Goiter In A Newborn With Congenital Hypothyroidism

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INTRODUCTION

- Congenital hypothyroidism occurs in 1:2-4,000 births; over 95% present without clinical manifestations and rarely with goiter (0.1-0.3:10,000 births)[1]
- Congenital goiter can be caused by a variety of disorders including thyroid dysgenesis, inborn errors of thyroid hormone metabolism or peripheral resistance, defects in the hypothalamic-pituitary axis, transplacental transfer of maternal antibodies, or iodine imbalance[2]
- Complications of congenital goiter include airway or esophageal compromise; there are no established guidelines for the diagnosis and management of congenital goiter in the perinatal period

CLINICAL COURSE

Prenatal	28 Weeks	• Ultrasound with 6.0 x 3.4 x 5.8 cm midline neck mass; growth restriction without evidence of polyhydramnios • Cordocentesis with elevated TSH (361 µIU/mL)
Birth	38 Weeks	• Planned Caesarean section • Intubated with size 2.5 endotracheal tube for respiratory distress refractory to pressure support
Postnatal	Day 1	• IV levothyroxine initiated (37.5 mcg) • Laboratory evaluation for relevant thyroid antibodies was unremarkable
	Day 17	• Extubated to nasal cannula • Weaned to room air over 4 days
	Day 46	• Discharged home • Enteral access for feeds

RESULTS AND CONCLUSIONS

- Our patient demonstrated a dramatic decrease in goiter size over a 6-week period (Right lobe: 57 to 16cc; Left lobe: 35 to 12cc)
- Thyroid function tests and thyroid antibody tests should be performed depending on prenatal maternal evaluation for hypothyroidism or iodine deficiency; these tests were negative in our patient
- In our patient, genetic testing was notable for a variant of unknown significance in the gene encoding for thyroid peroxidase (*TPO*)
- Serial color-doppler ultrasonography is a useful tool for monitoring size change and response to therapy[3]
- Infants with congenital hypothyroidism must be treated with exogenous thyroid hormone to ensure appropriate neurodevelopmental growth
- Surgical intervention for congenital should only be offered if there is progressive compressive symptoms with severe functional impairments

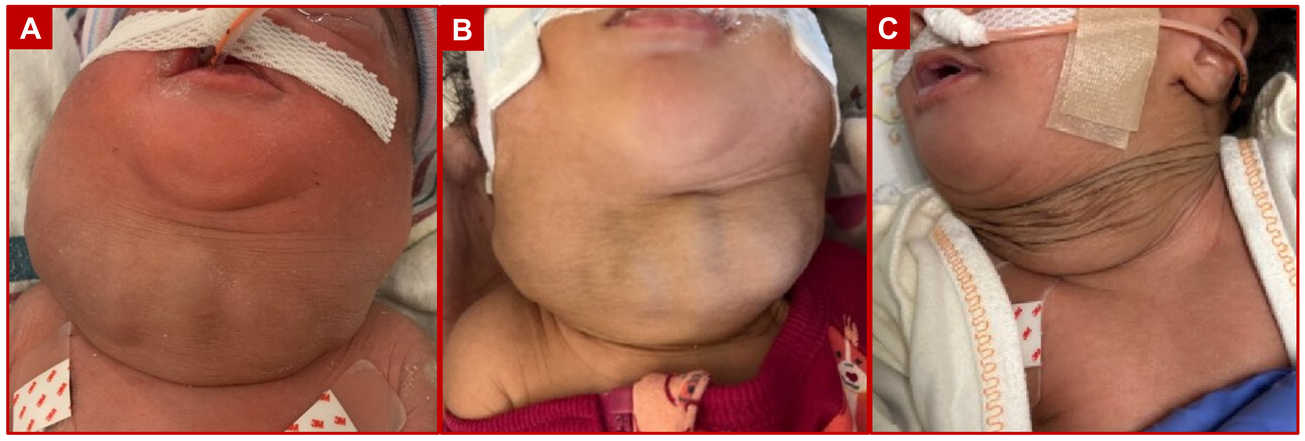

Figure 1. Representative photos of goiter at day-of life 1 (**A**), day-of life 13 (**B**) and day-of life 23 (**C**).

