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CASE REPORT - PEDIATRICS

"THE BABY HOUDINI" – A CASE OF CENTRAL CONGENITAL HYPOTHYROIDISM

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Abstract

Introduction : Central Congenital Hypothyroidism (CCH) is defined as an inadequate stimulation of the thyroid gland by the pituitary, at birth. 1 in 13000 children get affected by CCH. Although majority of CCH occur as a part of combined pituitary hormone deficiencies (CPHD), it can also occur rarely as an isolated condition. Upto 90% of isolated central CH cases have identifiable genetic causes (IGSF1, TBL1X, IRS4). This case report provides information on a practical approach to diagnosis and management of this alluring condition.

Conclusion : Due to the severity of the condition, early detection and treatment leads to good neurodevelopmental outcome.

Keywords : Congenital Central Hypothyroidism (CCH), New-born Screening, Combined Pituitary Hormone Deficiency (CPHD)

INTRODUCTION

Central congenital hypothyroidism (CCH) is a rare disorder at birth due to defective stimulation of a normal thyroid gland by thyroid stimulating hormone (TSH) with deficiency in thyroid hormone biosynthesis.[1,2] Thyroid hormone (TH) deficiency may be either due to defective development of thyroid gland or its function (primary or thyroidal CH), or due to inadequate stimulation of a normal thyroid gland by the pituitary (central CH). Although rarely an isolated condition, CCH often occurs as a part of combined pituitary hormone deficiencies (CPHD).[3,4] TH deficiency is difficult to recognize immediately after birth and it impedes the growth and development of brain.[5]

CASE REPORT

Figure 1 : Clinical Findings

A 2 month old male infant, first born of 3rd degree consanguineous marriage, was delivered at 38 week of gestation by Normal Vaginal Delivery with birth weight 2.45 kg, length 50cm , Head Circumference 35 cm falling between 10th-25th percentile, 25th-50th percentile and 50th-75th percentile respectively.

The child presented with history of yellowish discolouration of eyes noticed by the mother for 15 days, progressive in nature now involving whole body including palms and soles, history of high coloured urine 15 days, history of feeding difficulties and history of somnolence. No history of clay coloured stools

Antenatal History: uneventful

CLINICAL EXAMINATION

HEAD TO TOE EXAMINATION

Anterior Fontanelle (AF) - 5.5* 5 cms
Posterior Fontanelle (PF) - 6.5* 5 cms
Head Circumference (HC) - 37 cm
Eyes - icteric
Depressed nasal bridge
Small mouth
Protruding tongue
Dry skin
Abdomen - N
Genitalia - N
QUEBAC SCORING: 8/13
Other system examination: normal.

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INVESTIGATIONS

Complete Blood Count (CBC) : Mean corpuscular volume (MCV) - 104.9fl (elevated)

Total Biirubin (T.B) -**15.8**

Direct Bilirubin (D.B) - 0.8

Indirect Bilirubin (I.B)-**15**

Serum Glutamic-Oxalacetic Transaminase (SGOT) -50

Serum Glutamate Pyruvate Transaminase (SGPT) - 43

Random Blood Sugar (RBS) - 84

Thyroid Stimulating Hormone (TSH) - **0.094 mcg U/ml** (0.7-8.3)

Free Thyroxine (Ft4) - **0.02 mcgU/ml** (0.89-2.20)

USG (Neck): normally located thyroid gland.

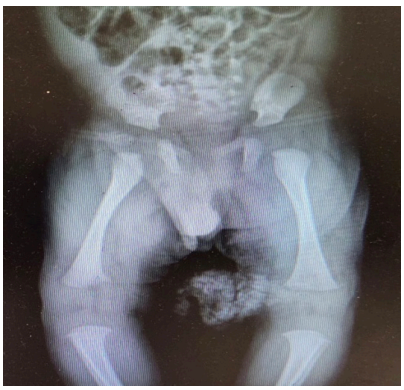

Figure 2 : X-ray B/L knee

X-ray both knee: Absence of epiphysis of both ends of femur.

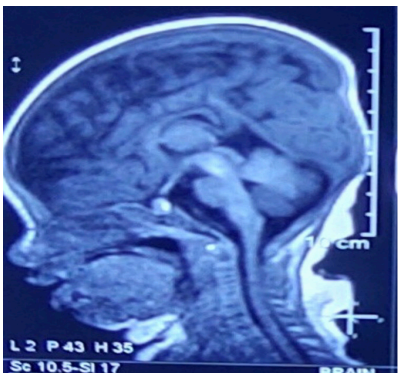

Figure 3 : Magnetic Resonance Imaging (MRI) brain

MRI brain : Pituitary visualized.

HORMONAL EVALUATION

S. Cortisol - 5.01 (2.8 – 23)

Testosterone - 91.7(60-400)

Growth Hormone (GH) - 5.06 (2- 10)

Follicle Stimulating Hormone (FSH) - 2.3(<3)

Luteinizing Hormone (LH) - 0.2(<0.3)

Prolactin - **103 ng/ml (4.05-15.2)**

Based on the above investigation, the diagnosis is confirmed to be isolated central hypothyroidism by detection of elevated prolactin levels.

TREATMENT

The child was started on T.thyroxine 10 mcg/kg/day. After 3 days of Thyroxine administration, icterus reduced, feeding improved.

Repeat **T. Bilirubin is 5.6 and D. Bilirubin is 0.4.**

CONCLUSION

Due to nonspecific signs such as feeding difficulty, hypoglycaemia and prolonged jaundice, clinical diagnosis is often missed despite early hospital admission. Since TSH levels are low, TSH based newborn screening cannot detect central hypothyroidism. As early diagnosis is missed, children are at risk of developmental delay and growth failure. Both isolated and CPHD, if diagnosed and treated early has an excellent neurodevelopmental prognosis.

LIMITATION

Thyrotropin-releasing hormone (TRH) stimulation test and genetic analysis is not done for this child.

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