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A Severely Neglected Paediatric Case: Congenital Hypothyroidism

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Abstract **Case Report**

Introduction: Congenital hypothyroidism is a common endocrinal disorder of newborn. Early diagnosis and management is essential to prevent the irreversible complication of congenital hypothyroidism. So, widespread implementation of newborn screening for this condition is very important [1]. ***Case report*:** A 14- year- old boy presented with short stature, chronic constipation, intellectual disability and delayed puberty as a result of untreated congenital hypothyroidism. ***Conclusion***: This case shows the importance of early diagnosis and management of congenital hypothyroidism by routine newborn screening and adequate replacement therapy.

Keywords: Hypothyroidism, Newborn Screening, Intellectual Disability, Short stature.

INTRODUCTION

Congenital hypothyroidism is a disorder of thyroid hormone deficiency that occurs due to dysgenesis of thyroid gland or dyshormonogenesis.

Most infants with congenital hypothyroidism are asymptomatic at birth [1-3]. But during the first few month of life, the symptom of hypothyroidism such as feeding problem, somnolence, hoarse cry and constipation usually developed [1, 4].

Newborn screening is a well-established system for the diagnosis of congenital hypothyroidism. But it is not practiced in many third-world countries [5].

This case report describes a neglected case of congenital hypothyroidism that was left untreated for 14 years and socially stigmatized as a disable child. Illiteracy and ignorance of the parent is the main cause of the parents negligence.

CASE REPORT

A 14 – year – old boy hailing from Pakundia, Kishoreganj, Bangladesh was brought by his parents to Health Aid Diagnostic and Hospital with complaints of growth failure, short stature, chronic constipation, intellectual disability and delayed puberty. He was born at home at term without any complications and was not screened for hypothyroidism at birth. He had a history of delayed passage of meconium and prolong jaundice in post natal period. The mother also stated that the boy was less active and excessively sleepy. He was treated by traditional healers and the parents never asked for medical advice before.

On examination, the boy was less active and less interested in his surroundings. The boy presented with dull-coarse facies, (Fig-1) large tongue, slurred voice, short and broad hands (Fig-2), dry–rough–cold skin (fig-3). He was moderately pale with normal vital signs.

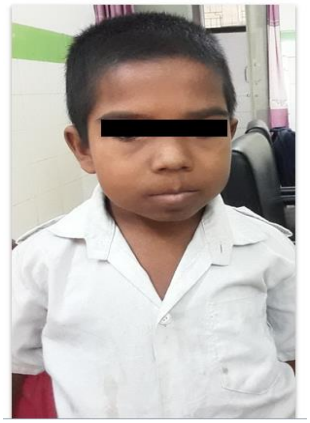

Fig-1: Dull coarse Facies with periorbital puffiness

Fig-2: Short – Broad hands

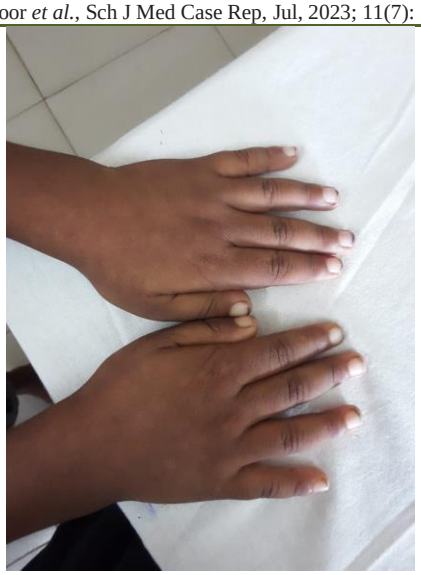

Fig-3: Dry rough skin

The boy was severely stunted (Height:105cm, Upper segment: 56cm, Lower segment: 49cm), US/LS ratio was 1.14: 1 (corresponds to 4.5 years) (Fig-4). He has generalized hypotonia with a positive Woltman sign. He has a developmental delay with a developmental age between four to seven years.

