Case Report

Congenital Hypothyroidism in Newborn - A Study on Early Detection and Management

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ABSTRACT

Congenital hypothyroidism is the most common neonatal disorder, Congenital hypothyroidism may have major detrimental effects on growth and neurological development, Thyroid gland disorder (aplasia or hypoplasia) is an important cause of congenital hypothyroidism and usually known as thyroidal dysgenesis. The diagnosis of congenital hypothyroidism is not always easy to make due to nonspecific symptoms, usually specific symptoms do not appear in newborn, Early intervention can lead to excellent outcomes. This case study presents a Congenital hypothyroidism in newborn detected and diagnosed early on day 4, appropriate treatment and follow up resulted in restoring the thyroid hormone levels to normal and excellent outcomes in maintaining clinical and biochemical Euthyroidism.

Keywords: Congenital Hypothyroidism, Euthyrodism, Aplasia, hypoplasia, thyroidal dysgenesis.

INTRODUCTION

Congenital hypothyroidism (CH), may have major detrimental effects on growth and neurological development, but early intervention can lead to excellent outcomes.[1] Congenital hypothyroidism can be defined as variable dysfunction of the hypothalamic-pituitary -thyroid axis present at birth. This dysfunction leads to insufficient thyroid hormone production and subsequently, to moderate or severe thyroid dysfunction[2]. Congenital Hypothyroidism is caused by absent or defective thyroid gland. It is classified into agenesis (22-42%) ectopy (35 -42%) and gland in place defects (24-36%).[3,4] It occurs 1:1300 to 1:4000 births worldwide.[5] The age at which symptoms appear depends on the impairment degree of thyroid function. The majority of new born affected by Congenital Hypothyroidism are unlikely to exhibit early symptoms, this is believed to be attributed to the transplacental transfer of maternal thyroid hormones and the residual thyroid activity in neonates. Classical clinical signs appear overtime and include macroglossia, facial edema, umbilical hernia, hypotonia, hypothermia, lethargy goiter, feeding difficulties, constipation, bradycardia, wide fontanelles, prolonged jaundice, and developmental delay.[6] In particular prolonged indirect hyperbilirubinemia, enlarged posterior fontanel (> 0.5 cm), stunted growth, and goiter give raise to the suspicion of congenital Hypothyroidism.

New born screening is very important in diagnosis of Congenital Hypothyroidism. It is generally performed in the first days of life (24 -72 hrs)[7] at this age the clinical diagnosis of Congenital Hypothyroidism is difficult or even impossible. International guidelines recommends immediate initiation of treatment with Levothyroxine (LT4), and closely monitoring hormonal levels, to facilitate appropriate adjustments in therapy[8]. The purpose of this study is to report a case of Congenital hypothyroidism in new born.

Case Presentation:

A moderate male preterm (32 week, 4 days) Dichorionic Diamniotic Twin 2, with birth weight 2.05 kg, born to 30 yr old primigravida mother history of hypothyroidism on Thyronorm 100 ug married since 3 yrs. Baby cried immediately after birth with normal Apgar score Routine new born care was given.

In view of persistent respiratory distress Neo puff ventilation for 10 mts and shifted to NICU for further care. In NICU baby kept on oxygen for 2 hrs. HR-154/min RR 48/min SpO_2 100%, As respiratory distress settled, baby was weaned to room air, Baby in room air maintained saturation with no distress, hence baby shifted to mother side baby started on paladai feeds baby tolerated well.

Diagnosis And Treatment:

On day 4 Total bilirubin was 6.67 mg/dl with direct bilirubin 0.43 mg/dl indirect fraction 6.24 mg/dl, below the phototherapy levels, FT3- 2.33pg/ml. FT4 -1.14 ng/dl, TSH 213.2 uIu/ml, Day 5 FT3 -2.34pg/ml, FT4- 1.14ng/dl, TSH- -273.9 uIu/ml. In view of high abnormal TSH on advice from pediatric endocrinologist Thyronorn 25 ug 1-0-0 & ½-0-0 on alternate days and continued the same line of management. Baby was hemodynamically stable, active, euglycemic, vitals stable, tolerating feeds discharged on advice to continue the same medication and feeds, immunization as per schedule, to report to nearest doctor if the baby develop any lethargy, poor feeding, Jaundice, decreased output, bluish discoloration, To follow Neuro

developmental assessment after 3 months, to do FT4 &TSH periodically and to monitor the Thyroid hormone levels.

