

Newborn Screening Tests for Your Baby

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Introduction

New born screening helps identify serious but rare health conditions at birth. All babies in the United States get new born screening. Each state decides which tests are required. Most new born screening test results are normal [1]. If a result is not normal, your baby gets a different kind of test called a diagnostic test. Health conditions that are found early with new born screening often can be treated. Early treatment is important, because it may help prevent more serious health problems for your baby.

New born screening

When your baby is 1 to 2 days old, he has some special tests called new born screening. New born screening checks a baby for serious but rare and mostly treatable health conditions at birth. It includes blood, hearing and heart screening [2]. Your baby can be born with a health condition but may not show any signs of the problem at first. If a health condition is found early with new born screening, it often can be treated. Early treatment is important, because it may help prevent more serious health problems for your baby. All babies in the United States get new born screening. About 4 million babies are screened every year [3].

How does new born screening

There are three parts to new born screening

Blood test: Most new born screening is done with a blood test to check for rare but serious health conditions. A health care provider pricks your baby's heel to get a few drops of blood. He collects the blood on a special paper and sends it to a lab for testing [4]. Blood test results are usually ready by the time your baby is 5 to 7 days old. To find out more about the timeframes for sending blood samples to lab and getting test results back, ask your baby's provider or the hospital staff.

Hearing screening: This test checks for hearing loss. For this test, your provider places tiny earphones in your baby's ears and uses special computers to check how your baby responds to sound [5].

Heart screening: This test is used to screen babies for a group of heart conditions called critical congenital heart defects (also called critical CHDs or CCHDs) [6]. It uses a simple test called pulse oximetry. Pulse oximetry checks the amount of oxygen in your baby's blood by using a pulse oximeter machine and sensors placed on your baby's skin (Figure 1).

New born screening tests

Newborn screening tests look for developmental, genetic, and metabolic disorders in the newborn baby. This allows steps to be taken before symptoms develop. Most of these illnesses are very rare, but can be treated if caught early [7]. The types of newborn screening tests that are done vary from state to state. By April 2011, all states reported screening for at least 26 disorders on an expanded and standardized uniform panel. The most thorough screening panel checks for about 40 disorders. However, because phenylketonuria (PKU) was the first disorder for which a screening test developed, some people still call the new born screen "the PKU test".

In addition to blood tests, screening for hearing loss and critical congenital heart disease (CCHD) is recommended for all newborns. Many states require this screening by law as well. kinds of health conditions does newborn screening test for Each state requires different tests, so ask your baby's health care provider which tests your baby will have. You also can visit babysfirsttest.org to find out what conditions your state tests for. March of Dimes works with states to ensure they are testing babies for the health conditions recommended by Department of Health and Human Services (also called HHS). The list of health conditions recommended by HHS is called RUSP or Recommended Uniform Screening Panel [8]. March of Dimes would like to see all babies in all states screened for at least 35 health conditions. Many

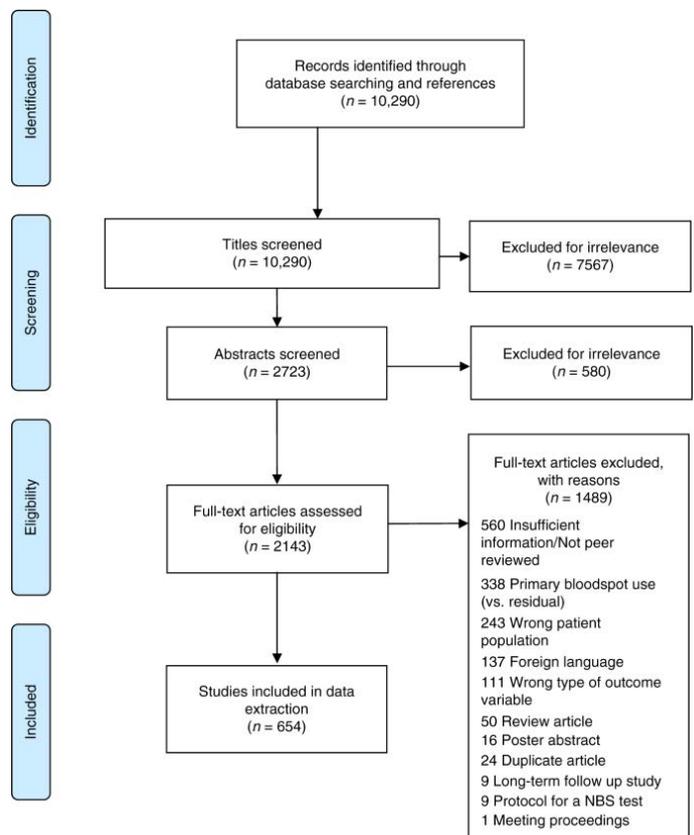


Figure 1: Shows New born Screening Stages.

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of these health conditions can be treated if found early. The health conditions are divided into six groups.

Organic acid metabolism disorders: Metabolism is how well and fast your body processes what you eat and drink. Babies with organic acid metabolism disorders don't break down food correctly, causing substances called organic acids to build up in the body [9].

Propionic acidemia (PROP)

Methylmalonic acidemia (methylmalonyl-CoA mutase deficiency) (MUT)

Methylmalonic acidemia (cobalamin disorders) Cbl A, B)

Isovaleric acidemia (IVA)

3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)

3-hydroxy-3-methylglutaric aciduria (HMG)

Holocarboxylase synthase deficiency (MCD)

Beta-ketothiolase deficiency (BKT)

Glutaric acidemia type 1 (GA-1)

Fatty acid oxidation disorders: During digestion, the body breaks down fat into fatty acids for energy [10]. A baby with fatty acid oxidation problems can't change fat into energy properly.

Carnitine uptake defect/carnitine transport defect (CUD)

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)

Trifunctional protein deficiency (TFP)

Amino acid metabolism disorders: Babies with these problems can't process certain amino acids properly in the body [11]. Amino acids help build protein in your body.

Argininosuccinic aciduria (ASA)

Citrullinemia type 1 (CIT)

Maple syrup urine disease (MSUD)

Homocystinuria (HCY)

Classic phenylketonuria (PKU)

Tyrosinemia type I (TYR I)

Endocrine disorders. These problems affect the glands that make hormones. Hormones are chemicals made by the body [12]. Hormones help with many processes in the body, like growth and development.

Primary congenital hypothyroidism (CH)

Congenital adrenal hyperplasia (CAH)

Hemoglobin disorders. These problems affect red blood cells. Red

blood cells carry oxygen to the rest of the body [13].

S,S disease (sickle cell anemia) (Hb SS)

S, beta-thalassemia (Hb S/βTh)

S, C disease (Hb S/C)

Other disorders

Biotinidase deficiency (BIOT)

Critical congenital heart disease (CCHD)

Cystic fibrosis (CF)

Classic galactosemia (GALT)

Glycogen storage disease type II (Pompe disease) (POMPE)

Hearing loss (HEAR)

Severe combined immunodeficiency (SCID)

Mucopolysaccharidosis type 1 (MPS I)

X-linked adrenoleukodystrophy

Spinal muscular atrophy due to homozygous deletion of exon 7 in SMN1.

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