Congenital Hypothyroidism – Precise Diagnosis with Dual Imaging

L.Kasongo[1], L.Rausin[1], R.Nicolescu[2]
[1]CHR Citadelle, Université de Liège, [2]Isosl Valdor, Liège

Session: Fetal, neonatal endocrinology and metabolism (to include hypoglycaemia) 1

INTRODUCTION

Primary congenital hypothyroidism (PCH), defined as thyroid hormone deficiency, can be viewed as an anatomical, clinical, biochemical and radiological spectrum. Its etiology includes thyroid dysgenesis (85%) with defects in thyroid gland development and thyroid dyshormonogenesis (15%) with inborn error of thyroxine synthesis or release.

There is growing evidence that precise characterization of thyroid dysfunction by dual imaging, scintigraphy and ultrasound (US) is helpful in improving both medical care and understanding of the underlying disease.

Scintigraphy is the gold standard method for assessing the diagnosis of ectopic thyroid tissue and color Doppler US can accurately measure the thyroid volume, reveal additional information regarding the anatomy of the gland and possible remnant of the thyroglossal duct.

CASE PRESENTATION

Patient 1

An 8-day-old female presented with prolonged jaundice and lethargy. **Neonatal screening** revealed a thyroid-stimulating hormone (TSH) level >100 µIU/mL

Repeated serum thyroid function tests

TSH >200 µIU/mL (0.20 – 4.20)

thyroxine (T4) 1.8 pg/mL (7 - 17)

triiodothyronine (T3) 0.4 pg/mL (0.9-1.95)

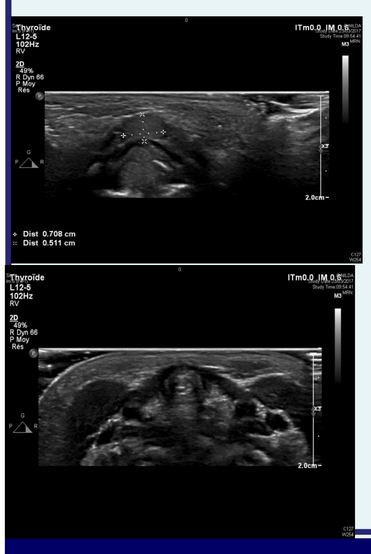
thyroglobulin 500 µg/L (1.4 - 78)

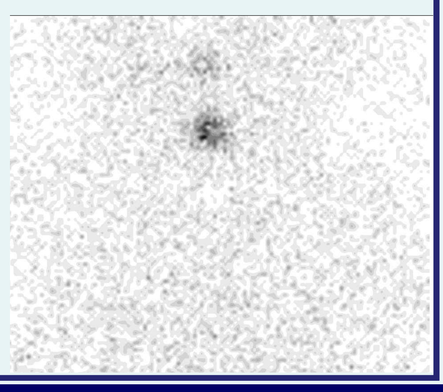
X-ray of the knee – bilateral absent epiphysis

Neck US - suspicious of thyroid agenesis and thyroglossal duct cyst.

The nuclear scan (^{123}I) - ectopic thyroid tissue in the wall of thyroglossal duct cyst.

Diagnosis – CH by thyroid dysgenesis and thyroglossal duct cyst.





Patient 2

An 8-day-old male newborn presented with hypo activity.

Neonatal screening – no available results.

The serum thyroid function tests

TSH 50 mIU/mL

T4 4 pg/mL,

T3 1 pg/mL

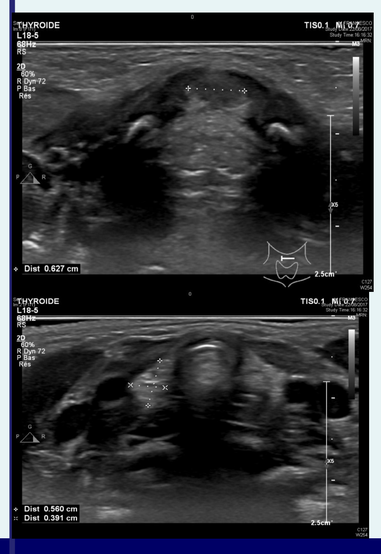
thyroglobulin 249 µg/L

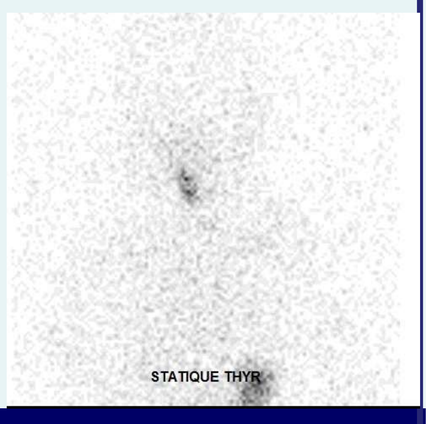
X-ray of the knee - normal ossification centers.

Neck US - hypoplasia of the thyroid tissue and thyroglossal duct cyst.

^{123}I scintigraphy - hemiagenesis, with absent left lobe.

Diagnosis – CH by thyroid dysgenesis and thyroglossal duct cyst.





CONCLUSIONS

Dual imaging allowed a specific diagnosis of the disorders of development of thyroid gland and abnormal obliteration of the thyroglossal duct. US and scintigraphy, as part of the optimal management of PCH, are complementary and provides the clinician maximal information on the anatomic, perfusion and functional status of the thyroid. Parents can be counseled on either the certainty of lifetime treatment (dysplastic thyroid) or the possibility of later discontinuing therapy (eutopic thyroid). Neonatal thyroid imaging requires an extensive amount of experience.

Randolph GW, Kamani DV. Thyroglossal duct cysts and ectopic thyroid. 2016 UpToDate

Léger J, Olivieri A, Donaldson M, Torresani T, Krude H, Vliet Gv, Polak M, Butler G. European Society for Paediatric Endocrinology Consensus Guidelines on Screening, Diagnosis,and Management of Congenital Hypothyroidism. 2014 Horm Res Paediatr, 81:80–103

Y-W Chang, D H Lee, Y H Hong, H S Hong. Congenital Hypothyroidism at US and Scintigraphy. 2011 Radiology, 258:872-79