

Original Article

Diagnostic Accuracy of Ultrasonography for Diagnosis of Congenital Hypothyroidism in Neonates Taking Scintigraphy as Gold Standard

Irfan Sharif¹, Tayyab Javed², Jawairia Aslam³, Iqra Waheed⁴

¹Tehsil Head Quarter Hospital Dunyapur, ²Rural Health Center Manawala,

³Hilal-e-Ahmer Hospital, Faisalabad, ⁴Private Services Lahore

Abstract

Objective: To assess the diagnostic accuracy of ultrasonography for diagnosis of congenital hypothyroidism in neonates taking scintigraphy as gold standard.

Methods: This Cross-sectional, validation study was undertaken in Department of Pediatrics, Mayo Hospital, Lahore during August 2017 to February 2018. A total of 202 neonates who met the inclusion criteria were included in the study. Then neonates underwent thyroid ultrasonography. Neonates were labeled as positive or negative for congenital hypothyroidism. Then all neonates underwent scintigraphy as per hospital protocol. Reports were assessed and neonates were confirmed as positive or negative for congenital hypothyroidism. All data was entered in specially designed Performa. The data was analyzed in SPSS version 20.

Results: The mean age of neonates was 13.02±7.55 days. There were 97 (48%) males and 105 (52%) females. The mean weight of neonates was 2.28±0.39 kg. On ultrasound, 76 (37.6%) neonates who have congenital hypothyroidism while 126 (62.4%) neonates had normal thyroid levels. On scintigraphy, 85 (42.1%) neonates who have congenital hypothyroidism while 117 (57.9%) neonates had normal thyroid levels. The sensitivity of ultrasound was 82.4%, “specificity was 94.9%, PPV was 92.1%, NPV was 88.1% and diagnostic accuracy was” 89.6% respectively taking scintigraphy as gold standard.

Conclusion: Thus the ultrasound is accurate enough that it can detect congenital hypothyroidism instead of going for interventional procedure of scintigraphy, which includes radioactive material.

Key words: Ultrasonography, congenital hypothyroidism, neonates, scintigraphy

How to cite this:

Sharif I, Javed T, Aslam J, Waheed I. Diagnostic accuracy of ultrasonography for diagnosis of congenital hypothyroidism in neonates taking scintigraphy as gold standard. J Pak Soc Intern Med. 2022;3(3): 230-232

Corresponding Author: Dr. Irfan Sharif

Email: irfanmalik210@gmail.com

Introduction

Thyroid hormone is an important hormone for the appropriate growth and maturation of human skeleton.¹ Congenital hypothyroidism is the most prevalent causes of mental retardation among pediatrics. It may occur in about 1:2,000-1:4,000 neonates.² Although it is prevalent but curable cause of mental retardation. The prevalence of Congenital hypothyroidism in Pakistan is high because of deficiency of neonatal screening program at national level, lack of training and education of parents, high number of consanguinity, and lack of training and suspicion of doctor may cause the delay in the diagnosis and raised incidence of the congenital hypothyroidism. This threat can be effectively resolved by a mass neonatal screening campaign and strong laws

to ensure that significant attempts are taken to eliminate this avoidable disease from Pakistan.³

Screening programmes for congenital hypothyroidism have been developed more than 30 years ago, the optimum screening technique and, in particular, the TSH threshold have not yet been successfully identified.⁴ One study, conducted in Faisalabad, showed that there were about 14% cases that were detected with congenital hypothyroidism in neonates of aged less than 3 months.⁵ Ultrasound detected congenital hypothyroidism with sensitivity, specificity, PPV, and NPV of 77%, 92%, 89% and 84%, respectively.⁶

The aim of this research was to determine the diagnostic accuracy of ultrasonography for detection of congenital hypothyroidism in neonates taking scintigraphy as gold

standard. Literature has shown that USG can be helpful in diagnosing congenital hypothyroidism in neonates instead of applying scintigraphy. For scintigraphy, radioisotope is injected in body of neonate and then scan is done which also contains radiations. Instead of that USG is non-invasive and easily accessible procedure. But there is no local study conducted before. So we conducted this study to confirm whether the ultrasonography can replace the scintigraphy. This study was undertaken to assess the diagnostic accuracy of ultrasonography for diagnosis of congenital hypothyroidism in neonates taking scintigraphy as gold standard.

Methods

Study design: Cross-sectional, validation study

Setting: Department of Pediatrics, Mayo Hospital, Lahore.

Study period: 6 months i.e. 5-8-2017 to 6-2-2018

Sample size: Sample size was 202 cases, estimated by using 95% confidence level, margin of error as 13%, percentage of congenital hypothyroidism i.e. 14% with sensitivity of ultrasound i.e. 77% and specificity of ultrasound i.e. 92%.

