CASE REPORT

Congenital hypothyroidism presenting as pseudo-obstruction in preterm infants

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SUMMARY

Congenital hypothyroidism (CH) presenting as acute pseudo-obstruction is uncommon. We report two premature infants presenting with acute bowel obstruction subsequently diagnosed to have CH. Both responded well to medical management with thyroid supplementation.

BACKGROUND

Congenital hypothyroidism (CH) is a well-known cause of preventable morbidity in children. Common clinical presentations in the newborn period include prolonged jaundice, respiratory distress, hypothermia, hypoactivity, poor feeding, delayed passage of meconium, vomiting and abdominal distension.¹ Symptoms are often absent soon after birth due to residual hormone from the mother. Presentation as intestinal pseudo-obstruction in the neonatal period is uncommon.

CASE PRESENTATION

Case 1

A female infant was born at 36 weeks gestation to a second gravida mother by a normal vaginal delivery following an uneventful pregnancy. She was born in good condition with a birth weight of 2.3 kg (25th centile). She developed bilious vomiting along with abdominal distension after starting enteral feeds on day 1. Upper gastrointestinal (GI) obstruction was suspected and a contrast study (barium meal) performed on day 2 of life was consistent with malrotation (figure 1). On day 3 she had an exploratory laparotomy; however, no evidence of malrotation was detected and the abdomen was closed without any intervention. Postoperatively, she continued to have persistent bilious aspirates. Further, medical workup revealed her thyroid-stimulating hormone (TSH) level was >500 mU/L with undetectable levels of T4. Intravenous leothyronine was started at a dose of 25 µg once daily and within 3 days she started tolerating full enteral feeds. Repeat thyroid function test a week later was normal. Ultrasound scan of the neck was suspicious of thyroid agenesis and this was confirmed by the nuclear scan performed 2 weeks after birth (figure 2).

Case 2

A preterm male infant born at 30⁺6 weeks gestation by emergency caesarean section to a second gravida mother with a birth weight of 1150g (25th centile). At birth, he was intubated, given exogenous surfactant and mechanically ventilated for respiratory distress. He was extubated to bilevel

positive airway pressure (BIPAP) at 6 h of age but on day 2 he developed hypotension, increasing respiratory distress and pulmonary haemorrhage and was reintubated. Dopamine infusion was started along with antibiotics for presumed sepsis. He deteriorated acutely on day 6, with vomiting, abdominal distension, temperature instability, deranged clotting profile and a drop in the haemoglobin level from 12 to 7g/dL. He was actively resuscitated with fresh frozen plasma, packed red blood cells and crystalloids. Serial abdominal radiographs showed persistent gaseous distension of bowel loops with thickened bowel walls (figure 3) and antibiotic cover was widened with metronidazole for possibility of necrotising enterocolitis. A week later he continued to have abdominal distension and nasogastric aspirates were still bilious. Upper GI contrast study after clinical improvement did not show evidence of malrotation. Owing to signs suggestive of pseudo-obstruction, he was investigated for thyroid disorders and cystic fibrosis on day 15. He was negative to a standard panel of cystic fibrosis mutations; however, his TSH was

Figure 1 Barium meal showing duodenojejunal flexure which lies low, approximately one vertebral body lower than the pylorus and just to the left of the left pedicle, with proximal loops of jejunum lying centrally, suggestive of malrotation.

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Figure 2 Nuclear medicine thyroid scan shows no evidence of thyroid tissue.

>100 mU/L and free T4 <5.2pmol/L, suggestive of CH. Intravenous leothyronine was started and his abdominal distension improved and was tolerating feeds 2 days later. Neck ultrasound suggested absence of any thyroid tissue. Unfortunately, newborn screening test was not performed in the first two weeks of life.

OUTCOME AND FOLLOW-UP

Both of them had regular follow-up postdischarge and at 6 months still continued on thyroid replacement therapy and are thriving well with age-appropriate milestones.

Figure 3 Abdominal X-ray anteroposterior view shows significantly dilated bowel loops suggestive of intestinal obstruction.

