

Case Study on Rare Disease: Central Hypothyroidism in Case of Pituitary Hypoplasia

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Abstract: Central hypothyroidism is defined as hypothyroidism due to insufficient stimulation by thyroid stimulating hormone (TSH) of an otherwise normal thyroid gland. It has an estimated prevalence of approximately 1 in 80,000 to 1 in 120,000. It can be secondary hypothyroidism (pituitary Hypoplasia) or tertiary hypothyroidism (hypothalamus) in origin. In children, it is usually caused by craniopharyngiomas or previous cranial irradiation for brain tumors or Pituitary Hypoplasia or hematological malignancies. In adults, it is usually due to pituitary macroadenomas, pituitary surgeries or post - irradiation.

Keywords: Central hypothyroidism, Pituitary Hypoplasia, Pituitary, Craniopharyngioma

1. Introduction

Central hypothyroidism is an uncommon condition characterized by insufficient thyroid gland stimulation by TSH, owing to hypothalamic and/or pituitary dysfunction. It is rarely isolated but more often occurs in conjunction with deficiencies of other pituitary hormones, as well as with neurologic symptoms and signs owing to hypothalamic/pituitary lesions. The diagnosis rests on documentation of clinical and biochemical hypothyroidism with an inappropriately low or nonelevated serum TSH level. Recent studies suggest that the temporal pattern of TSH secretion, as well as TSH structure, is altered in central hypothyroidism, providing a mechanism for the induction of the hypothyroid state in this condition.

2. Case Report

A 6 month old male child born of non - consanguineous marriage presented to paediatrics opd with complain of High grade fever since 4 day, Cough since 4 day, facies with prominent eye balls since 6 month of age. Patient having past history of PICU admission for Recurrent Cough and Cold. No other complaints at present.

On detailed history the child having Neck Holding and Sit with support at age of 6 month absent, palmar Grasp present, Mouthing Present, Cooing Present, Mother Regard Present, Patient On CNS examination tone of both upper limb was Normal, deep tendon reflex was brisk. Other skeletal, eyes, ear examination was normal. On Head to Toe examination child having Prominent Eye balls, Umbilical Herniation.

All routine investigations (blood) was within normal limit except WBC count was 14,100, CRP Was 24, chest x - ray was s/o Bilateral Perihilar Mid Lower zone Consolidation. Hence Acute Bronchiolitis With secondary Bacterial infection labeled. USG NECK S/O both lobe of thyroid and

isthmus and submandibular gland shows normal. All major vessels are normal. **MRI BRAIN** done which suggestive of Small Pituitary Gland (Height of 2.5mm) Represent Pituitary Hypoplasia, Mild Prominence of all ventricles, 2D ECHO S/O Normal Heart Study.

On Blood investigation (9 Dec 2022) [TSH - 0.39mIU/L Normal 0.58 - 5.56 SGPT 188U/L, S. Creat - 0.39mg/dl, LH - 0.62, Testosterone - <2.50, S. Cortisol (8am) - 12.39 CSF LDH 31U/L ADA 64 U/ml Glucose 57 mg/dl Microprotein 35mg/dl 9 Dec 2022 Vit D3 24.58 ng/ml T3 1.40 pg/ml, S. Free T4 0.759 (age specific Range 0.9 - 2.9)]

3. Management

Majority of the patients are treated with levothyroxine. The principles of treatment include replacement of thyroid hormone as well as treatment of co - existent pituitary hormone deficiencies, in particular glucocorticoid replacement, as administration of levothyroxine in a glucocorticoid - deficient individual may precipitate an adrenal crisis. The dose of levothyroxine required should be tailored to each individual. Children may require higher doses (around 4 mcg/kg daily) to meet the demands of growth and development. Normal infants and children have higher free - thyroid hormone levels. Therefore, higher levothyroxine doses are recommended in this age group. Treatment should be started promptly in order to avoid adverse neurological sequelae associated with hypothyroidism.

The recommended initial dose of T₄ is 10–15 µg/kg per day, or 50 µg daily, for the average term infant weighing 3–4.5 kg. Therapy should be monitored at 4 - to 6 - week intervals during the first 6 months, at 2 - to 3 - month intervals between ages 6 and 24 months and at 3 - to 6 - month intervals thereafter. If the initial diagnosis cannot be established definitively, levothyroxine can be withdrawn for

30 days at age 2–3 years without compromising brain maturation to allow reassessment.

For this patient we have given levothyroxine 50 mcg daily. And ask for follow - up and for umbilical hernia only conservative management was advised.

