CLINICAL AND LABORATORY STANDARDS INSTITUTE®

1st Edition

CLSI NBS10™

Newborn Screening for Congenital Hypothyroidism

CLSI NBS10 describes a newborn screening system for detecting congenital hypothyroidism (CH). It discusses both first-tier and second-tier screening tests performed on newborn dried blood spot specimens, as well as screening strategies for identifying newborns at increased risk for CH.

A guideline for global application developed through the Clinical and Laboratory Standards Institute consensus process.

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Newborn Screening for Congenital Hypothyroidism

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Abstract

Clinical and Laboratory Standards Institute NBS10—*Newborn Screening for Congenital Hypothyroidism* describes newborn screening (NBS) processes used worldwide to identify newborns at increased risk for congenital hypothyroidism (CH). CH is one of the most common diseases detected by NBS, occurring in as many as 1 in 2000 live births and even more frequently in iodine-deficient areas. Presymptomatic detection through NBS can lead to early diagnosis and treatment, which reduces or eliminates the permanent intellectual disability and growth failure that occurs in individuals with untreated CH. CLSI NBS10 describes the laboratory screening tests for detecting primary CH, as well as the various laboratory screening algorithms in use, including the advantages and disadvantages of each. It also describes other types of CH that may be detected by NBS, such as central, transient, or subclinical CH. CLSI NBS10 is intended for use by health care providers, birthing facilities, NBS laboratories, regulatory agencies, public health policy makers, and manufacturers of instruments, reagents, and related NBS products.

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Contents

Contents (Continued)

Foreword

The goal of newborn screening (NBS) is presymptomatic detection of at-risk newborns, generally through use of dried blood spot (DBS) specimens that are analyzed in specialized NBS laboratories. Ideally, NBS systems provide well-organized, highly effective, population-based services that apply preventive medicine principles to reduce death and disability from many congenital diseases or disorders. NBS systems should be comprehensive. They may include health care providers (HCPs), birthing facilities, public health programs, policy makers, insurers, and families, among others. NBS programs should be linked to follow-up HCPs for rapid diagnosis and initiation of treatment. NBS systems encompass preanalytical, analytical, and postanalytical activities, which include education; collection and laboratory analysis of DBS specimens; results reporting; referral to clinical care (short-term follow-up); diagnosis, intervention, programmatic evaluations, and evaluation of health outcomes (long-term follow-up); quality assurance; and quality improvement.

Congenital hypothyroidism (CH) is one of the most common diseases detected by NBS, occurring in as many as 1 in 2000 live births in most populations and even more frequently in iodine-deficient areas. Presymptomatic detection through NBS leads to early diagnosis and treatment, which drastically reduces or eliminates the permanent intellectual disability and growth failure that occurs in children with untreated CH. Since the 1970s, NBS for CH has been introduced in many countries. Presently, all high-resource and many low-resource jurisdictions screen newborns for CH. However, two-thirds of the world's newborns are not screened.[1] In those locations, including places where iodine deficiency makes CH more common, newborns with CH are usually not detected or treated early. The economic and social burden of intellectual disability due to CH remains a public health challenge.

CLSI NBS10 describes laboratory screening algorithms for CH detection that use thyrotropin (ie, thyroid-stimulating hormone), total thyroxine (T4), or both as first-tier screening tests. It also summarizes variations in screening protocols, including measurement of free T4 and thyroxine-binding globulin and point-of-care testing. The advantages and disadvantages of each strategy are explained. This globally applicable guideline is intended to help new and existing programs evaluate and refine procedures and practices for all aspects of CH NBS.

NOTE: The content of this guideline is supported by the CLSI consensus process and does not necessarily reflect the views of any single individual or organization.

KEY WORDS

congenital hypothyroidism
dried blood spot specimens
newborn screening
thyroid-stimulating hormone
thyrotropin
thyroxine
thyroxine-binding globulin

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Chapter 1

Introduction