Newborn Disease Screening Panels

Linden Young, RPh, PGY-1 Pharmacy Resident, Aurora Medical Center Grafton

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Disclosures

The planner(s) and speaker(s) have indicated that there are no relevant financial relationships with any ineligible companies to disclose.

Learning Objectives

At the end of this session, learners should be able to:

- Outline the purpose and public health impact of newborn screening.
- Identify core conditions included in screening panels.
- Recognize the healthcare provider’s role in supporting screening, follow-up care, and medication therapy management.
- Name state-specific differences in screening practices.
- Discuss clinical implications of positive results.

Outline

- Introduction to Newborn Screening
- Core Conditions in Screening Panels
- Interpreting Positive Results
- State-Specific Variations
- Specific Conditions and Pharmaceutical Considerations
- Conclusion and Q&A

Abbreviation Key

- ADHD = Attention deficit hyperactivity disorder
- CAH = Congenital adrenal hyperplasia
- CNS = Central nervous system
- CFTR = Cystic fibrosis transmembrane conductance regulator
- CYP = Cytochrome P450
- DHS = Department of Health Services
- DNA = Deoxyribonucleic acid
- G-6PD = Glucose-6-phosphate dehydrogenase
- GA = Georgia
- GAMT = Guanidinoacetate methyltransferase
- HHS = Health and Human Services
- IL = Illinois
- IT = Intrathecal
- IV = Intravenous
- Kg = Kilogram
- mEq = Milliequivalent
- Mg = milligram
- MSUD = Maple syrup urine disease
- NBS = Newborn screening
- NC = North Carolina
- NGS = Next-generation sequencing
- NICU = Neonatal intensive care unit
- NK = Natural killer
- ODT = Oral disintegrating tablet
- PKU = Phenylketonuria
- PN = Parenteral nutrition
- PO = By mouth
- RBC = Red blood cell
- RUSP = Recommended uniform screening panel
- SMN = Survival motor neuron
- WETRAC = Wisconsin Electronic Tracking and Reporting System
- WI = Wisconsin

Newborn Screening Defined

- Newborn screening:
  - "The process of checking babies to identify those who might have certain serious health conditions that can benefit from early treatment or intervention"
- Newborn screening panel:
  - "A list of conditions for which newborns receive screening at or shortly after birth"

ADVOCATE HEALTH

Glossary | Newborn Screening. Hrsa.gov. Published December 15, 2024. Accessed December 31, 2025. https://newbornscreening.hrsa.gov/about-newborn-screening/glossary#newborn-screening

History of Newborn Screening

- Began in the 1960's in the United States
  - Dr. Robert Guthrie, a microbiologist and pediatrician, developed a blood test for phenylketonuria
    - First example of population-based screening
    - Many people refer to all NBS as the PKU test
      - Not accurate as not all inclusive

Robert Guthrie Symposium 2022. Buffalo.edu. Published July 19, 2023. Accessed January 27, 2026. https://medicine.buffalo.edu/departments/pediatrics/news_and_events/rgs/2022.html

Gleason CA. Avery's Diseases of the Newborn - E-Book. Elsevier Health Sciences; 2023.
Avades and Kokhanov; Neoreviews December 2025; 26 (12): e809–e819
Cao et al. The Journal of Applied Laboratory Medicine, Volume 9, Issue 4, July 2024, Pages 820–832

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Benefits of Early Detection

- Primary congenital hypothyroidism
  - Prevalence: 1 in 3,000
  - If untreated: serious intellectual, developmental, and physical disabilities
  - If treated early: normal development
- Cystic fibrosis
  - Prevalence: 1 in 3,700
  - If untreated: lifelong health problems, lung damage, and possible early death
  - If treated early: longer and healthier life
- Severe combined immunodeficiency
  - Prevalence: 1 in 75,000
  - If untreated: death within 1-2 years after birth
  - If treated early: normal, healthy life