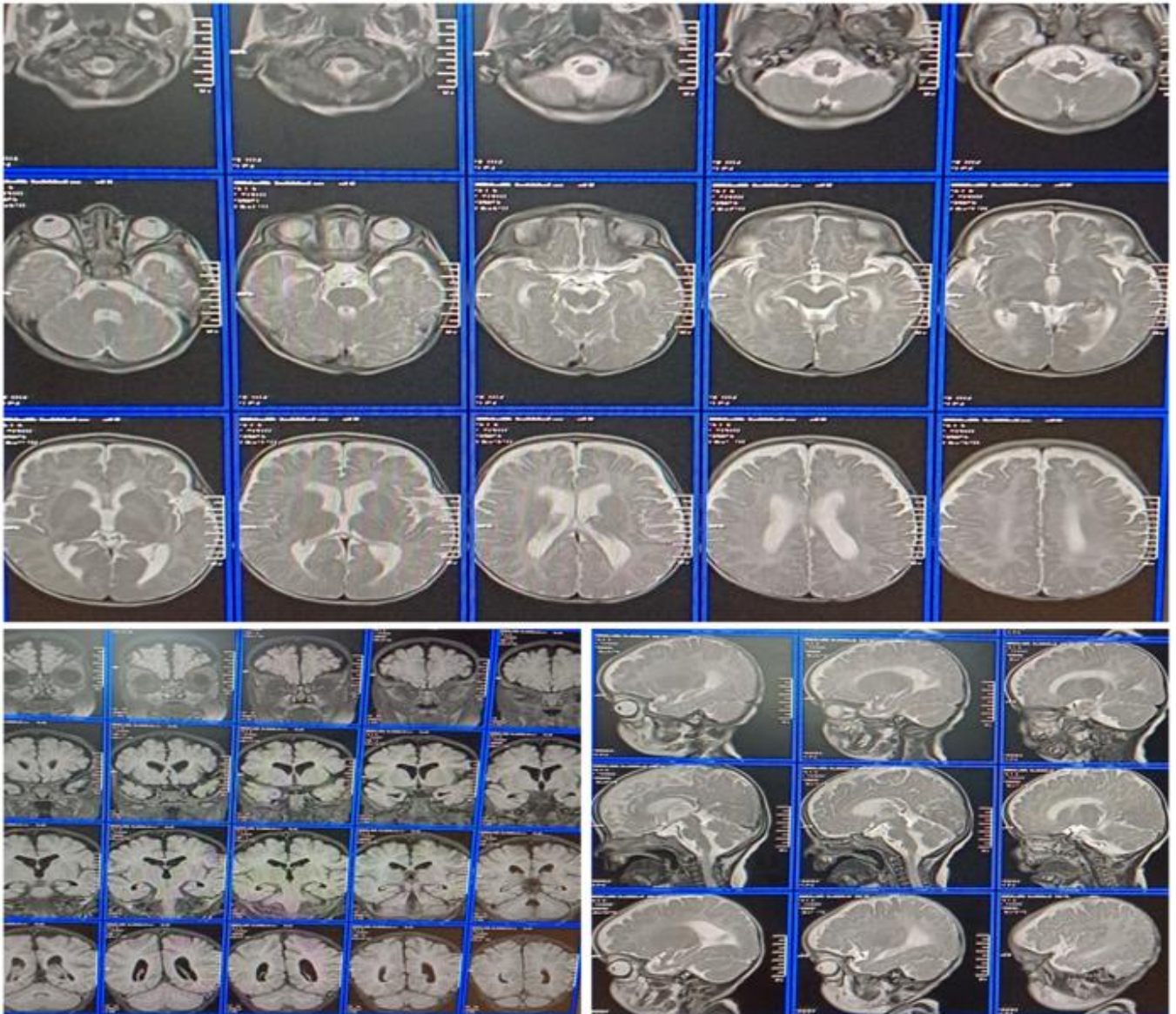
4. Conclusion

Central hypothyroidism is most often caused by diseases of the pituitary or hypothalamus. The diagnosis is suspected by the finding of low FT₄ and inappropriately low, normal or

slightly increased TSH. A delayed TSH response to TRH (TRH stimulation test) supports the diagnosis. In neonates, central hypothyroidism goes undetected as most centers only use TSH evaluation. This can result in a delay in diagnosis and severe hypothyroidism, with mental and skeletal abnormalities. Fatigue can be the only presenting feature of central hypothyroidism in the absence of other pituitary hormone abnormalities. Treatment with levothyroxine is very reassuring, with dramatic improvement in symptoms. Monitoring of adequacy of therapy needs to be followed - up with serial serum T₄ and T₃ levels, with the aim of maintaining them in the upper range of normal



Figure 1: Characteristics Facies of Central Hypothyroidism with Umbilical Hernia



[MRI findings Suggestive of Small Pituitary Gland (Height of 2.5mm) Represent Pituitary Hypoplasia, Mild Prominence of all ventricles]

References

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