Rare Presentations of Congenital Hypothyroidism

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Congenital hypothyroidism is mostly asymptomatic at birth. It is usually detected by targeted neonatal screening for CH that is performed after birth. Nonetheless, there are reports of rare and vague presentations of CH. These include protracted icterus, large tongue, abdominal distension, skin mottling, muscle hypotonia, prolonged gestation, large posterior fontanel, respiratory distress, hypothermia, peripheral cyanosis, hypoactivity, poor feeding, lag in onset of stooling, abdominal distension with vomiting, edema [1,2] and chylothorax [3]. We describe here two newborn infants with rare clinical presentations that could be attributed to CH. The crucial role of neonatal screening in the diagnosis of CH is highlighted.

PATIENT DESCRIPTIONS

PATIENT 1

A 2550 g term female infant, born to a group B Streptococcus-carrier mother, had abdominal distension at discharge examination [Figure A]. The infant nursed well without vomiting and passed meconium. There was no organomegaly or palpable abdominal masses. Sepsis workup was performed and empiric ampicillin and cefotaxime were started.

CH = congenital hypothyroidism

A decompressive nasogastric tube was inserted and intravenous fluids were initiated. Abdominal radiograph was normal except for mild bowel distension [Figure B]. Sepsis workup was negative. Abdominal ultrasonography was normal. On day 4 of life, the Israeli National Neonatal Screening Program reported a high thyroid-stimulating hormone level (> 400 mU/L). Venous sample of free thryoxine 2.4 pmol/L (normal 12–22) and TSH > 100 mU/L (normal < 10 mU/L) were compatible with CH. Thyroid hormone supplementation was started and 4 days later abdominal distension subsided; the infant nursed well and was discharged home. Follow-up 2 weeks later revealed a healthy infant with normal FT4 and TSH.

PATIENT 2

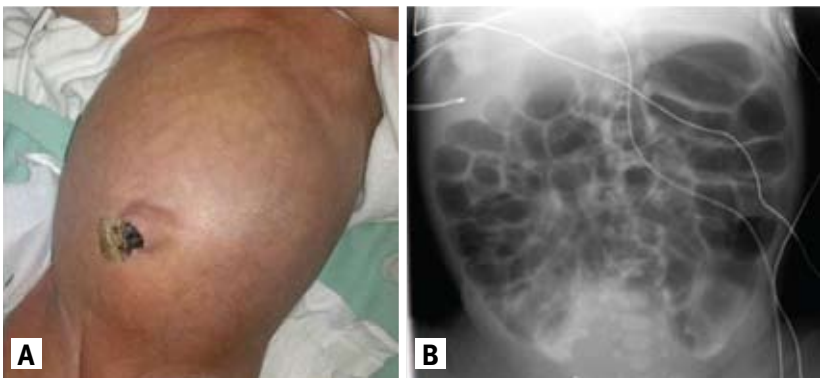
A term female infant weighing 4110 g presented with a systolic murmur and barely palpable pulses. Blood pressure (four limbs) was normal. Echocardiography was normal except for patent foramen ovale. The infant was asymptomatic but pulses remained weak. At day 4, the Israeli National Neonatal Screening Program reported a high TSH level (> 400 mU/L). The venous sample of FT4 = 2 pmol/L and TSH > 100 mU/L was compatible with CH. Thyroid hormone supplementation was started and 10 days later the pulses became normal and were easily palpable.

TSH = thyroid-stimulating hormone
FT4 = free thyroxine

COMMENT

In this report we wish to raise the awareness of neonatologists regarding uncommon clinical presentations of CH, where the clinical signs disappeared following thyroid hormone supplementation. In the first patient, abdominal distension was likely the sole sign of CH and was apparently caused by decreased intestinal motility often observed with CH [2]. In the second patient, weak pulses could have been due to CH which has been associated with decreased beat-to-beat variability of fetal heart rate [4] and with decreased left ventricular function [5].

Abdominal radiographs: **[A]** Significant bowel distension at discharge examination (day 2), **[B]** Mild bowel distension (day 4).



These cases also demonstrate the crucial role of neonatal screening, which allowed early and timely treatment of CH.

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