DISCUSSION

CH presenting as functional bowel obstruction/pseudo-obstruction is uncommon. As far as we are aware there are two reported cases of preterm infants with hypothyroidism presenting with features suggestive of intestinal obstruction.^{2 3} Both infants presented with abdominal distension and bilious emesis diagnosed secondary to hypothyroidism. Our patients were both premature and also presented with persistent abdominal distension and bilious aspirates, which resolved once thyroid hormone therapy was initiated.

CH occurs 1 in 3000 to 4000 live-births, 85% of which are due to thyroid dysgenesis and 15% due to inborn errors of thyroid metabolism.⁴ Thyroid hormone is important for the maturation of many target tissues and key for brain and skeletal development. CH may not present clinically in the immediate newborn period due to transplacental passage of some maternal thyroid hormone and some residual thyroid function as with thyroid hypoplasia, dyshormonogenesis and ectopic thyroid.⁵ The classical signs and symptoms of the disease are only recognisable at an advanced stage while at early stages the clinical signs are subtle. With the introduction of newborn screening programme over the past three decades, majority of infants with CH are diagnosed, and treatment initiated at an early presymptomatic stage, enabling normal mental development. However, diagnosing CH in small preterm infants with screening tests may prove challenging due to several reasons. Full-term infants have a normal physiological rise in TSH in the early postnatal period which causes a surge in T4 and T3 levels between day 1 with a peak at 6 h and remains elevated till day 7 of postnatal life. In contrast, preterm infants have a relatively immature hypothalamic–pituitary–thyroid axis and some preterm infants with CH develop a late rise in TSH levels.⁵ In addition, concurrent administration of dopamine and glucocorticoids in sick premature infants could lower the levels of TSH by suppression of the thyrotropes in the anterior pituitary gland, or by their action on the hypothalamus.⁶ Infants with central hypothyroidism which is due to disorders of pituitary or hypothalamus can have inappropriately low levels of TSH despite low T4 levels. Many newborn screening tests only measure TSH levels and not combined levels of T4 and TSH and it is possible that central hypothyroidism may be missed with current screening tests.⁵ However, these conditions are extremely rare in neonates.

Learning points

- ▶ The purpose of this report is to stress that despite availability of neonatal screening tests, awareness of this serious but preventable disease is essential.
- ▶ Diagnosing congenital hypothyroidism (CH) in preterm infants with thyroid-stimulating hormone based screening tests can be difficult and it is important to repeat the screening tests pre-discharge or on day 28 postnatal age.
- ▶ CH must be strongly suspected as a possible differential diagnosis in apparent bowel obstruction in neonates, if an obvious cause of mechanical bowel obstruction is absent.

Hypothyroidism has been reported in preterm infants at the repeat screening after 36 weeks gestation despite normal screening at 5–8 days after birth.⁷ The relatively low levels of TSH in preterm infants, combined with the effect of medications especially in very sick preterms, necessitate the need to repeat the screening test later.⁵ Our case 2 was extremely unwell and unfortunately newborn screening was not performed. Current newborn screening guidelines in the UK advise repeat screening tests for preterm babies less than 32 weeks gestation at 28 days postnatal age, or the date of discharge home, whichever is the sooner.

Low thyroid hormone levels affect GI motility and increase intestinal transit time. Several mechanisms including deceleration in myoelectric activity, autonomic neuropathy, intestinal oedema, ischaemia, reduction in adrenergic receptors have been implicated.⁸ In adults hypothyroidism has been reported to present as ileus and pseudo-obstruction although the exact incidence of this presentation is unknown.⁹ Bilious vomiting in a preterm infant must be investigated thoroughly and could be

due to intestinal obstruction, sepsis, electrolyte imbalance, etc. Intestinal obstruction is usually ruled out by contrast studies. Our patient 1 incidentally had a false positive radiological diagnosis of malrotation which is reported to occur in 1–2% of infants.¹⁰

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Competing interests None.

Patient consent Obtained.

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