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Review Article

Diagnostic Radiologic Imaging in Patients with Congenital Hypothyroidism

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A b s t r a c t

Neonatal screening programmes to detect congenital hypothyroidism (CH) revealed an incidence of approximately one (in 2500-5000). Primary CH may be due to an absence or hypoplastic gland, an ectopic gland, or an inborn error of thyroid hormone metabolism. Determination of the cause of CH is of genetic, epidemiological and prognostic importance. Diagnostic imaging, therefore, is of paramount importance. The various radiological imaging methods are highlighted in this brief review.

K e y w o r d s

Congenital hypothyroidism
Diagnostic Imaging
Iodine^{123}
Perchlorate Technetium 99m Discharge test
Ultrasound
Scintigraphy

Introduction

Congenital hypothyroidism (CH) is a preventable cause of mental retardation with a worldwide incidence of 1 in 2500 to 5000 live births. The paucity of early signs and symptoms and the need for early treatment led to the development of screening. Primary CH may be due to an absence or hypoplastic gland, an ectopic gland, or an inborn error of thyroid hormone metabolism which is inherited as an autosomal recessive disorder. Determination of the cause of CH is of genetic, epidemiological and prognostic importance, and is gaining momentum (El Desouki et al., 1995; Leger et al., 2014). The pediatric radiologist, in collaboration with the endocrinologist can outline this. In this brief review, various diagnostic modalities used in the assessment of patients with congenital hypothyroidism are highlighted.

Anatomy and Physiology

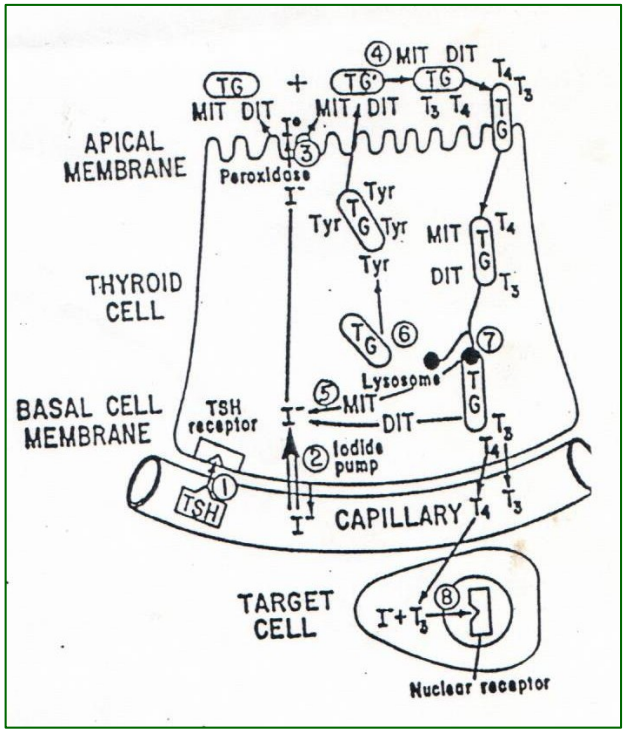
The thyroid gland is visible in the three week embryo as an endodermal projection between the first and second bronchial arches, a point marked by the foramen cecum at the base of the tongue. During the subsequent three weeks it migrates to lie in front of the thyroid cartilage. The factors that control its migration are unknown. Originally, it is attached to the foramen cecum by the thyroglossal duct, which usually atrophies. Colloid formation appears by 10 weeks.

The fetal thyroid gland is capable of trapping iodine by 8-10 weeks and producing thyroxine (T4) by 12 weeks of gestation. Production of hypothalamic thyrotropin releasing hormone (TSH) occur about

the same time but integration and function of the hypothalamic-pituitary-thyroid axis with negative feedback does not occur until the second half of pregnancy. Prior to mid-gestation, the fetus appears to be dependent on maternal thyroid hormone for normal development. Recent studies show that approximately one-third of maternal T4 crosses the placneta to the fetus. After birth, there is a surge of TSH which peaks at 30 min in the range of 70-100 mU/L, resulting in increased serum T4 and T3 levels which gradually fall down over the first four weeks of life. In the premature infant, however, serum T4 levels are lower, but rise to meet full term infant levels by approximately six weeks.

Essentially, all the steps involved in thyroid hormone synthesis including iodine trapping, oxidation organification, coupling and secretion (Fig. 1) are under control of TSH. The majority of T4 and T3 are carried in the circulation by binding proteins, hence, TBG deficiency or excess will affect measurements of total hormones concentration. Measurements of free hormones levels, therefore, are more accurate.

Fig. 1: Diagrammatic representation of thyroid hormone synthesis, secretion and utilization
(La Franchi, 1993).



Absent or mal-descent of the thyroid gland or an inborn error of thyroid hormone synthesis leads to congenital hypothyroidism, because the vast majority of infants do not display any manifestation of hypothyroidism during birth. In the absence of prompt replacement therapy, the developing brain would be damaged. Screening for congenital hypothyroidism was initiated in Quebec, Canada in 1972. Since then, this practice has spread to most of the industrialized world. Racial differences in the incidence of CH were noted (La Franchi, 1993).