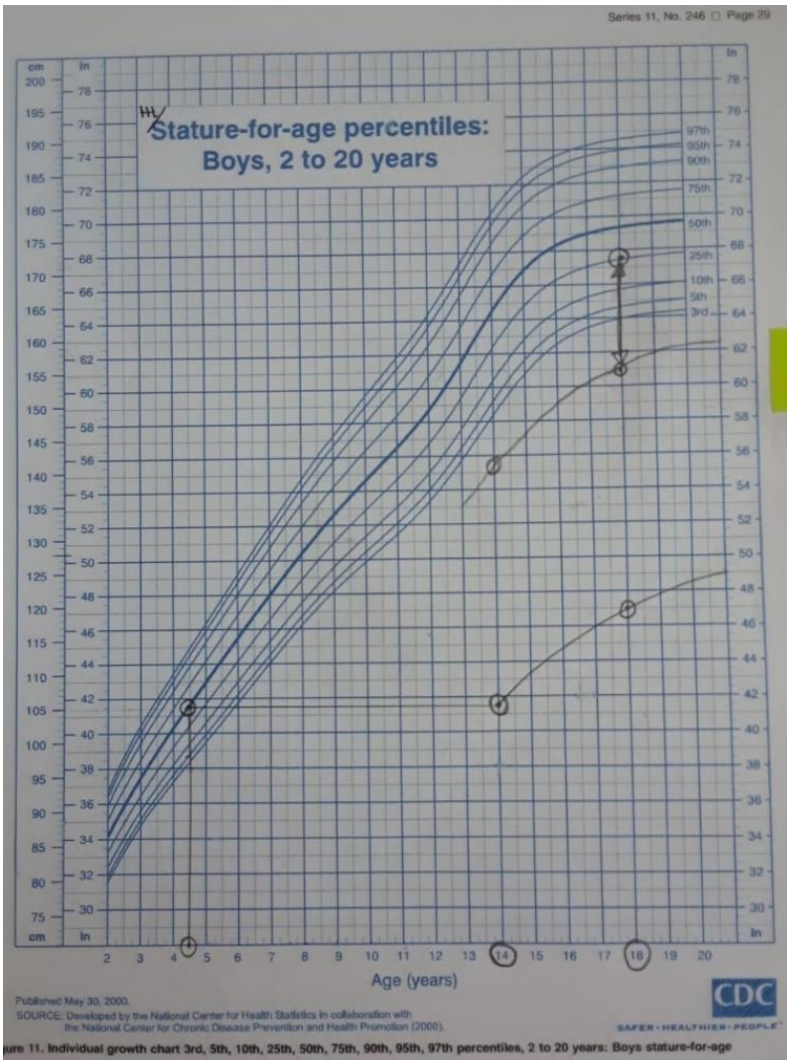

Fig-4: Growth Chart showing pathological short stature

His investigation reports revealed anemia (Hb 8.9 gm/dl), elevated level of S.TSH (100 uIU) with a decreased level of S. FT4 (2.26PmoI/L) along with delayed bone age and skeletal dysgenesis in the skeletal survey (Fig 5 & 6).

