Case study

A Unusual case of Congenital Hypothyroidism

Comment [WK1]: It needs to be arranged in terms of working style and according to the spelling guide.

ABSTRACT

Congenital Hypothyroidism is a common preventable cause of mental retardation.The incidence of CH is 1 in 2500 to 1 in 3000 newborns.Most common causes are thyroid dysgenesis and dyshormonogenesis. Some disorder like maternal autoantibodies, maternal intake of anti thyroid medication, iodine deficiency or iodine excess can result in transient CH.

Comment [WK2]: open should be written

Common symptoms include decreased activity and increased sleep, feeding difficulty, constipation, and prolonged jaundice. On examination, common signs include myxedematousfacies, large fontanels, macroglossia, a distended abdomen with umbilical hernia, and hypotonia.

Thyroid dysgenesis accounts for 85% of permanent, primary CH, while inborn errors of thyroid hormone biosynthesis (dyshormonogeneses) account for 10-15% of cases. Secondary or central CH may occur with isolated TSH deficiency, but more commonly it is associated with congenital hypopitiutarism. Transient CH most commonly occurs in preterm infants born

CASE REPORT

A 3 day old baby was admitted to SNCU with chief complain of yellowish discoloration upto abdomen and respiratory distress. The baby was born to 2nd Gravida with history of one abortion at 2 month which was spontaneous and the mother was on antithyroid medication for last 6 month before the delivery. The baby was born at 38 weeks 2days with birth weight 1.75kgs with birth history uneventful. Baby was fed within one hour of life and passed stool within 24hrs and urine within 48hrs.

General physical examination revealed a well alert and active baby with tachypnea and mild retraction and icteric upto abdomen with facial dysmorphism , Quebec score-4.

On Examination their was open posterior fontanelle with facial dysmorphism(upslanting palpebral fissure, depressed nasal bridge, low set ears, no skeletal deformity, no umbilical hernia. Assymetrical IUGR (ponderal index - 1.9). Respiratory examination shows tachypnea with mild intercoastal retraction and mild subcoastal retraction.

So the baby was put on oxygen support and single surface phototherapy for 2days for neonatal jaundice

And on 6th day of admission their was and episode of regurgitation of the feed and feed intolerance was seen so drop domperidone and drop ranitidine was given followed by burping after half feed their were so sign of constipation.

Investigation shows normal blood picture , sepsis profile positive, USG neck shows both thyroid lobes were normal, TORCH- rubella IgG- 104 IU/ml and CMV-93 IU/ml , blood culture and sensitivity

shows MRCONS, CSF analysis was in normal limits, Urine culture and sensitivity was positive for entercoccus , Mother anti TPO antibody - 0.84IU/ml which was in normal limit,

Serum TSH- 10.6, freeT4-1 ng/dl , free T3- 2.2 pg/dl

Dried blood spot test revealed urea cycle defect to be presumptive positive with abnormal amino acid metabolism and decrease in citrulline levels

In view of TSH- 10.6. Tab Levothyroxine was started and The patient was discharged after a hospital stay of 17 days on the attendant's persistent request on Tab Levothyroxine.

A provisional diagnosis of Term/VD/ Assymetrical IUGR/Neonatal Jaundice/ Late onset neonatal sepsis/ Dysmorphic facies/ Urosepsis / Congenital Hypothyroidism/ Quebec score- 4

DISCUSSION

In greater than 95% of newborn infants with CH, there are no symptoms or signs of CH when the diagnosis is suspected by newbornscreening.Congential Hypothyroidism with citrullinemia is one of urea cycle defects and is caused by argininosuccinic acid synthetase deficiency which includes a failure to synthesize urea, resulting in the accumulation of ammonium, glutamine, and citrulline. The neonatal form presents the most severe onset and a fatal course with vomiting, failure to feeding, irritability, apnea, convulsion,lethargy, stupor and coma with respiratory arrest.Althoughcitrullinemia is reported as a rare urea cycle.

CONCLUSION

The deficiency of thyroid hormones in the neonate has been known since antiquity.Though still a prevalent nutritional disease worldwide, iodine deficiency rarely causes congenital hypothyroidism (CH) in western countries. Permanent neurodevelopmental deficits were known to occur when CH was not recognized and adequately treated by 2 to 3 months of postnatal age. Since the advent of newborn screening for CH in 1973, intellectual disability as a consequence of CH has been virtually eradicated among affected infants detected by screening within the first 2-3 weeks of age. The incidence is approximately 1:4,000 in iodine sufficient populations. The etiology in 70 to 80% of the non-familial cases is unknown. Maternal hypothyroidism may adversely affect the fetus to rarely cause findings of CH, or to be associated with mildly decreased IQ outcomes when maternal hypothyroidism occurs during the first half of pregnancy despite normal fetal and neonatal thyroid function. So neonatal screening for thyroid function should be done after 72 hours of life and should repeat Thyroid function test on 15th day of life.