Diagnostic imaging studies

The pediatric radiologist and a nuclear medicine consultant play an important role in determining the aetiology of CH. An ultrasound (Fig. 2) can be performed, even after starting therapy, which is not feasible for isotope scanning. Although, it had a high specificity, yet it is not reliable in detecting ectopic tissue, and an operator dependent (Cone et al., 1988; Takashima et al., 1995; Delaney et al., 2012).

Thyroid scan using Tc-99, pertechnetate (Figs. 3, 4, 5 and 6) OR Iodine-123 with or without Perchlorate Discharge Test (PDT) (Wells et al., 1986; Schoen et al., 2004; Heyman et al., 1982; O'Connor et al., 1982; Arnold et al., 1976; Gulenchyn et al., 1981; Treves et al., 1985), when indicated, may be undertaken prior to the onset of therapy to determine the underlying etiology. For thyroid scanning, Tc-99m is preferred to Iodine-123 because it is available around the clock, much less expensive, employs shorter scanning time, yields less radiation dose, and better defines the thyroid in relation to the surrounding tissue.

The usual dose of Tc-99m pertechnetate in children is 13.5 kBq (500 uCi) administered intravenously and imaging is performed 15 to 20 min after that using a gamma camera equipped with a low energy general purpose collimator. An initial zoomed image and another unzoomed image, to show the salivary glands and the stomach, are obtained. Radioactivity in the syringe before and after injecting is measured to give the corrected administered dose to measure thyroid uptake. The normal Tc-99m-uptake in children is in the range between 0.5% and 4.0%.

Fig. 2: An ultrasound of a normal thyroid gland (A) and a patient with congenital hypothyroidism due to thyroid aplasia (B).

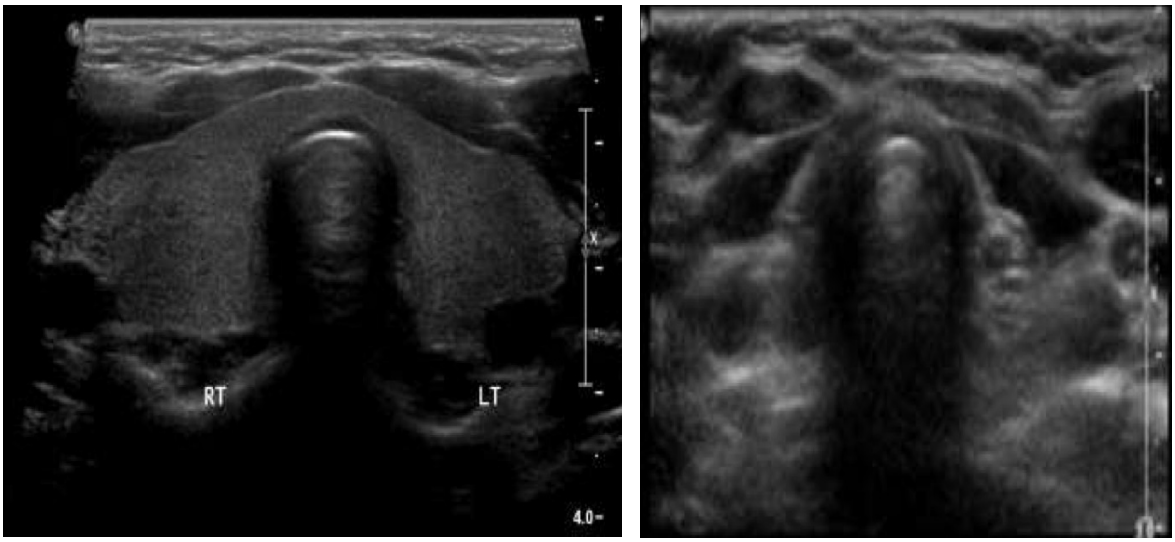
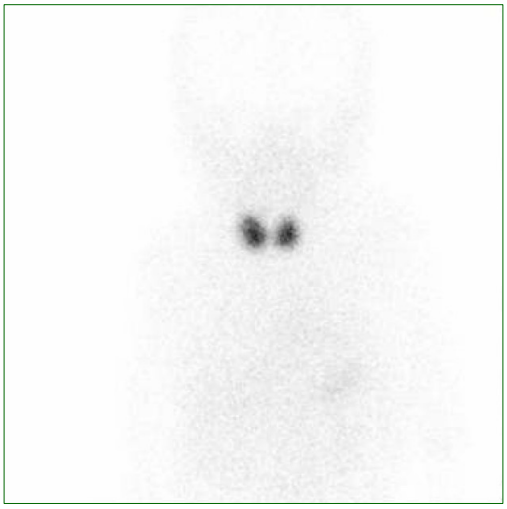