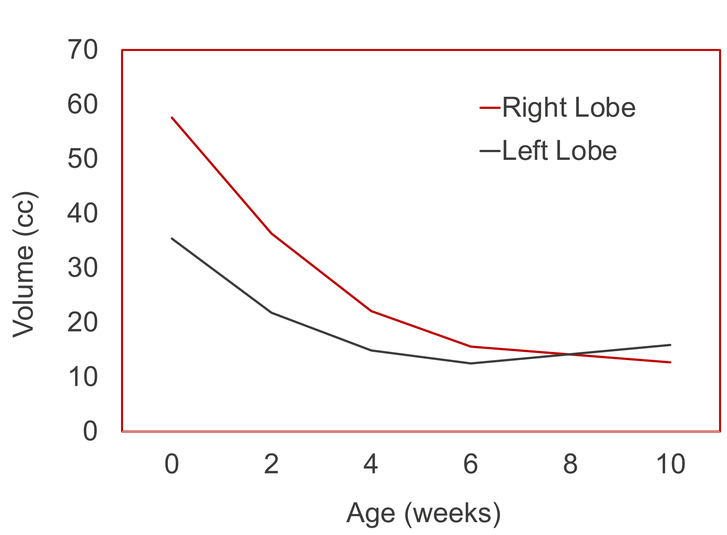

Figure 2. Thyroid lobe volume measured by ultrasound

Age (weeks)	Levothyroxine (mcg)	TSH (µIU/mL)	Free T4 (ng/dL)
0	37.5	*16.7 (H)*	0.67
2	25.0	0.919	1.19
4	12.5	0.149	1.57
6	12.5	0.719	1.17
8	12.5	0.157	1.53
10	12.5	1.296	1.19

Table 1. Relationship between age and levothyroxine dose with relevant laboratory markers of thyroid function

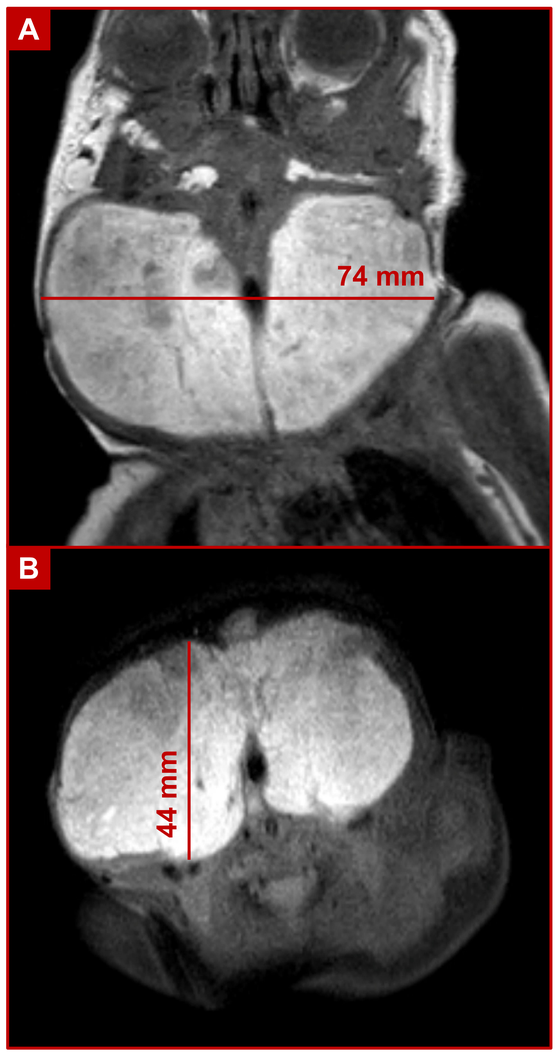

Figure 3. T1-weighted MRI in coronal (**A**) and axial (**B**) section; hyperintensity consistent with thyroid parenchyma

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3. Perry KD, Hope J, Yang J. Dyshormonogenetic goiter-like changes in a child with congenital hypothyroidism and a euthyroid adult. *Diagnostic Cytopathology*. 2013;41(8):720-4.