CASE REPORT

Neck mass: an obstructive cause of respiratory distress with medical management

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SUMMARY

Congenital goitre is a known cause of hypothyroidism in newborn. Congenital goitre can be due to defective synthesis of thyroxine or administration of antithyroid drugs to the mother during pregnancy. In this case report, we report an instance of a preterm male infant with antenatally detected goitre presenting as a neck mass with congenital hypothyroidism. Hormonal replacement therapy was started immediately after birth which lead to resolution of the mass and normalisation of thyroid function.

BACKGROUND

Neonatal goitre is not a common entity, but it is an important finding as it can lead to obstruction of airway and need for resuscitation in labour room. Furthermore, the resulting thyroid dysfunction is an important cause of preventable mental impairment. Timely diagnosis and intervention in the form of L-thyroxine can prevent morbidity in later life.

CASE PRESENTATION

A 24-year-old G3P0110 mother had a normal antenatal course until the seventh month of gestation. During the routine ultrasonography (USG) at 28 weeks of gestation, it was noticed that the fetus had a highly vascular, soft, bilobed mass in the anterior aspect of the neck, with size of right lobe 10.78 mL and left lobe 14.63 mL. The location, consistency and bilobed appearance of the mass were suggestive of fetal goitre. There were no features suggestive of polyhydramnios. The mother was clinically euthyroid and there was no family history of thyroid disease. There was also no history suggestive of iodine deficiency or excess. Maternal thyroid function tests (thyroid-stimulating hormone (TSH): 3 mIU/L, antibodies 21 IU/mL (normal thyroid

Figure 2 Regression of neck mass after initiation of therapy with L-thyroxine.

peroxidase antibody: <35 IU/mL) and USG (volume –12 mL) of the neck were normal. The mother went into spontaneous labour at 33 weeks. Expecting a difficult airway, laryngeal mask airway was arranged in the delivery room besides other routine equipment. An emergency lower-segment caesarean section was done for non-progression of labour. The infant's birth weight was 2.368 kg, with Apgar scores of 8 and 8 at 1 and 5 min, respectively. There was a mass in the anterior part of the neck (figure 1). On palpation, the mass was soft in consistency, mobile and non-cystic, and there were no signs of inflammation and tenderness. Furthermore, no bruit was audible. The remainder of the examination was unremarkable. The infant had mild respiratory distress at birth for which oxygen by hood was given for 12 hours. An USG of the neck

Figure 1 Goitre present as midline mass at birth in a neonate.

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was performed and was suggestive of a marked diffuse enlargement of the thyroid gland, with right lobe of 21×23×36 mm (17.4 mL) and left lobe of 27×26×38 mm (26.7 mL); echotexture was normal. The thyroid profile findings of low serum free thyroxine (FT4) of 6.13 pmol/L (normal value: 12±22 pmol/L) and high serum TSH level of >37.69 mIU/L (normal value: 0.4±10 mIU/L) confirmed the diagnosis of primary hypothyroidism. Hormonal replacement treatment with L-thyroxine (15 µg/kg/day) was immediately started on day 2. The mother was asked to avoid giving concomitant soy formula, iron and calcium and vitamin D preparation. The baby was started on feeds on day 2 and was on full feeds by day 8 of life. By day 7 of life, there was reduction in goitre size on physical examination which was confirmed by repeat USG, which showed right lobe size reduced to 21×19×29 mm (11.6 mL) and left lobe 17×20×25 mm (8.5 mL). Repeat thyroid profile on day 14 revealed TSH value of 11.64 and FT4 of 7.247; therefore, dose of L-thyroxine was increased to 20 µg/kg/day. The child was discharged on the 21st day of life with thyroid gland barely visible on inspection (figure 2). He was continued on L-thyroxine and is under regular follow-up.

INVESTIGATIONS

- ▶ USG of the neck: diffuse enlargement of the thyroid gland, right lobe 21×23×36 mm (17.4 mL) and left lobe 27×26×38 mm (26.7 mL; normal thyroid gland volume 0.9±0.2 mL).
- ▶ Thyroid profile: FT4=6.13 pmol/L (normal FT4 12±22 pmol/L), TSH=37.69 mIU/L (normal TSH 0.4±10 mIU/L).
- ▶ Repeat USG of the neck on day 14: right lobe 21×19×29 mm (11.6 mL) and left lobe 17×20×25 mm (8.5 mL).
- ▶ Repeat thyroid profile on day 14: TSH 11.64 mIU/L and FT4 6.247 pmol/L.

