

Thyroid Imaging Study for the Diagnosis of Congenital Hypothyroidism with Thyroid Dysgenesis

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Introduction

Primary congenital hypothyroidism can be divided into thyroid dysgenesis and thyroid hormone synthesis disorders. Thyroid dysgenesis is divided into aplasia(33%), hypoplasia and Ectopia(66%). Ectopia is functioning thyroid tissue found anywhere other than usual anatomic location of thyroid gland. It occurs 1 in 1,000,000-3,000,000 of newborns. Rarely it could be found in more than two location. About 1/3 of ectopic thyroid is symptomatic with congenital hypothyroidism, but thyroid hypertrophy is prominent during adolescence and pregnancy, so it may be difficult to diagnose it in infancy. The purpose of this study was to evaluate the imaging patterns of patients with congenital thyroid hypoplasia and to evaluate the diagnostic value of scan and ultrasound in thyroid dysplasia.

Subjects and Methods

Initially, the subjects who performed thyroid scintigraphy between 2002 and 2017 at department of Pediatrics, Dankook University Hospital were included in the study. Medical records were reviewed about clinical information, laboratory results, and imaging studies. Finally, only the subjects who were referred to Dankook University Hospital due to abnormal neonatal screening test or abnormal thyroid function test, neck mass, or suggestive symptoms of congenital hypothyroidism were included in the study. A total of 64 patients with suspected congenital hypothyroidism underwent thyroid scintigraphy. Nineteen patients with ectopic thyroid or invisible thyroid tissue on thyroid scintigraphy were included in the study (Fig. 1). Results of thyroid scintigraphy and sonography were analyzed based on the diagnosis.

Results

There were more females in the study (M:F 1:2.8). The most common cause of thyroid imaging study was abnormal result of neonatal screening test. Four preterm babies and one schizencephaly were included in the study (Table 1). Final clinical diagnosis was done based on clinical information and imaging studies (Table 1). In cases with hypoplasia or aplasia in imaging studies, more careful interpretation may be necessary because there may be mismatch in the interpretation between sonography and scintigraphy. Thyroid imaging studies with ectopic thyroid or invisible thyroid tissue on thyroid scintigraphy were included in the study (Table 2). Sometimes, re-reading or reevaluation of thyroid imaging studies lead to the different interpretations (Table 3 & Fig. 2). There were mismatch with the results of sonography and scintigraphy, especially in hypoplasia and aplasia (Table 3). Thyroid sonography was very useful to detect normally positioned thyroid tissue, and scintigraphy was more sensitive to detect ectopic thyroid (Table 3).

Table 1. Clinical characteristics of study subjects

Sex	M = 5, F = 14 (1:2.8)	
Age (month) at 1st visit	4.47 ± 9.79 (0.26-33.83)	
Age (yr) at recent visit	6.86 ± 3.95 (0.82-15.10)	
Follow-up period (yr)	6.54 ± 4.26 (0.46-15.60)	
Initial presentation		
Abnormal NST	16	84.21 %
Developmental delay & low T4	1	5.26 %
Tongue base mass	1	5.26 %
Constipation	1	5.26 %
Accompanied symptom		
Prematurity	4	21.05 %
Schizencephaly	1	5.26 %

January, 1st, 2002 – April, 30th, 2017
Underwent Thyroid scintigraphy below 3 years old with congenital hypothyroidism (N=6.)