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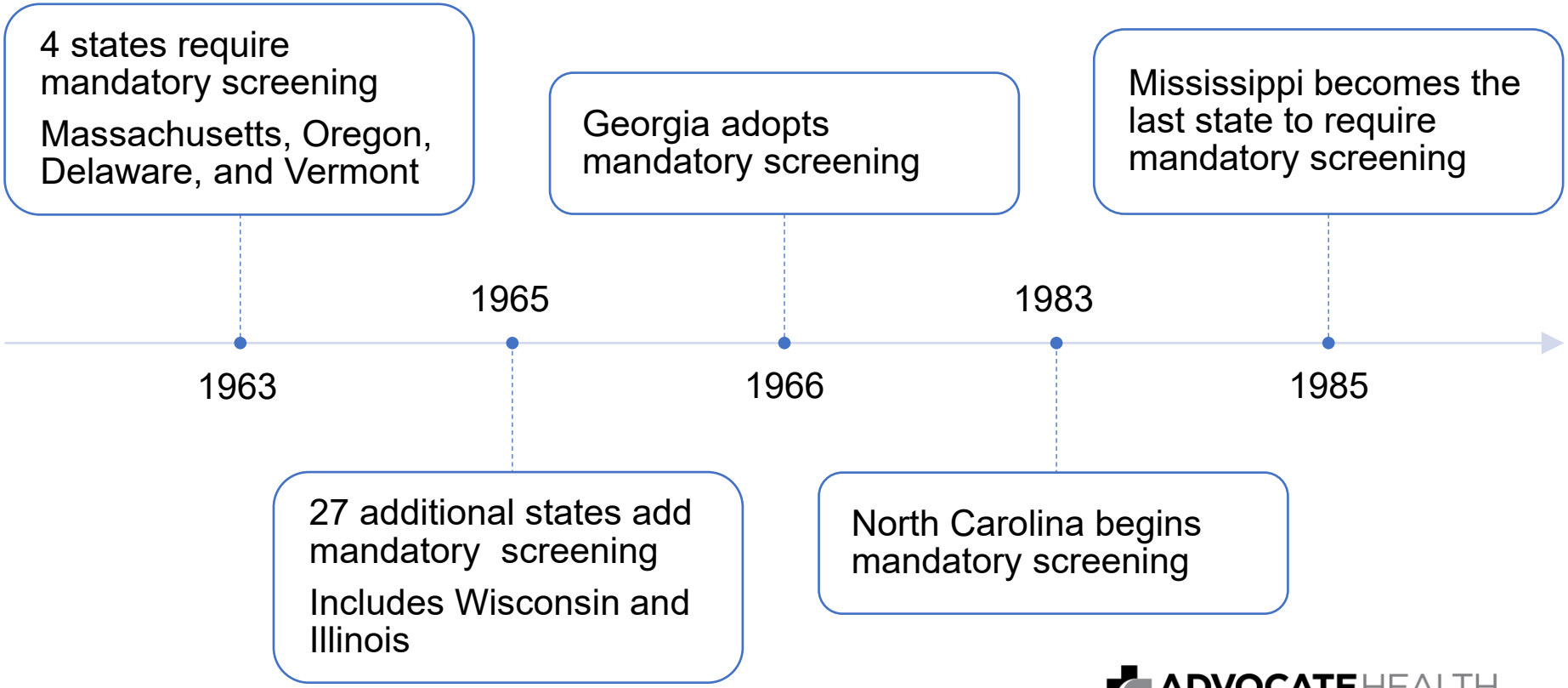
The Newborn Screening Story: How One Simple Test Changed Lives, Science, and Health in America. Association of Public Health Laboratories; 2013.