Sampling-technique: Nonprobability, Consecutive sampling

Selection criteria: Neonates of age 1-28 days of either gender were included. While preterm neonate (gestational age < 37 weeks), very low birth weight neonates (< 1.5 kg), congenital heart disease were excluded.

Data collection: 202 neonates who met the inclusion criteria were included in the study from emergency. After taking consent form from parents, demographics were also taken. Then neonates underwent ultrasonography by a single senior radiologist having at least 4 years' residency experience. Thyroid ultrasound was done on entire neck when patient lie vertically. The neck was then, hyperextended and HONDA (Japan) ultrasound machine, with HLS-475M, 7.5 MHZ linear transducer was used to image the thyroid. Neonates were labeled as positive or negative for congenital hypothyroidism if there will be presence or absence of ectopic or agenesis thyroid tissue, size hyperplastic (>50 + normal), increased echogenicity and increased degree of thyroid vascularity. Then all neonates underwent scintigraphy as per hospital protocol. Reports were assessed and neonates were confirmed as positive or negative for congenital hypothyroidism if there will be ectopic or agenesis thyroid tissue will be present, hyperplastic size (>50+ normal), increased echogenicity and increased degree of thyroid vascularity. All data was entered in Performa.

Data analysis: Collected data was entered and analyzed in SPSS v. 20. 2×2 table was developed to the calculate

“sensitivity, specificity, PPV, NPV & diagnostic accuracy” of ultrasound keeping scintigraphy as gold standard.

Results

The mean age of neonates was 13.02 ± 7.55 days. There were 97 (48%) males and 105 (52%) females. The mean weight of neonates was 2.28 ± 0.39 kg. Table 1

The sensitivity, specificity, PPV, NPV and diagnostic accuracy were 82.4%, 94.9%, 92.1%, 88.1% and 89.6% respectively taking scintigraphy as gold standard. Table 2

Table 1: Baseline characteristics of neonates

n	202
Age (days)	13.02 ± 7.55
Gender	
Male	97 (48.0%)
Female	105 (52.0%)
Weight (kg)	2.28 ± 0.39

Table 2: Diagnostic accuracy of ultrasound in detection of congenital hypothyroidism

		Scintigraphy		Total
		Positive	Negative	
Ultrasound	Positive	70	6	76
	Negative	15	111	126
Total		85	117	202

Sensitivity: 82.4%, Specificity: 94.9%, PPV: 92.1%, NPV: 88.1%, Diagnostic accuracy: 89.6%

Discussion

“Congenital hypothyroidism”, is a highly prevalent endocrine disorder observed in neonates. It can be noticed in about 1 to 3,000-4,000 live-births. The incidence may vary from race to race and ethnicity to ethnicity.⁷ Congenital hypothyroidism leads to significant neuro-developmental dysfunction when care is missed, hence, several countries adopt the neonatal screening programmes for early diagnosis of congenital and neonatal hypothyroidism.⁸

Congenital hypothyroidism as also be reported relatively common in Iranian population; the average prevalence of the disorder has been reported as 1:914 in Tehran, to 1:1433 in Fars and 1:370 in Isfahan.^{9,10} While most infants are now diagnosed with Congenital hypothyroidism after diagnosis by neonatal screening programmes by using thyroid stimulating hormone level, thyroid imaging is required for aetiology recognition.¹¹

Determining the aetiology of the condition is particularly critical for choosing effective therapeutic methods and recognizing the effects of inheritance and prognosis.^{12,13}

Thyroid scintigraphy by using ^{99m}Tc or ^{123}I , is a reliable diagnostic tool for diagnosis of congenital thyroid disorder. However, thyroid scintigraphy is mostly recommended for diagnosis of congenital hypothyroid and its related causes, ultrasound can also replace scintigraphy as it is non-invasive and usually can also evaluate the anatomical changes in thyroid gland.¹⁴

Ultrasound of thyroid gland may ignore few cases of ectopic glands, therefore scintigraphy is found to be more reliable for detection of dysgenetic ectopic thyroid glands and physically intact, but non-functional glands than ultrasound.¹⁵⁻¹⁷

In our study, the mean age of neonates was 13.02 ± 7.55 days. There were 97(48%) males and 105(52%) females. The mean weight of neonates was $2.28 \pm 0.39\text{kg}$. The sensitivity, specificity, PPV, NPV & diagnostic accuracy were 82.4%, 94.9%, 92.1%, 88.1% & 89.6% respectively taking scintigraphy as gold standard. Hashemipour et al., found that ultrasound have sensitivity, specificity, PPV, and NPV of 77%, 92%, 89% and 84%, respectively for detection of congenital hypothyroidism.⁶

But, ultrasound has certain limitations in the diagnosis of ectopic thyroid glands. Hashemipour et al., found that about 33% of ectopic thyroid glands were detected on ultrasound.⁶ In another study, De Bruyn et al., performed the ultrasound on 54 infants with congenital hypothyroidism, only 19.2% cases having ectopic thyroid gland were detected on ultrasound.¹⁹

In another related study, conducted in Korea, conflicting cases of congenital hypothyroidism were examined through comparison of ultrasound and scintigraphic findings; in 2% cases, ultrasound was unable to identify the ectopic thyroid glands and the sensitivity was reported as 78%, which was higher than we observed in our study. The precision of ultrasound was 100%, which is closer to the findings of our study.²⁰

Conclusion

Thus the ultrasound is accurate enough that it can detect congenital hypothyroidism instead of going for interventional procedure of scintigraphy which includes radioactive material.