DIFFERENTIAL DIAGNOSIS

1. Cystic hygroma.
2. Haematoma.
3. Thyroglossal cyst.
4. Lymphangioma.
5. Lymphadenopathy.

TREATMENT

L-thyroxine (20 µg/kg/day).

OUTCOME AND FOLLOW-UP

The infant is under regular follow-up. At 5 months of age, the infant is thriving well with weight of 4.8 kg and has a normal neurological examination and evoked auditory brainstem response (screening auditory brainstem response was abnormal). Repeat thyroid profile is also normal. The plan is to do thyroid function as per the American academy of Pediatrics (AAP) guidelines, monitoring for T4 and TSH values every 1–2 months for up to 6 months, every 3–4 months from 6 to 12 months and every 6–12 months from 3 years to completion of the growth. Re-evaluation is also scheduled on follow-up to delineate the cause of hypothyroidism.

DISCUSSION

Neonatal goitre presents as a neck mass at birth or shortly thereafter. Congenital goitre can be because of multiple reasons which

include defective iodine uptake and transport, defective thyroglobulin synthesis, transplacental passage of thyroid antibodies, maternal ingestion of antithyroid and tumour.^{1 2} In our case, dysmorphogenesis was thought to be the most reasonable justification of the hypothyroidism. Goitre most commonly presents as firm, non-tender, bumpy mass noted at birth or incidentally thereafter by a parent.³ Even though majority of the infants with goitre are euthyroid, it may present with both hypothyroidism and hyperthyroidism.⁴ The hearing screen was also done and was reported to be normal. A similar case of congenital goitre presenting with hypothyroidism was reported from Portugal and Karnataka in India.^{5 6} The long-term goals are to achieve normal growth and development, by restoring the serum T4 concentration to normal as soon as possible immediately after birth. In our case, we started hormonal replacement therapy within 48 hours of life much earlier than reported in previously reported case reports. Re-evaluation is a must in cases where diagnosis is not clear. According to the European guidelines, re-evaluation is indicated when there is a 'normally located gland, with or without goitre, in neonates with positive thyroid antibodies, in children who have required no increase in L-T4 dose since infancy, and in children in whom no enzyme defect has been identified'.⁷

Learning points

- ▶ Neonatal goitre presents as midline mass and can present as difficult airway at birth.
- ▶ Laryngeal mask airway is an alternative to intubation in cases of difficult airway.
- ▶ Neonatal hypothyroidism is a preventable cause of neurodevelopmental impairment.
- ▶ Neonatal goitre can present with hypothyroidism, hyperthyroidism or normal thyroid function.

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REFERENCES

- 1 Nicola JP, Nazar M, Serrano-Nascimento C, *et al.* Iodide transport defect: functional characterization of a novel mutation in the Na⁺/I⁻ symporter 5'-untranslated region in a patient with congenital hypothyroidism. *J Clin Endocrinol Metab* 2011;96:E1100–E1107.
- 2 Peteiro-Gonzalez D, Lee J, Rodriguez-Fontan J, *et al.* New insights into thyroglobulin pathophysiology revealed by the study of a family with congenital goiter. *J Clin Endocrinol Metab* 2010;95:3522–6.
- 3 de Vries L, Bulvik S, Phillip M. Chronic autoimmune thyroiditis in children and adolescents: at presentation and during long-term follow-up. *Arch Dis Child* 2009;94:33–7.
- 4 Kurtoğlu S, Özdemir A. Fetal neonatal hyperthyroidism: diagnostic and therapeutic approach. *Turk Pediatri Ars* 2017;52:1–9.
- 5 Kim MJ, Chae YH, Park SY, *et al.* Intra-amniotic thyroxine to treat fetal goiter. *Obstet Gynecol Sci* 2016;59:66–70.
- 6 Pereira RC, Barroso LM, Mendes MJ, *et al.* A newborn with neck mass. *Einstein* 2011;9:78–80.
- 7 Léger J, Olivieri A, Donaldson M, *et al.* European society for paediatric endocrinology consensus guidelines on screening, diagnosis, and management of congenital hypothyroidism. *J Clin Endocrinol Metab* 2014;99:363–84.

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