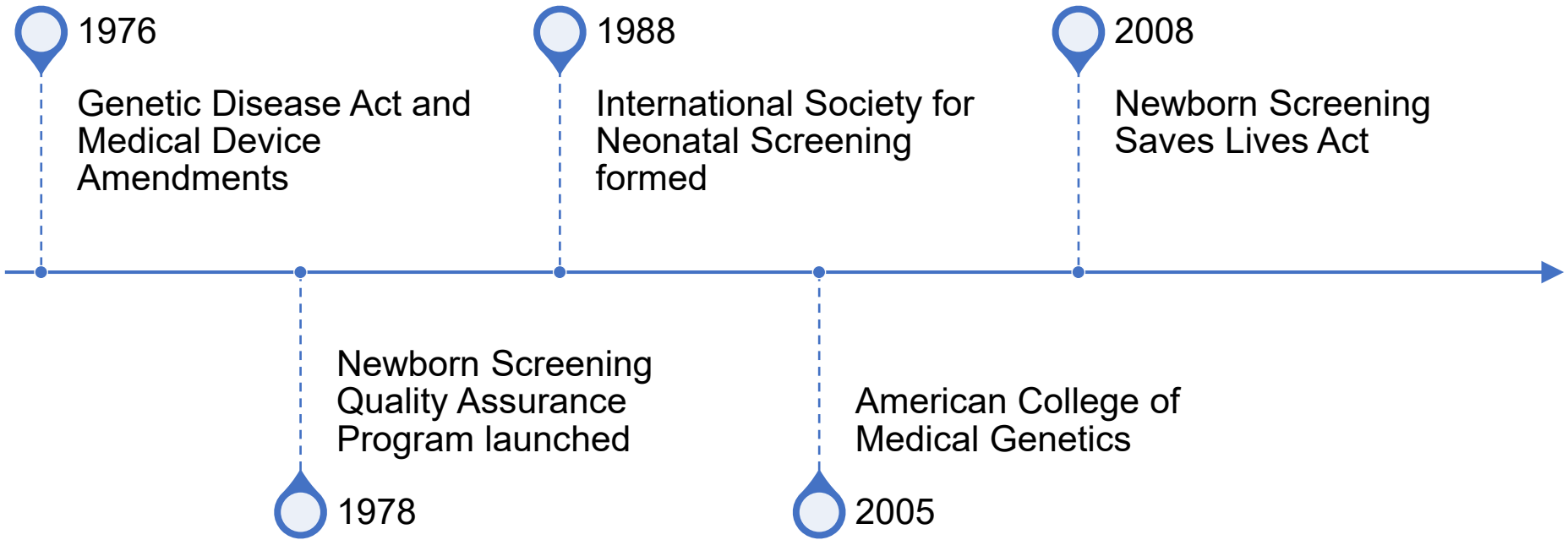
Mandatory Screening Timeline



ADVOCATE HEALTH

The Newborn Screening Story: How One Simple Test Changed Lives, Science, and Health in America. Association of Public Health Laboratories; 2013.

Milestones



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The Newborn Screening Story: How One Simple Test Changed Lives, Science, and Health in America. Association of Public Health Laboratories; 2013.

Assessment Question #1

Blood testing for which disease is credited with initiating modern newborn screening programs?

a. Phenylketonuria
b. Maple syrup urine disease
c. Cystic fibrosis
d. Severe combined immunodeficiency

Newborn Screening Process

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Timing

- Babies need screening during the first few days of life
  - Typically, between 24 and 48 hours after birth
    - Retesting may be needed if before 24 hours
- Situations where timing may be delayed:
  - Transfusion
  - Dialysis
  - Neonates requiring medications/nutrients

Avades and Kokhanov; Neoreviews December 2025; 26 (12): e809–e819
HRSA. Newborn Screening Process | Newborn Screening. December 2024. Accessed January 5, 2026. https://newbornscreening.hrsa.gov/newborn-screening-process.

State Law

- Law varies according to each state
- General considerations:
  - May require all babies born in hospital to have a newborn screening before leaving the hospital
  - Babies born at home must be tested within a week (ideally within a few days)
  - Screening may be refused for religious beliefs/practices or personal views
  - Refusal is typically documented

Wisconsin legislature: 253.13. December 19, 2025. Accessed January 5, 2026.
Newborn screening: Blood screening for newborns. Wisconsin Department of Health Services. October 24, 2025. Accessed January 6, 2026.

NBS Models

- One-screen model
  - Babies screened once between 24-48 hours old
  - Looks for all conditions on the state's NBS panel
  - Used by most states (includes WI, NC, GA and IL)
- Two-screen model
  - All babies are screened at 24-48 hours old and again at 1-2 weeks old as part of a well-baby visit
  - Used by thirteen states

HRSA. Newborn Screening Process | Newborn Screening. December 2024. Accessed January 5, 2026.
Newborn Screening (NBS) | Georgia Department of Public Health. Georgia.gov. Published 2019.
A Baby's First Step in Life - A Newborn Screening Guide for Parents. dph.illinois.gov.
Resources for Parents | Wisconsin State Laboratory of Hygiene. Wisc.edu. Published 2026. Accessed January 27, 2026.
NC DPH. N.C. DPH: State Lab > Newborn Screening. Ncdhhs.gov. Published 2019.