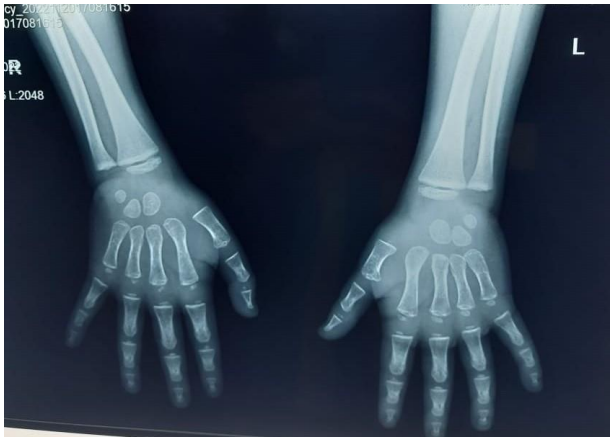

Fig- 5: X ray wrist showing delayed bone age

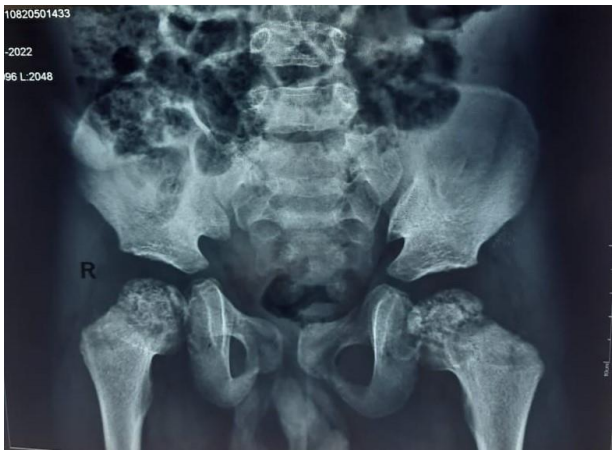

Fig-6: X- Ray Hip showing skeletal dysgenesis

Ultrasonography of the thyroid gland showed hypoplasia of both thyroid lobes. Accordingly, he was diagnosed with congenital hypothyroidism and treated with replacement therapy; Levothyroxine. The boy was on regular follow-up and showed clinical as well as biochemical improvement. After six months of treatment, the boy become more oriented and interested in surroundings, correction of facial puffiness (Fig-7), slurred voice, and constipation.

Fig-7: Follow up picture after treatment

DISCUSSION

The thyroid gland produces iodine-containing thyroid hormones that are very important for the overall growth and development of the human body specially brain development, body metabolism, and skeletal growth [1, 6]. Congenital hypothyroidism occurs when there is a defect in thyroid gland development (agenesis or dysgenesis: 80%) or a defect in thyroid hormone synthesis (dyshormonogenesis: 20%) [7, 8].

Family history as well as maternal history of a thyroid disorder should be carefully evaluated for information about a suspected congenital hypothyroidism patient [9, 10].

The diagnosis of congenital hypothyroidism at birth based on clinical evidence is quite impossible, as most infants with this condition are normal at birth [11]. Usually, specific symptoms do not appear in newborns and these cases are identified within the first two to three weeks of life [7]. So, most infants are diagnosed by the neonatal screening program.

The characteristic clinical features of congenital hypothyroidism are delayed passage of meconium, prolonged physiological jaundice, lethargy, constipation, hoarse cry, large protruded tongue with feeding problems, and coarse facies with periorbital puffiness. There may be an umbilical hernia, dry-rough skin, hypothermia, cardiomegaly, bradycardia, and asymptomatic pericardial effusion [1] hypotonia and delayed relaxation of deep tendon reflex [1, 12], delayed achievement of developmental milestones, intellectual disability and short stature are also important clinical presentation [1]. As the child grows, infantile body proportions are maintained and the child usually has disproportionate short stature [7].

Early diagnosis and adequate treatment ensure normal linear growth and intelligence. Thyroid hormone is critical for brain development and effective treatment should be started immediately to prevent irreversible brain damage [4, 13, 14].

Diagnosis of congenital hypothyroidism is confirmed by elevated levels of thyroid stimulating hormone (TSH) and decreased levels of serum thyroid hormone (total or free T4). Other supplemental investigations are; X- rays of different bones (as per age) showed a delayed bone age [1, 12], USG and CT scan of thyroid gland to detect the cause of hypothyroidism.

The drug of choice for congenital hypothyroidism is thyroxine hormone replacement for life. Treatment must be continued and the patient should be monitored clinically and biochemically (S.T3 & T4 level) after 4 to 6 weeks of initial therapy, then every 3 months during 2nd & 3rd year of life and thereafter every 6 months [15]. Every effort should be exerted to maintain s. T4 level in the upper half of the normal range [15].

CONCLUSION

Early diagnosis and adequate treatment within first 14 days of life results normal linear growth and intelligence. So, to prevent irreversible complications of congenital hypothyroidism, nationwide establishment of neonatal screening program is a must. Additionally, health education to parents, early replacement therapy and regular careful follow up of patient are also similarly essential [17].

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