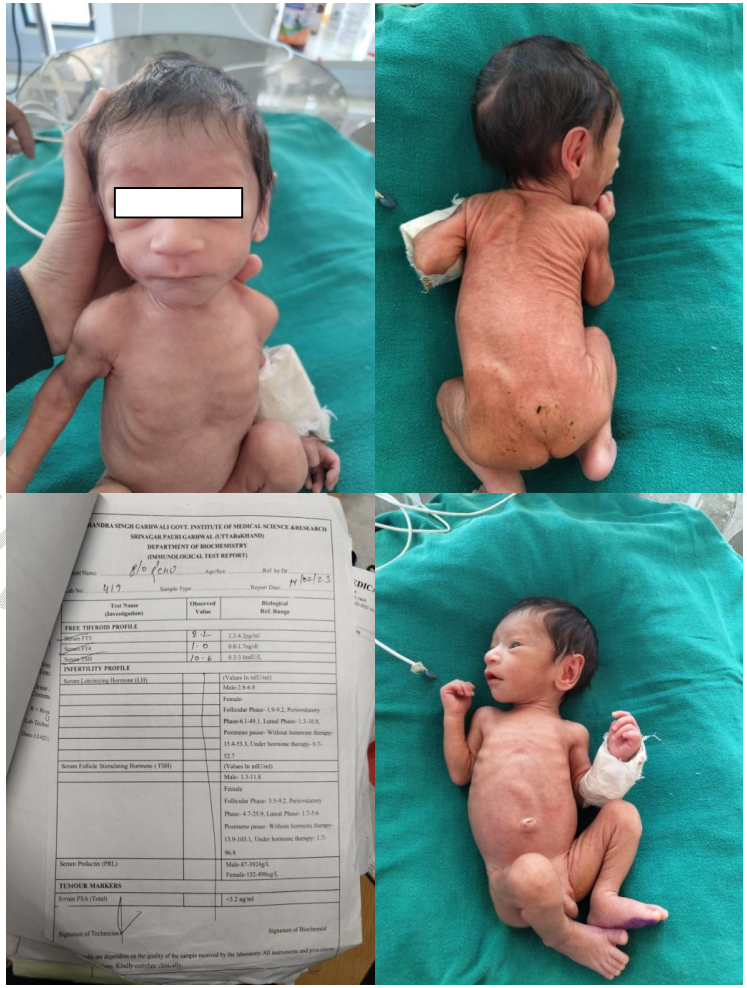
REFERENCE

- American Academy of Pediatrics, Rose SR, Section on Endocrinology and Committee on Genetics, et al: Update of newborn screening and therapy for congenital hypothyroidism, Pediatrics 117:2290–2303, 2006.
- Maestri NE, Clissold DB, Brusilow SW. Long-term survival of patients with argininosuccinatesynthetase deficiency. J Pediatr 1995; 127:929-35.

- Tokatlı A, Çokun T, Özalp .Citrullinemia. Clinical experience with 23 cases. Turk J Pediatr 1998; 40:185-193.
- National institute of health

Summer M, Tuchman M. Proceedings of a consensus conference for the management of patients with urea cycle disorders.Pediatr 2001; 138:S6-S10.
Maestri NE, Hayser ER, Bartholomew D, Brusilow SW. Prospective treatment of urea cycle disorders. J Pediatr 1991; 119:923,8.
La Franchi S. Disorders of the Thyroid Gland. In: Behrman RE, Kleiman RM, Jenson HB (ed). Nelson Textbook of Pediatrics. 16thed. Philadelphia: Saunders 2000; 1698-1702.
Summer M. Current strategies for the management of neonatal urea cycle disorders.Pediatr 2001; 138:S30-S39.
Rezvani I. Defects in Metabolism of Amino Acids. In: Behrman RE, Kleiman RM, Jenson HB (ed). Nelson Textbook of Pediatrics.16th ed. Philadelphia: Saunders 2000; 344-377.
Tokatlı A, Çokun T, Özalp .Citrullinemia. Clinical experience with 23 cases. Turk J Pediatr 1998; 40:185-193.
Tokatlı A, Çokun T, Özalp . Fifteen years' experience with 212 hyperammonaemic cases at a metabolic unit. J InherMetab Dis 1991; 14:698-706.

Fig 1 Congenital Hypothyroidism and pathological report



SRINAGAR PAURI GARHWAL (UTTARAKHAND)
DEPARTMENT OF BIOCHEMISTRY
(IMMUNOLOGICAL TEST REPORT)

Name: B/o Renu Age/Sex: Ref. by Dr.:
Lab No.: 417 Sample Type: Report Date: 14/02/23

Test Name (Investigation)	Observed Value	Biological Ref. Range
FREE THYROID PROFILE		
Serum FT3	3.2	2.2-4.2pg/ml
Serum FT4	1.0	0.8-1.7ng/dl
Serum TSH	10.6	0.3-5.6mIU/L
INFERTILITY PROFILE		
Serum Luteinizing Hormone (LH)		(Values In mIU/ml)
		Male-2.4-6.8
		Female-
		Follicular Phase- 1.9-9.2, Periovulatory
		Phase-6.1-49.1, Luteal Phase- 1.3-10.8,
		Postmeno pause- Without hormone therapy-
		15.4-53.3, Under hormone therapy- 0.7-
		19.7
Serum Follicle Stimulating Hormone (FSH)		(Values In mIU/ml)
		Male- 1.3-11.8
		Female
		Follicular Phase- 3.5-9.2, Periovulatory
		Phase- 4.7-25.9, Luteal Phase- 1.7-5.6
		Postmeno pause- Without hormone therapy-
		13.9-103.1, Under hormone therapy- 1.7-
		96.8
Serum Prolactin (PRL)		Male-87-3924g/L
		Female-132-498ug/L
TUMOUR MARKERS		
Serum PSA (Total)		<3.2 ng/ml

Signature of Technician Signature of Biochemist

...ts are dependent on the quality of the sample received by the laboratory. All instruments and procedures ... Kindly correlate clinically.