

Large goiter in a patient with congenital hypothyroidism.

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OBJECTIVES

Description of a rare clinical case of congenital hypothyroidism with goiter, leading to compression of the trachea.

METHODS

Congenital goiter presenting in the newborn period is very rare. Here we present a case of congenital hypothyroidism with a large goiter, leading to trachea compression symptoms. Hormone replacement therapy was started leading to normal levels of TSH, FT₄ and of thyroid volume. In face of maternal normal thyroid levels, dyshormonogenesis considered to be the most probable cause of hypothyroidism.

RESULTS

A male newborn to non-consanguineous healthy parents, was born at term with normal height and weight. Neck mass was first diagnosed by ultrasound at 33 weeks of gestation. The boy intubated soon after birth due to respiratory disorders and asphyxia, which were caused by trachea compression. He was diagnosed with congenital goiter and thyroid deficiency at day 1 of life (Tab1., Pic 1.)

In addition, the boy was found to have tricuspid valve insufficiency, cardiomegaly and congenital pneumonia. Therapy with levothyroxine was started on the second day of life at the initial dosage of 15 mkg/kg/day, what lead to immediate reduction of the goiter volume and possibility of extubation. Normalization of TSH and FT₄ levels achieved at 29 days of life. Goiter volume decreased down to 13 ml by the 29th day of life.

Patient's DNA was analyzed for a wide genetic panel, including *NKX2-1*, *UBR1*, *GLIS3*, *AITD3*, *TRH*, *SLC26A4*, *SECISBP2*, *DUOXA2*, *THRB*, *IYD*, *SLC5A5*, *DUOX2*, *TRHR*, *TSHR*, *DUOX1*, *THRA*, *SLC16A2*, *TPO*, *TSHB*, *FOXE1*, *PAX8*, *GNAS*, *NKX2-5* genes, by using massive parallel sequencing, no mutations were found.

Table 1

Day of life	1	4	7	15	22	29	35
TSH (mIU/L)	>75.0	-	>75.0	>75.0	11.5	3.9	3.9
FT ₄ (pmol/L)	<4.5	-	<4.5	<4.5	-	17.6	24.7
Goiter volume (ml)	55	26.8	23	23	-	13	13
Levothyroxine dose (mkg/day)	50	50	75	75	75	75	50

Picture 1



1st day of life



4th day of life



30th day of life

CONCLUSIONS

Congenital hypothyroid goiter can grow huge volume. In our case, it led to trachea compression and need for mechanical ventilation. Even though we were not able to find mutations in genes, known to be involved in thyroid formation, we still suspect dyshormonogenesis as the most possible cause of goiter development in our patient. Additional genetics studies should be performed in attempt to disclose other possible mechanisms.