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Congenital hypothyroidism presenting with severe neonatal anaemia.

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Abstract

A 26 days old female neonate presented with abdominal distension, hoarse cry, difficulty in feeding since birth, and progressive paleness noticed over one week before her mother sought medical advice. On examination she was very pale with clinical features suggestive of congenital hypothyroidism. Conducted investigations revealed congenital hypothyroidism with severe macrocytic normochromic anaemia. The neonate improved with packed red cell transfusion and levothyroxine replacement therapy.

Keywords: Severe anaemia, Congenital hypothyroidism, Neonatal anaemia.

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Introduction

Congenital hypothyroidism (CH) occurs in approximately 1 in 4,000 newborns worldwide and is one of the most common causes of preventable mental retardation in children [1]. Early identification by neonatal screening tests and early initiation of levothyroxine replacement, preferably within the first two weeks of life, are crucial in preventing cognitive and neuro-motor impairment in affected children [2]. The clinical manifestations of CH in neonatal period can be variable and include temperature instability, lethargy, hoarse cry, dry skin, goitrous swelling of neck, persistent posterior fontanel, prolonged jaundice, umbilical hernia, and delayed appearance of upper epiphysis of the tibia. Coarse facies, macroglossia, constipation, and short trunk may become evident either during or beyond the neonatal period. These features may be subtle or may not be present at all. Therefore, it would not be wise to wait until typical clinical features develop to diagnose congenital hypothyroidism. Anaemia is a common finding in CH and the severity of anaemia depends on the degree of hypothyroidism [3]. However, CH presenting with severe anaemia in the neonatal period has not been commonly described to the best of our knowledge. We report a neonate who was not offered screening for CH at birth and was brought to our attention on day 26 of life with established hypothyroidism and severe anaemia requiring transfusion.

Case Report

A 26 days old female baby was brought to our attention with complaints of difficulty in feeding and hoarse cry since birth and progressive paleness noticed over one week with abdominal distension for 5 days. There was no history of fever, vomiting, or respiratory distress. The baby was delivered at term by spontaneous vaginal delivery at another hospital and was of appropriate weight for gestational age (3.28 kg). There was no history of delayed passage of meconium or prolonged jaundice. There was no history of any maternal illness complicating pregnancy. There was no family history suggestive of haemolytic anaemia or blood dyscrasias.

On examination baby weighed 3.5 kg, her length was 56 cm. She was severely pale with a wide open and flat anterior fontanel (3 × 3 cm), open posterior fontanel, depressed nasal bridge, macroglossia, dry skin, and an umbilical hernia measuring 2 × 3 cm. There was no icterus, cephalhaematoma, or hepatosplenomegaly. Lab evaluation revealed decreased free tetraiodothyronine (FT_4), elevated thyroid stimulating hormone, severe hypoproliferative anaemia with a Hb of 4.5 gm/dL and Reticulocyte Production Index (RPI) of 0.22 **[Table 1].** Considering the craniofacial features, clinical

Table I. *Pre and post treatment investigations in the reported neonate with CH.*

Lab parameters	Pre treatment	Post treatment
Haemoglobin	4.5 g/dl	8.1 gm/dl
PCV	16.4	23.8
MCV	105	81.1
MCH	30	27.6
MCHC	28.5	34
Reticulocyte count	1.5%	2
Reticulocyte Production Index (RPI)	0.22	0.41
FT_4	0.152 ng/dl	2.65 ng/dl
TSH	100 micro IU/ml	8.42 micro IU/L

evaluation, and the results of the performed investigations, a provisional diagnosis of congenital hypothyroidism was made and therapy with Levothyroxine was started at 15 microgram/kg/day. Neck Ultrasound showed no evidence of thyroid gland in the neck. X-ray knee showed stippled epiphysis of the femur. Packed red blood cells (PRBC) was transfused to correct severe anaemia. One month later, the baby was reviewed and was noted to have social smile, improved facial and cutaneous features, decreased hoarseness of voice. Haemoglobin and RPI improved to 8.1 gm/dl and 0.41 respectively with TSH falling to 8.42 micro IU/ml **[Table 1].** She started to have adequate feeding and gained weight appropriately.

Discussion

CH presenting with severe anaemia in the neonatal period is rare. Anaemia due to hypothyroidism is usually macrocytic or normocytic and is associated with decreased height/ length [4]. Anaemia in CH has been believed to be due to defective erythropoietin action or production. In vitro studies have shown that thyroxine is necessary for potentiation of erythropoietin action on erythroid colony formation [5]. Moreover, decreased tissue oxygen requirements due to decreased basal metabolic rate as a consequence of hypothyroidism may itself lead to physiological adaptation which in turn leads to decreased erythropoietin production and consequent decreased erythropoiesis [6]. There is an evidence of hypo proliferative erythropoiesis in CH based on ferro-kinetic studies [7].

A study on the haematological parameters of hypothyroid and hyperthyroid subjects, Kawa MP et al from Poland (2010) demonstrated that thyroid hormone deficiency resulted in decreased blood counts and clonogenic potential of Erythroid Burst Forming Units (BFU-E) ultimately leading to decreased erythropoiesis [8]. In the currently reported neonate, the haemoglobin improved with ten millilitres per kilogram packed red cell transfusion and levothyroxine supplementation without requiring the need for other nutritional supplementation indicating the role of levothyroxine in restoring normal erythropoiesis. The reason for severe anaemia could be attributable to severe hypothyroidism as reflected by TSH values and failure to demonstrate thyroid gland by ultrasonography indicating the possibility of athyreosis. A thyroid scintigraphy was planned but could not be done at our institute to document ectopic thyroid due to non availability.

The neonate described in this report had not been offered screening for CH at the time of birth or before discharge from the place of birth which resulted in presentation to our hospital in the late neonatal period with manifest hypothyroidism and severe anaemia as a complication. Such incidents are being regularly reported and have not resulted in significant change in medical care practices in our country. There are instances where a diagnosis of CH had been made beyond five years of age and even during adolescence and adulthood [9,10]. New born screening for CH has been in vogue for more than 35 years worldwide and yet it is grossly underutilized in our country citing lack of resources. The central government recently launched its flagship program, the Rashtriya Bal Swasthya Karyakram (RBSK) which was launched to combat four "D" s, -defects, diseases, deficiencies, and developmental delays by incorporating universal screening for the above conditions. We urge the RBSK program managers to implement universal and free screening for CH for all newborns so as to safeguard the mental well being and productivity of future citizens of India.

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