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A CASE REPORT OF CONGENITAL HYPOTHYROIDISM

Paediatrics

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ABSTRACT

Congenital hypothyroidism (CH) is one of the most common preventable causes of mental retardation in children. The incidence in India is estimated to be 2.1 per 1000 live births which is 8 times higher than what is reported in western literature. Newborn screening programs should be confirmed by finding an elevated serum TSH and low T4 or free T4 level. Cord blood remains a very practical alternative for screening purposes, and thus is the practice in some Asian countries. The IAP recommends universal newborn screening by elevated serum TSH and low T4 or free T4 level by the use of cord blood samples. Dysgenesis and dyshormonogenesis are the most common causes of congenital hypothyroidism.

KEYWORDS

Congenital Hypothyroidism, Cord Blood Screening, TSH,, free T3, FreeT4

INTRODUCTION

Congenital hypothyroidism (CH) is one of the most common preventable causes of mental retardation in children. The incidence in India is estimated to be 2.1 per 1000 live births which is 8 times higher than what is reported in western literature. Newborn screening programs should be confirmed by finding an elevated serum TSH and low T4 or free T4 level. Cord blood remains a very practical alternative for screening purposes, and thus is the practice in some Asian countries. The IAP recommends universal newborn screening by elevated serum TSH and low T4 or free T4 level by the use of cord blood samples. Dysgenesis and dyshormonogenesis are the most common causes of congenital hypothyroidism.

Case Study

A 6 months old female patient was bought by her mother with History of Cough and cold, noisy breathing, sluggishness in activity and not interested in surroundings and decrease frequency of stool since 5 months, not gaining weight since 1.5 months and crying during micturation and feeding difficulty since 15 days. Patient visited multiple times to pediatrician for cough and cold. He was born out of nonconsanguineous marriage with uneventful birth history. Only birth vaccine taken. History of bottle feeding with formula feed present. Patient had gross development delay. She had short stature with length 52 cm (< 0.5 pg/mL, Free T4 - < 0.1 ng/dl and TSH - > 100 µIU/ml. USG neck – Thyroid gland is not visualized at its normal position in the neck. Possibility of absent/hypoechoic/ectopic thyroid gland.USG abdomen Suggestive of bilateral medullary nephrocalcinosis. OAE - both ear fails. -ENT and endocrinology refer was done. They advised 56µg of Thyroxine tablet once orally in the morning one hour before food was started at 6 months of age, warning his parents not to stop the drug at any instance. The patient was advised initial 1-2 month follow up in 1 st year, every 2-3 months till 3 years and 6 months follow up till 10 years as literacy in family was low frequent follow was necessary. During follow up improvement was noted after 6 week of treatment, the patient became more oriented and interested in the surroundings.

DISCUSSION:

Congenital hypothyroidism (CH) is defined as thyroid hormone deficiency present at birth. Congenital hypothyroidism is classified into permanent and transient CH. Screening a newborn for congenital hypothyroidism is very important as mental retardation can be prevented in 85% of the cases with early initiation of thyroxine supplementation therapy before 3 months of age. The incidence of CH in India varies from 1:2500 to 1:2800 live births. Common symptoms include decreased activity and increased sleep, feeding difficulty, constipation, and prolonged jaundice. On initial examination, the most common signs are a distended abdomen with umbilical hernia, macroglossia, myxedematous facies and cold or mottled skin. Hoarse cry, constipation, neonatal hyperbilirubinemia more than 3 weeks due to immaturity of hepatic glucuronyl transferase are common features seen. Thyroid hormone is also important in the formation and maturation of bone. Deficiency leads to a wide posterior fontanel of > 5 mm and absent femoral epiphyses in up to 54%.Neurologic examination findings include hypotonia with delayed reflexes. CH is associated with an increased risk of congenital malformations. Other associated malformations include spiky hair, cleft palate, neurologic abnormalities and genitourinary malformations. Newborn screening is the most important method to identify infant with CH. Most screening program measures level of TSH which is elevated in primary hypothyroidism often to >100mU/L. Other diagnostic studies are Thyroid radionuclide uptake in scan, Thyroid USG, Serum thyroglobulin measurement, Anti-thyroid antibody determination, urine iodine determination and genetic mutation. Levothyroxine is the treatment of choice - recommended initial dose is 10-15 mcg/kg/day. It is important to monitor frequently serum T4 or free T4 and TSH during 1st 3 year of life to ensure optimal therapy and to avoid overtreatment.

CONCLUSION:

Growth retardation and mental retardation of children due to hypothyroidism can be totally prevented by – 1.Implementing universal neonatal screening program for hypothyroidism and educating parents about the sign and symptoms of hypothyroidism. 2.Adequate and timely L-thyroxin replacement therapy. Very careful follow-up of patients and adjustment of the L-thyroxin dose to maintain serum T4 levels in the upper half of the normal range during the first year of life. Any diagnostic delay, inadequate treatment or even poor compliance to treatment is always associated with irreversible damage as those manifested in this case

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