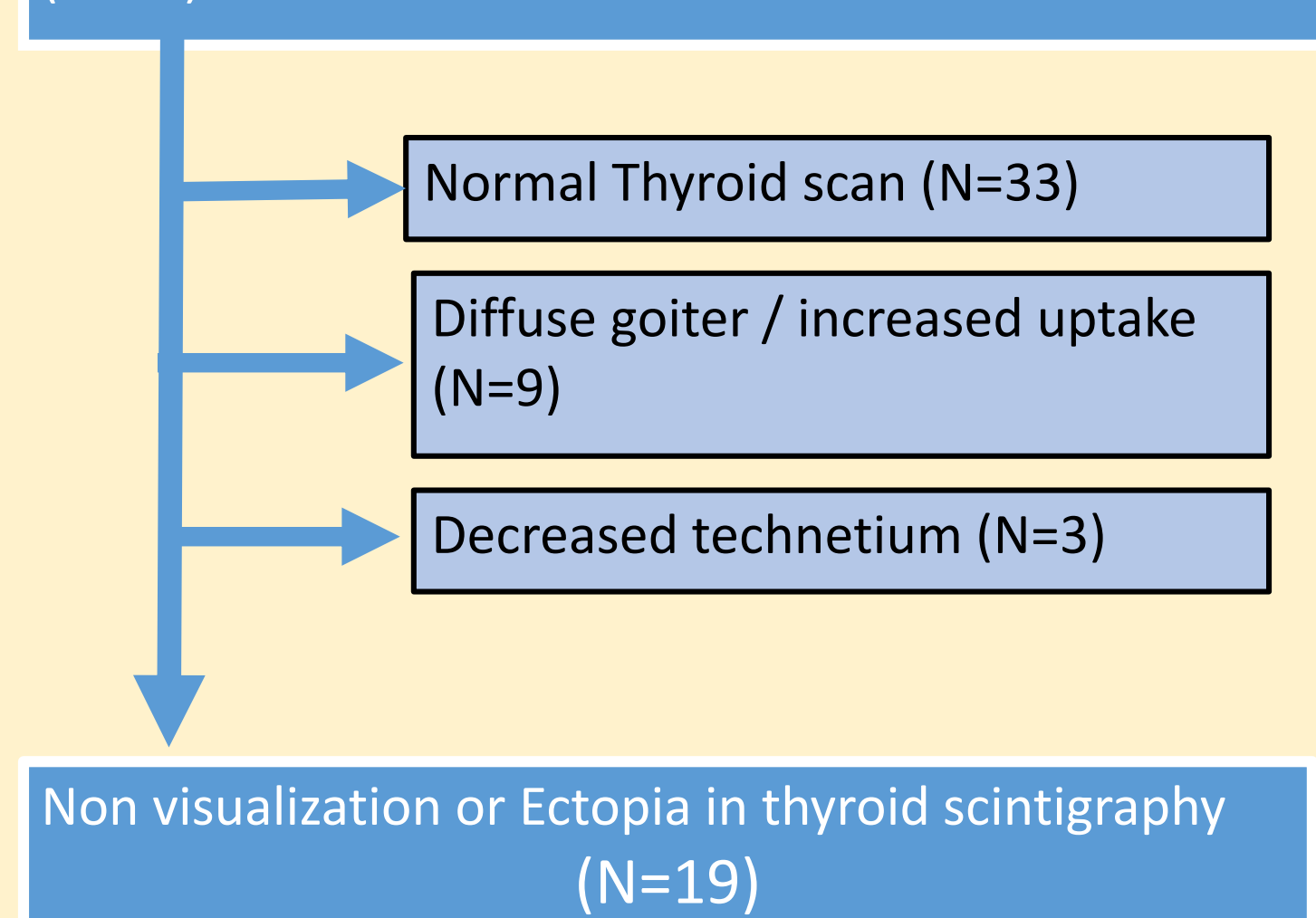


Table 2. Final diagnosis based on imaging studies

Final diagnosis (N=19)	N	%
Ectopia	9	47.4
Hemithyroid	1	5.3
Hypoplasia	3	15.8
Aplasia	1	5.3
Normal	5	26.3