Parts of Screening

- Blood spot screening
- Pulse oximetry (critical congenital heart disease) screening
- Hearing screening

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HRSA. Newborn Screening Process | Newborn Screening. December 2024. Accessed January 5, 2026.

Blood Spot Screening

- Also called "heel stick" or "24-hour test"
  - Newborn's heel is given a pinprick
  - A few drops of blood are collected on a card
  - The card is sent to the laboratory for testing
- Determines if a baby might have a serious condition
- Disorders screened varies widely by state
- Results
  - In-range (negative, normal, low risk)
  - Out-of-range (positive, abnormal, high risk)
  - Borderline (inconclusive, medium risk)

HRSA. Newborn Screening Process | Newborn Screening. December 2024. Accessed January 5, 2026. https://newbornscreening.hrsa.gov/newborn-screening-process.
Wisconsin. Wisconsin | Newborn Screening. September 2025. Accessed January 6, 2026. https://newbornscreening.hrsa.gov/your-state/wisconsin.
Newborn Blood Screening Panel of Diseases. Wisconsin State Laboratory of Hygiene. Accessed January 6, 2026. https://www.slh.wisc.edu/clinical/newborn/health-care-professionals-guide/nbs-test-panel-of-diseases/

Pulse Oximetry Screening

- Determines if a newborn might have critical congenital heart disease
- Group of serious heart conditions present at birth
- More testing may be required:
  - Echocardiogram, electrocardiogram, chest X-ray
- Results
  - Pass
  - Fail

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HRSA. Newborn Screening Process | Newborn Screening. December 2024. Accessed January 5, 2026.

Hearing Screening

- Done any time after 12 hours of life
  - Performed in a quiet room when baby is calm
- Two types of hearing screening
  - Otoacoustic emissions
  - Automated auditory brainstem response
- Both uses earbuds to test baby's hearing
- Results
  - Pass
  - Fail

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HRSA. Newborn Screening Process | Newborn Screening. December 2024. Accessed January 5, 2026.
Newborn Hearing Screening. American Academy of Audiology. April 11, 2022. Accessed January 6, 2026

Assessment Question #2

Which of the following are not part of the recommendation for NBS?

a. Critical Congenital Heart Disease Screening
b. Hearing Screening
c. Blood Spot Screening
d. Pulmonary Screening

Recommended Uniform Screening Panel

- List of disorders that the HHS recommends for states to screen as part of NBS programs
- Standardized list of disorders
- Conditions listed must be covered by non-grandfathered health plans
- 38 Core conditions
- 26 Secondary conditions

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Recommended Uniform Screening Panel. HRSA. July 2024. Accessed January 6, 2026.

Challenges of Screening

- False-positives
  - Examples: congenital adrenal hyperplasia, congenital hypothyroidism, galactosemia, MSUD, cystic fibrosis
- False-negatives
  - If sample is taken too early, can be difficult to detect the level of metabolic activity in certain conditions
    - Examples: PKU, MSUD
- Repeat testing recommended if done at <24 hours of age

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Avades and Kokhanov; Neoreviews December 2025; 26 (12): e809–e819

Provider Follow-up

- WI: WE-TRAC
  - Available for birth providers and audiologists
  - Follow up on missing hearing screening and heart screening results
- GA: multiple programs responsible for follow-up depending on condition
- IL: Newborn Screening Follow-Up Program
- NC: Newborn Screening Program

Newborn Screening Program. Wisconsin Department of Health Services. November 28, 2025. Accessed January 6, 2026
Newborn Screening Reports. Illinois.gov. Published 2025. Accessed January 27, 2026.
Newborn Screening Services | NCDHHS. Ncdhhs.gov. Published 2025.
Newborn Screening (NBS) | Georgia Department of Public Health. Georgia.gov. Published 2019.

Assessment Question #3

Which of the following is true regarding software platforms such as WE-TRAC?

A. They are used to help providers follow up with positive newborn screening results.
B. They are used to schedule routine well-child visits.
C. They are designed to track adult vaccine records.
D. They are used solely for billing of newborn screening.

