequency of follow up	Once in 3 -6 months	Once in 12-18 months
Status in Dec 2013	Normal growth	Poor growth
	Mild delay in mental age	Severe developmental delay
	Achieved speech and personal social milestones	Achieved rudimentary social communication

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Diagnostic Focus and Assessment

The developmental delay, stunted growth, facial features and raised TSH pointed to congenital hypothyroidism. The diagnosis was confirmed from pediatrician and endocrinoloigst. The main challenge was the delay in instituting thyroxine supplementation.

Therapeutic Focus and Assessment

The patients were started with Tab. levo-Thyroxine 50 micrograms daily once in early morning on empty stomach. Comprehensive developmental therapy services were provided at this centre with occupational and speech therapy. The caretakers were conselled for regular thyroxine supplements and developmental therapy.

Follow Up and Outcomes

Thyroxine dose was adjusted according to clinical response and TSH level in both cases. Clinical psychology for behaviour management and special education for school readyness were provided at CRC-Bhopal.

Case 1: She had fever and tachycardia intermittently that was managed by reducing dose of thyroxine. Presently at age of 10 years she has attained 140cm height, 33kg weight, IQ = 55, speaks and walks well and is attending 2nd standard in a normal school. (Figure 1)

Case 2: required frequent hospitalizations for associated rickets, hernia and gastrointestinal dysmotility. At age of 8½ years, she has 89 cm height, 8 kg weight, IQ = 21, stands holding support, walks with help of CP walker, responds with smile and cry and is attending a special school. (Figure 2)

Patient Perspective

Case 1: The mother is happy with normal growth of the patient but is anxious about her school performance.

Case 2: The caretaker is pleased with improved social responsiveness and walking ability of the child but worried about her poor growth and bloated abdomen.

Figure 1: Case 1 achieved normal growth

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Figure 2: Case 2 achieved supported mobility

DISCUSSION

We were able to curb impairment in both cases to varying extent by thyroxine supplementation and comprehensive habilitation despite delay in diagnosis. Counselling ensured follow up for proper dose and developmental inputs. Prevalence of congenital hypothyroidism is 1:2050 in China (Gu *et al.,* 2008). The incidence is reported to be increasing in USA with a caution for close follow up of transient hypothyroidism (Olney *et al.,* 2010). In India, more than 80% of the districts surveyed pointed to risk of iodine deficiency that may cause wide range of impairments viz. abortion, stillbirth, intellectual deficiency, deafness, squint, dwarfism, goitre etc. (DGHS, MoHFW, GOI, 2006). Congenital hypothyroidism is identified at average age of 4 years in India (Virmani *et al.,* 1989).

The case 1, who was treated at 1½ years age achieved motor and speech milestones with normal growth curve. In contrast, the case 2, treated at 5 years age achieved fewer motor and social milestones and was stunted in growth.

Conclusion

We conclude that rehabilitation medicine inputs with thyroxine supplementation should be instituted as soon as possible in cases of congenital hypothyroidism. It is high time to implement active case identification, treatment and rehabilitation of congenital hypothyroidism for all infants so that children achieve maximum potential of development and growth.

Informed consent – Obtained.
Competing interests – Nil
Ethics approval - The authors followed the guidelines of WMA Declaration of Helsinki.
De-identification – Yes

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