

## Case study

### A Unusual case of Congenital Hypothyroidism

#### 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.

Common symptoms include decreased activity and increased sleep, feeding difficulty, constipation, and prolonged jaundice. On examination, common signs include myxedematous facies, large fontanelles, 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 hypopituitarism. 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 there was open posterior fontanelle with facial dysmorphism (upslanting palpebral fissure, depressed nasal bridge, low set ears, no skeletal deformity, no umbilical hernia. Asymmetrical IUGR (ponderal index - 1.9). Respiratory examination shows tachypnea with mild intercostal retraction and mild subcostal retraction.

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

And on 6th day of admission there was an 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 there were no signs 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

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

**Comment [WK2]:** open should be written

shows MRCONS, CSF analysis was in normal limits, Urine culture and sensitivity was positive for enterococcus , 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 newborn screening. Congenital 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. Although citrullinemia 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

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Fig 1 Congenital Hypothyroidism and pathological report

UNDER PEER REVIEW



**Specialty**  
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**LABORATORY INFORMATION**

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**TEST INFORMATION**

TEST NAME: HEMATOLOGY  
 TEST CODE: HEM 100  
 TEST DESCRIPTION: Hematology (CBC) with platelet count and reticulocyte count. Includes hemoglobin, hematocrit, red blood cell count, white blood cell count with differential, and platelet count.

**RESULTS**

TEST NAME	RESULT	REFERENCE RANGE
Hemoglobin (Hb)	14.5 g/dL	13.8 - 15.2 g/dL
Hematocrit (Hct)	45.0 %	41.0 - 47.0 %
Red Blood Cell Count (RBC)	4.80 x 10 <sup>12</sup> /L	4.50 - 5.50 x 10 <sup>12</sup> /L
White Blood Cell Count (WBC)	12.50 x 10 <sup>9</sup> /L	4.00 - 11.00 x 10 <sup>9</sup> /L
Neutrophils	75 %	50 - 70 %
Lymphocytes	15 %	20 - 40 %
Monocytes	5 %	2 - 8 %
Eosinophils	2 %	1 - 5 %
Basophils	3 %	0 - 2 %
Platelet Count	150,000 /mm <sup>3</sup>	150,000 - 400,000 /mm <sup>3</sup>
Reticulocyte Count	0.5 %	0.1 - 1.5 %

**LABORATORY COMMENTS:** Hematology results for the patient listed above. Hemoglobin and hematocrit are within normal limits. White blood cell count is elevated, consistent with a leukocytosis. The differential shows a neutrophilic leukocytosis with a left shift. Platelet count is within normal limits. Reticulocyte count is slightly elevated.

**TEST INFORMATION:** Hematology (CBC) with platelet count and reticulocyte count. Includes hemoglobin, hematocrit, red blood cell count, white blood cell count with differential, and platelet count.

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**TEST INFORMATION**

TEST NAME: CHEMISTRY  
 TEST CODE: CHEM 100  
 TEST DESCRIPTION: Chemistry (Basic) including glucose, urea nitrogen, creatinine, total protein, albumin, total bilirubin, and aspartate aminotransferase (AST).

**RESULTS**

TEST NAME	RESULT	REFERENCE RANGE
Glucose	100 mg/dL	70 - 100 mg/dL
Urea Nitrogen (BUN)	12 mg/dL	7 - 20 mg/dL
Creatinine	1.2 mg/dL	0.7 - 1.3 mg/dL
Total Protein	7.5 g/dL	6.0 - 8.0 g/dL
Albumin	4.5 g/dL	3.5 - 5.0 g/dL
Total Bilirubin	1.2 mg/dL	0.1 - 1.2 mg/dL
Aspartate Aminotransferase (AST)	25 U/L	0 - 37 U/L

**LABORATORY COMMENTS:** Chemistry results for the patient listed above. Glucose is within normal limits. Urea nitrogen and creatinine are within normal limits. Total protein and albumin are within normal limits. Total bilirubin is slightly elevated. Aspartate aminotransferase (AST) is within normal limits.

**TEST INFORMATION:** Chemistry (Basic) including glucose, urea nitrogen, creatinine, total protein, albumin, total bilirubin, and aspartate aminotransferase (AST).

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**TEST INFORMATION**

TEST NAME: URINALYSIS  
 TEST CODE: URIN 100  
 TEST DESCRIPTION: Urinalysis including color, appearance, pH, specific gravity, glucose, ketones, bilirubin, urobilinogen, heme, and leukocytes.

**RESULTS**

TEST NAME	RESULT	REFERENCE RANGE
Color	Yellow	Yellow
Appearance	Clear	Clear
pH	5.0	5.0 - 8.0
Specific Gravity	1.020	1.010 - 1.030
Glucose	None	None
Ketones	None	None
Bilirubin	None	None
Urobilinogen	None	None
Heme	None	None
Leukocytes	None	None

**LABORATORY COMMENTS:** Urinalysis results for the patient listed above. Urine is yellow and clear. pH is 5.0, specific gravity is 1.020. No glucose, ketones, bilirubin, urobilinogen, heme, or leukocytes are present.

**TEST INFORMATION:** Urinalysis including color, appearance, pH, specific gravity, glucose, ketones, bilirubin, urobilinogen, heme, and leukocytes.

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**TEST INFORMATION**

TEST NAME: COAGULATION  
 TEST CODE: COAG 100  
 TEST DESCRIPTION: Coagulation studies including prothrombin time (PT), international normalized ratio (INR), and partial thromboplastin time (PTT).

**RESULTS**

TEST NAME	RESULT	REFERENCE RANGE
Prothrombin Time (PT)	13.5 sec	11.0 - 13.5 sec
International Normalized Ratio (INR)	1.1	0.8 - 1.2
Partial Thromboplastin Time (PTT)	32.0 sec	28.0 - 35.0 sec

**LABORATORY COMMENTS:** Coagulation results for the patient listed above. Prothrombin time (PT) is 13.5 sec, INR is 1.1, and partial thromboplastin time (PTT) is 32.0 sec. All values are within normal limits.

**TEST INFORMATION:** Coagulation studies including prothrombin time (PT), international normalized ratio (INR), and partial thromboplastin time (PTT).

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