Table 1: Periodical Thyroid Hormone Levels

S.No	DAYS	FT3 pg/ml	FT4 ng/dl	TSH ulu/ml
1	Day 4	2.33	1.19	213.2
2	Day 5	2.34	1.14	273.9
3	Day 29	-	1.39	90.48
4	Day 60	-	1.45	48.48
5	Day 90	-	1.81	25.31
6	Day 150	-	1.45	5.84

Normal Thyroid Function Test For New Born

Free T3 - 2.08 - 4.42 pg/ml

Free T4 - 2.20 - 5.30 ng/dl

TSH - 1.00 - 39.0 ulu/ml

DISCUSSION

Thyroid gland disorder (aplasia or hypoplasia) is an important cause of congenital hypothyroidism and usually known as thyroidal dysgenesis. If the hypothyroidism developed early during fetal life, the effect might appear in certain organ systems, including the central nervous system and skeleton, However, most infants with congenial hypothyroidism are normal at birth based on clinical evidence is rarely possible[9-11]. The diagnosis of congenital hypothyroidism is not always easy to make due to nonspecific symptoms. Usually, specific symptoms do not appear in newborns. Since symptoms and signs develop gradually after birth, the timing of clinical features will vary depending on the severity of hypothyroidism. Hence confirmatory Thyroid Function Test at appropriate time is crucial in diagnosing congenital hypothyroidism and initiating early treatment options. In our case study no specific symptoms noticed except for respiratory distress and hence it was very difficult to diagnose the new born child for congenital hypothyroidism. However, the Thyroid Function Test (TSH -213.2 ulu/ml) reports on day 4 was very useful in suspecting and diagnosing for congenital hypothyroidism. Congenital Hypothyroidism for most of the infants are diagnosed by[12] newborn screening programs reports which takes between 5 to 10 days, moreover most of the new born screening programs measure TSH levels only, even though it has high sensitivity. Congenital hypothyroidism screening can have false negative results due to delayed rise in TSH levels that often occurs during the first week of life, additionally the use of glucocorticoids, somatostatin dopamine observed to inhibit TSH release , Also the reports of Thyroid Function Test gives the values of FT3 & FT4 levels along with TSH it was very much useful in initiating the treatment with Levo thyroxine immediately, This case advocates the need for performing the Thyroid Function Test for new born suspected for congenital hypothyroidism with or without specific symptoms, also the Thyroid Function Test are becoming much easier with short Turnaround time with different methodology of Chemiluminescence (CLIA), Electrochemiluminescence (ECLIA), Enzyme-linked Fluorescent Assay (ELFA), with significant sensitivity and specificity and also with small sample volume for analysis.

Early diagnosis and adequate treatment in the first week of life result in the normal linear growth and intelligence. The longer delay in diagnosing Congenital hypothyroidism, the higher the risk of mental retardation and various neurological sequelae.[13] Thyroid hormone is critical for normal brain development and effective treatment must be initiated promptly to prevent irreversible brain damage[14,15].

In our case study since the treatment started on day 5, there was significant and periodical decrease in TSH levels (Table 1), and TSH reached almost normal in 90 days. Early diagnosis, intervention leads to excellent outcomes especially in growth and neurological developments.

CONCLUSION

Congenital hypothyroidism is one of the most common treatable causes of mental retardation, The timing of therapy is crucial to growth and neurologic outcomes, indeed there is an inverse relationship between intelligence quotient (IQ) and age at which diagnosed, even when diagnosed early, neurological development may suffer if treatment is not optimized in the first two to three years of life. Therefore, it is important for Congenital infants to receive early treatment and close follow up will result in rapidly restoring the FT4 and TSH to the normal levels and maintaining clinical and biochemical euthyroidism.

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