Conflict of Interest: None

Funding Source: None

References

1. Patidar PP, Philip R, Toms A, Gupta K. Radiological manifestations of juvenile hypothyroidism. *Thyroid Res Pract.* 2012;9(3):102.
2. Anjum A, Afzal MF, Iqbal SMJ, Sultan MA, Hanif A. Congenital hypothyroidism in neonates. *Indian J Endocrinol Metab.* 2014;18(2):213.
3. Ali M, Zia A, Siddiqui SE. New born screening in preventing congenital hypothyroidism. *J Ayub Med Coll Abbottabad.* 2015;27(4):953-4.
4. Raza H, Riaz S, Jamal M, Shirazi H, Gul S. Congenital Hypothyroidism Newborn Screening-The PIMS Experience. *Ann Pak Inst Med Sci.* 2013;9(3):198-200.
5. Malik BA, Butt MA. Is delayed diagnosis of hypothyroidism still a problem in Faisalabad, Pakistan. *J Pak Med Assoc.* 2008;58(10):545-9.
6. Hashemipour M, Rostampour N, Nasry P, Hovsepian S, Basiratnia R, Hekmatnia A, et al. The role of ultrasonography in primary congenital hypothyroidism. *J Res Med Sci.* 2011;16(9):1122.
7. Rastogi MV, LaFranchi SH. Congenital hypothyroidism. *Orphanet J Rare Dis* 2010;5:17.
8. Gruters A, Krude H. Update on the management of congenital hypothyroidism. *Horm Res.* 2007;68 (S-5): 107-11.
9. Dalili S, Rezvani SM, Dalili H, Mohtasham Amiri Z, Mohammadi H, Abrisham Kesh S, et al. Congenital hypothyroidism: etiology and growth-development outcome. *Acta Med Iran.* 2014;52(10):752-6.
10. Hashemipour M, Amini M, Iranpour R, Sadri GH, Javaheri N, Haghighi S, et al. Prevalence of congenital hypothyroidism in Isfahan, Iran: results of a survey on 20,000 neonates. *Horm Res.* 2004;62(2):79-83.
11. Hashemipour M, Rostampour N, Nasry P, Hovsepian S, Basiratnia R, Hekmatnia A, et al. The role of ultrasonography in primary congenital hypothyroidism. *J Res Med Sci.* 2011;16(9):1122-8.
12. Schoen EJ, Clapp W, To TT, Fireman BH. The key role of newborn thyroid scintigraphy with isotopic iodide (^{123}I) in defining and managing congenital hypothyroidism. *Pediatrics.* 2004;114(6):e683-8.
13. Dias VM, Campos AP, Chagas AJ, Silva RM. Congenital hypothyroidism: etiology. *Journal of pediatric endocrinology & metabolism: JPEM* 2010;23(8):815-26.
14. Rose SR, Brown RS, Foley T, Kaplowitz PB, Kaye CI, Sundararajan S, et al. Update of newborn screening and therapy for congenital hypothyroidism. *Pediatrics.* 2006;117(6):2290-303.
15. Kobayashi H, Tashita H, Hara H, Hasegawa Y. Utility of computed tomography in identifying an ectopic thyroid in infants and pre-school children. *Endocr J.* 2005;52(2):189-92.
16. Kreisner E, Camargo-Neto E, Maia CR, Gross JL. Accuracy of ultrasonography to establish the diagnosis and aetiology of permanent primary congenital hypothyroidism. *Clin Endocrinol.* 2003;59(3):361-5.
17. Garel C, Leger J. Thyroid imaging in children. *Endocr Develop.* 2007;10:43-61.
18. Poyhonen L, Lenko HL. Ultrasonography in congenital hypothyreosis. *Acta Paediatr Scand.* 1984;73(4):523-6.
19. De Bruyn R, Ng WK, Taylor J, Campbell F, Mitton SG, Dicks-Mireaux C, et al. Neonatal hypothyroidism: comparison of radioisotope and ultrasound imaging in 54 cases. *Acta Paediatr Scand.* 1990;79(12):1194-8.
20. Chang YW, Lee DH, Hong YH, Hong HS, Choi DL, Seo DY. Congenital hypothyroidism: analysis of discordant US and scintigraphic findings. *Radiology.* 2011; 258(3):872-9.