Fig. 1. The schematization of the study protocol

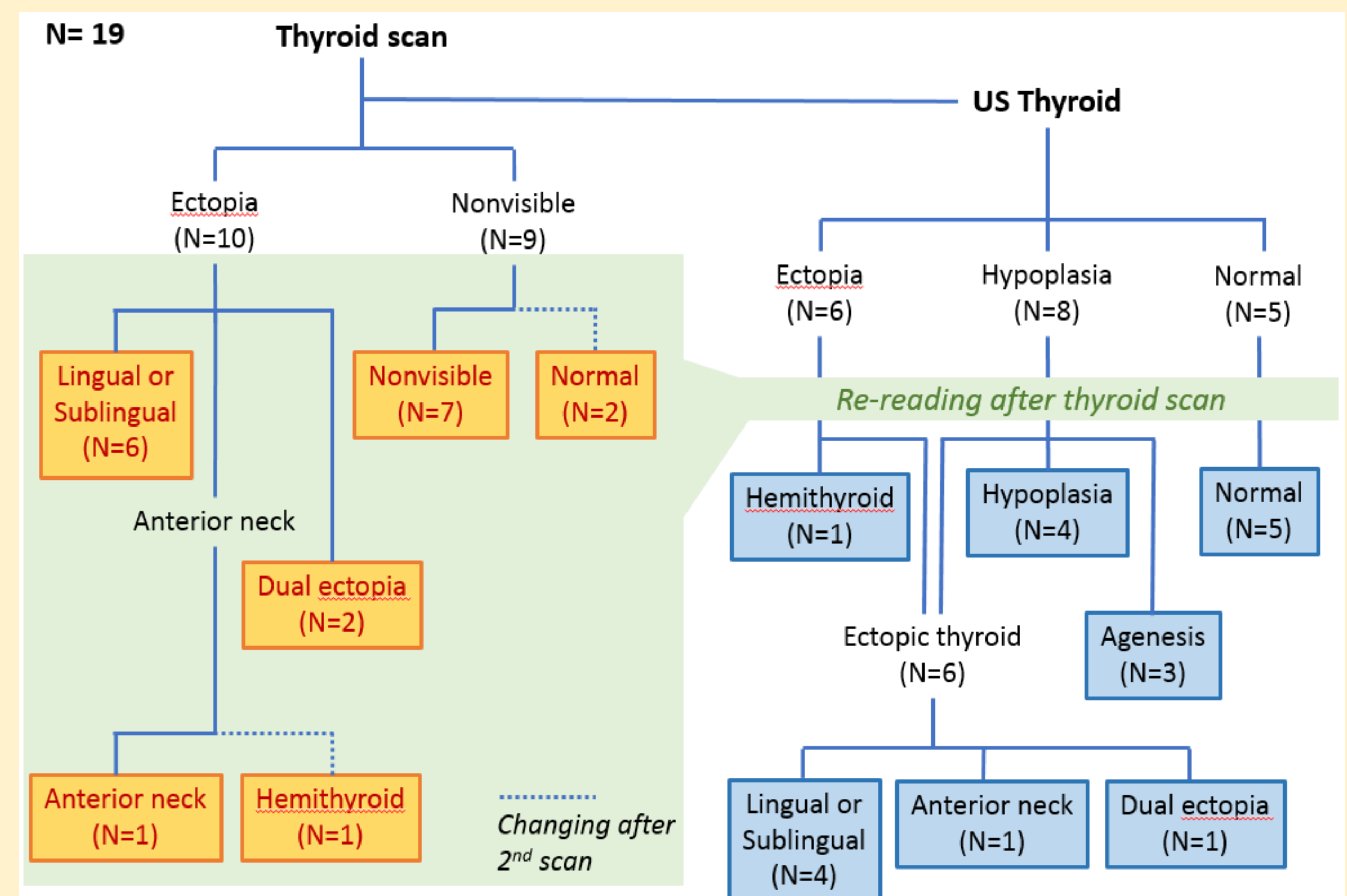


Fig. 2. Initial and final results of thyroid imaging studies

Table 3. Diagnostic value of thyroid scintigraphy and sonography

	N	Thyroid Scintigraphy				
		Sensitivity	Specificity	PPV	NPV	Accuracy
Ectopia	9	100	100	100	100	100
Hemithyroid	1	100	100	100	100	100
Hypoplasia	3	0	100	0	84.21	84.21
Aplasia	1	100	58.82	12.5	100	57.89
Normal	5	20	92.9	50	76.5	73.7
	N	Thyroid Sonography				
		Sensitivity	Specificity	PPV	NPV	Accuracy
Ectopia	9	66.7	100	100	76.9	84.2
Hemithyroid	1	100	100	100	100	100
Hypoplasia	3	10	93.75	75	100	94.74
Aplasia	1	100	88.89	33.33	100	89.47
Normal	5	100	100	100	100	100

Conclusion

Thyroid sonography was very useful to detect normally positioned thyroid tissue, and scintigraphy was more sensitive to detect ectopic thyroid. Both sonography and scintigraphy are required for the correct diagnosis of thyroid dysgenesis, especially in the case with suspected thyroid hypoplasia or aplasia.