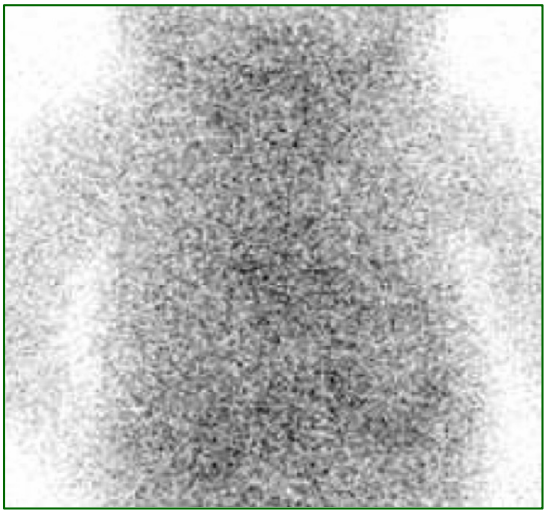
Fig. 3: Tc-99-m pertechnetate thyroid scan of a patient with congenital hypothyroidism.
Note: the eutopic enlarged gland in a patient with "dyshormono genesis"



Iodine-123 thyroid scan is performed by giving 1.35 kBq (50uCi) orally, imaging the thyroid and measuring the uptake at 6 and 24 h.

The PDT is performed using 1.35 kBq (50 uCi) of Iodine-123 instilled directly in the mouth followed by water or milk to wash the mouth. Thyroid uptake is measured using a scintillation probe and scaler at 1 and 2 h before administering a 400 mg dose of potassium perchlorate to measure the rate of iodine-washout every 15 min for 1 h and every 30 min for another 1 h. When organification defect is suspected, PDT test is performed for confirmation. A discharge rate of more than 50% indicates a virtually complete organification defect while a discharge rate between 20% and 50% indicates a partial defect.

Fig. 4: Tc-99-m pertechnetate thyroid scan of a patient with congenital hypothyroidism.
Note: the absence of thyroid tissue "thyroid aplasia".



If an ectopic thyroid gland is identified, a permanent form of hypothyroidism exists. If the thyroid scan is compatible with aplasia (i.e. no uptake) as well as infants whose mothers had an autoimmune disorder resulting in the production of thyrotropin receptor blocking antibodies. In this setting, if an ultrasound examination of the thyroid confirms aplasia, again a

permanent form of hypothyroidism has been established. However, if an ultrasound examination shows a normal thyroid gland, further diagnostic studies must be undertaken. If permanent hypothyroidism has not been diagnosed prior to the onset of therapy, it is recommended that treatment discontinued sometime after the age of 3 years for 30 days to determine permanency of the hypothyroidism.

Fig. 5: Tc-99-m pertechnetate thyroid scan of a patient with congenital hypothyroidism:
Note: the lingual gland in a patient with "ectopic thyroid gland".

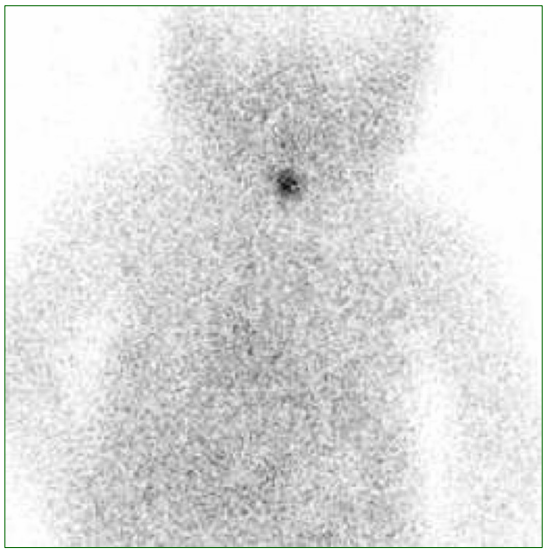
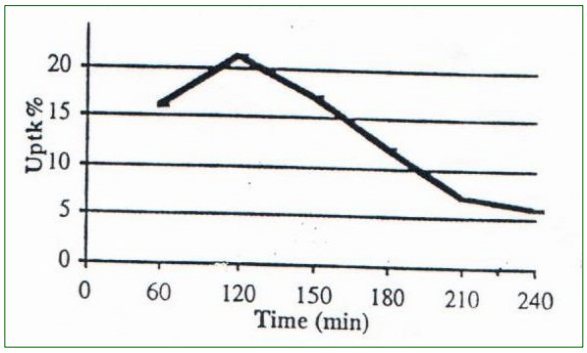


Fig. 6: Perchlorate discharge test (PDT): positive test with 2 h discharge of 65% from patient with dyshormonogenesis



In conclusion, ultrasound as the primary imaging modality was suggested, as it had a high specificity, however, it is operator dependent and was not reliable in detecting ectopic thyroid. Quantitative 99mTc pertechnetale and PDT can add useful aetiological, genetic and prognostic information in the clinical evaluation of infant suspected of having CH as a result of neonatal screening.

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