



ORIGINAL ARTICLE

Detecting congenital hypothyroidism with newborn screening: the relevance of TSH cutoff values[☆]

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KEYWORDS

Congenital hypothyroidism;
Newborn screening;
Neonatal screening;
Thyrotropin

Abstract

Objectives: To assess the prevalence of congenital hypothyroidism and the ability of various neonatal thyroid-stimulating hormone (TSHneo) cutoff values to detect this disease.

Methods: This cohort study was based on the retrospective collection of information available from the Reference Service for Newborn Screening database for all live births from January 1, 2010, to December 31, 2012, assessed using the Newborn Screening Program of a Brazilian state. The infants were divided into two groups: I – Control: infants with normal newborn screening tests and II – Study: infants with congenital hypothyroidism. Analysis included comparing the TSHneo levels from both groups. A receiver operating characteristic (ROC) curve was constructed to assess the TSHneo cutoff values.

Results: Using a TSHneo cutoff value of 5.0 μIU/mL, 50 out of 111,705 screened infants had diagnosis of congenital hypothyroidism (prevalence 1:2234 live births). The ROC curve showed that TSHneo value of 5.03 μIU/mL had 100% sensitivity and the greatest associated specificity (93.7%). The area under the curve was 0.9898 ($p < 0.0001$).

Conclusions: The ROC curve confirmed that the TSHneo cutoff value of 5.0 μIU/mL adopted by the Newborn Screening Program of a Brazilian state was the most appropriate for detecting congenital hypothyroidism and most likely explains the high prevalence that was found.

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PALAVRAS-CHAVE

Hipotireoidismo congênito;
Triagem de recém-nascidos;
Triagem neonatal;
Tirotopina

Detecção de hipotireoidismo congênito com triagem de recém-nascidos: a relevância dos valores de corte de TSH**Resumo**

Objetivos: Avaliar a prevalência do hipotireoidismo congênito e a capacidade de vários valores de corte do hormônio estimulante da tireoide neonatal (TSHneo) detectar essa doença.

Métodos: Este estudo de corte teve como base a coleta retrospectiva de informações disponíveis no banco de dados do Serviço de Referência em Triagem Neonatal sobre todos os nascidos vivos de 1° de janeiro de 2010 a 31 de dezembro de 2012, avaliados utilizados o Programa de Triagem Neonatal de um estado brasileiro. Os neonatos foram divididos em dois grupos: I–Controle: neonatos com testes de triagem neonatal normais e II–Estudo: neonatos com hipotireoidismo congênito. A análise incluiu a comparação entre os níveis de TSHneo dos dois grupos. Uma curva da Característica de Operação do Receptor (ROC) foi criada para avaliar os valores de corte de TSHneo.

Resultados: Utilizando um valor de corte de TSHneo de 5,0 µIU/mL, 50 dos 111.705 neonatos examinados foram diagnosticados com hipotireoidismo congênito (prevalência de 1:2.234 nascidos vivos). A curva ROC mostrou que o valor do TSHneo de 5,03 µIU/mL possuía sensibilidade de 100% e a maior especificidade relacionada (93,7%). A área abaixo da curva foi 0,9898 ($p < 0,0001$).

Conclusões: A curva ROC confirmou que o valor de corte de TSHneo de 5,0 µIU/mL adotado pelo Programa de Triagem Neonatal de um estado brasileiro foi o mais adequado na detecção do hipotireoidismo congênito e provavelmente explica a alta prevalência constatada.

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Introduction

Congenital hypothyroidism (CH) is a common pediatric endocrine disorder^{1,2} that can cause mental retardation.^{3,4} The importance of early diagnosis and treatment, to prevent irreparable brain damage^{4,5} and growth retardation,^{3,6} justified the establishment of newborn screening programs (NSPs) for the detection of CH.^{4,7} These NSPs proved to be significantly cost-effective to society at large.^{8,9}

Measuring the concentration of thyroid-stimulating hormone (TSH) is considered the best screening test strategy for detecting primary CH⁴ because it exhibits high sensitivity^{4,10} and accuracy.¹¹ However, CH detection depends directly on the neonatal TSH (TSHneo) cutoff value,^{4,12} which varies among different NSPs^{13,14} and may have an influence on the recall rate.¹³ According to available evidence, many cases of CH would go undetected if the TSHneo cutoff value was increased.^{9,15} On the other hand, reducing the cutoff value requires prior judicious assessment of the laboratory work quality¹⁵ and costs to the screening program.^{4,9} The criteria used for choosing the TSHneo cutoff for detecting CH should be adapted to the target disease definition.⁴

Some programs worldwide have lowered the screening test cutoffs^{5,7} in order to increase the sensitivity of the assay.¹⁶ In accordance with that, cutoff values close to 10.0 µIU/mL^{2,8} or as low as 5.0 µIU/mL^{3,17} or 4.5 µIU/mL¹⁸ are used in several NSPs worldwide to improve the detection of children at risk.

The prevalence of CH in Brazil is approximately one case per 2500 live births; regional variation has been noted and according to the 2010 Brazilian Health Ministry

recommendations, a TSH value higher than 15.0 µIU/mL per immunometric assay should be considered positive.¹⁹

Because the efficiency of any NSP depends on its ability to cover the largest possible population and to detect the largest number of cases, obtaining reliable TSH values is necessary to maximize the detection of CH cases. However, the lack of agreement on TSH cutoff values in the literature makes this task difficult. Since November 2009, the TSHneo value of 5.0 µIU/mL has been the cutoff point adopted by the NSP in Mato Grosso (MT), Brazil. Consequently, the present study aimed to assess the CH detection ability of several TSHneo cutoff values and their effects on the current screening program.

Methods**Study design and population**

This was a transversal study nested in a cohort study, based on retrospective information collected from the database of the Reference Service for Newborn Screening (RSNS) of MT for all live births, 2010–2012, that were assessed by the NSP-MT. Data were also collected from the clinical records of all individuals with all forms of CH.

The infants included in the study were divided in two groups: Group I – control group ($n = 220$), infants with a normal newborn screening test (NST), and Group II – study group ($n = 44$), infants with confirmed CH, *i.e.*, serum TSH higher than 10.0 µIU/mL and normal or low free thyroxine (T4) levels, according to the laboratory reference values. For every

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