

IMAGES IN PEDIATRIC ENDOCRINOLOGY

Congenital Hypothyroidism Associated with Neonatal Tooth, Pierre-Robin Syndrome and Congenital Heart Defects

A female infant was born as the second child of healthy unrelated parents at 37 weeks of gestation by Caesarian section at a local hospital with birth weight 2,880 g, length 51 cm, and head circumference 33 cm. The pregnancy was complicated by polyhydramnion and placenta previa. The infant had stigmata of Pierre-Robin syndrome, and on the 5th day of age a tooth eruption in the region of the lower central incisor was noted. She required nasogastric tube feeding. Jaundice occurred during the first week of life. The patient was recalled because of a positive screening test for congenital hypothyroidism. At the time of screening on day 4, thyroid-stimulating hormone (TSH) was 136 mIU/l. On admission to our hospital, on the 13th day of age, physical examination revealed temperature 38.7°C, tachypnoea, peripheral cyanosis, slight oedema of the eyelids, low frontal and posterior hairline, micrognathia, retrognathia, cleft soft palate, muscular hypotonia. In addition, a continuous murmur with weak radial and femoral pulses were noted. An ultrasound examination of the thyroid gland was carried out confirming the absence of any thyroid tissue in the thyroid area at the base of the neck. At the same time, an ultrasound examination of the knee was done. She had evidence of delayed bone maturation based on absence of the distal femoral epiphyses. She was immediately started on 25 µg/day of L-thyroxine before awaiting biochemical confirmation of suspected diagnosis. Thyroid function tests done on the 13th day of life revealed TSH >100 mIU/l (normal range: 0.15-3.2), free thyroxine (fT4) 0.10 pmol/l (normal range: 10-25), total thyroxine (T4) 0.0 nmol/l (normal range: 65-160), total triiodothyronine (T3) 0.5 nmol/l (normal range: 1.04-2.5), and thyroglobulin (Tg) 6.1 ng/ml (normal range: 2-70). Roentgenography of the knee demonstrated absence of distal femoral epiphyses. Thyroid scintigraphy was not done. Echocardiography showed coarctation of the aorta, patent ductus arteriosus and patent foramen ovale with aneurysma of interatrial septum. Cytogenetic analysis demonstrated normal female karyotype. Because of a high degree of mobility, the tooth was extracted on the 20th day of life. Her clinical condition markedly improved and she was discharged from hospital on the 36th day of life. During the following weeks

she demonstrated signs of airway obstruction and failure to thrive. Unfortunately the child died from cardiorespiratory insufficiency on the 65th day of age at a local hospital.

Higher incidence of congenital extrathyroid anomalies in infants with congenital hypothyroidism than in the general population has been reported and especially congenital heart diseases^{1,2}. However, neonatal teeth with congenital hypothyroidism, as in our patient, has not to our knowledge been previously published. Further studies on congenital malformations in the general population and those associated with congenital hypothyroidism are still needed to help us understand the role of local genetic and environmental factors.

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