State-Specific Panels

State	Number of Conditions	Missing Core Conditions
Georgia	37	Early-Onset Metachromatic Leukodystrophy
Illinois	66	Duchenne Muscular Dystrophy GAMT Deficiency
North Carolina	60	GAMT Deficiency Infantile Krabbe Disease Mucopolysaccharidosis Type II
Wisconsin	51	GAMT Deficiency Infantile Krabbe Disease Mucopolysaccharidosis Type II

Newborn Blood Screening Panel of Diseases. Wisconsin State Laboratory of Hygiene. Accessed January 6, 2026.
Recommended Uniform Screening Panel. HRSA. July 2024. Accessed January 6, 2026.
North Carolina | Newborn Screening. December 2025, Accessed January 26, 2026. newbornscreening.hrsa.gov.
Georgia | Newborn Screening. newbornscreening.hrsa.gov. Published December 2025. Accessed January 27, 2026.
Illinois | Newborn Screening. Hrsa.gov. Published July 5, 2024. Accessed January 27, 2026.

Please fill out the form entirely with **LEGIBLE** and **ACCURATE** information

FORM EXPIRATION DATE: 2026-09-30

WI State Laboratory of Hygiene
465 Henry Mall
Madison WI 63706

UXXXXXX

Submitter Use/Label

Baby's Name (LAST / FIRST)
LAST FIRST

Multiple births (Twin, etc.) #____ OF ____

SEX F M

Baby's Birthdate Time (Military)
MM / DD / YY :

Baby's ID # (MRN/Alt. ID)

Baby's Primary Care Provider NPI#
LAST FIRST

Specimen Collection Date Time (Military)
:

PCP's Clinic Name

City Phone # ( )

☐Mother OR ☐Guardian Name (LAST / FIRST):
LAST FIRST

Ordering Physician NPI#
LAST FIRST

Weight at birth (grams)
g

Gestational age at birth
wks days

Baby's Race (Circle all that apply)
Black Native American White Asian/Pacific Isle

Hispanic? N Y

Baby in ICU? N Y ☐Collection at Discharge

Repeat Specimen? N Y

Transfusion(s)? (any product) N Y
Last Txn Date/Time:

Baby on TPN now? N Y

Place of Birth (required)
NAME CITY / STATE

Mother's Hepatitis B Surface Antigen: (HBsAg): Neg Pos

J. Schauer, Director
E. Rider, CAP Director
WSS 253-13 HYG:213

Blood Not Submitted (mark reason)
☐ Declined ☐ Transferred ☐ Deceased
☐ Other

Hearing Screen Date
MM / DD / YY

Pulse Ox Screen Date Time (Military) ☐ Pass ☐ Fail
MM / DD / YY :

Not Screened (mark reason) ☐ Declined ☐ Transferred ☐ Deceased
☐ Echo normal ☐ Confirmed heart disease ☐ Other

Circle Hearing Screen Method
ABR OAE BOTH

Right Ear ☐ Pass ☐ Refer

Left Ear ☐ Pass ☐ Refer

Hearing Not Screened (mark reason)
☐ Transferred ☐ NICU ☐ Declined
☐ Deceased ☐ Other

This box for Newborn Screening Laboratory USE ONLY

PARENTS COPY

COMPLETELY FILL ONE CIRCLE AT A TIME

FILL ALL FIVE CIRCLES APPLY TO ONE SIDE ONLY

Wisc.edu. Published 2022. Accessed January 27, 2026. https://www.slh.wisc.edu/wp-content/uploads/2022/12/NBS-Blank-Card_2022.jpg

Nominating a Condition

- Healthcare providers can nominate a condition to add to the state-specific newborn screening panel
  - For example: In WI, 9 criteria must be included for the condition
- Can also nominate for the RUSP
  - Includes answering 15 questions regarding the condition, screening, and impact of screening
  - Similar 9 criteria for consideration

Newborn Screening: Nominate a Condition for the Newborn Screening Panel. Wisconsin Department of Health Services. November 11, 2025. Accessed January 6, 2026.
Nominate a Condition. HRSA. July 2024. Accessed January 6, 2026.

Examples: Recent WI Nominations

- Critical congenital heart disease (Added 7/3/2014)
- Krabbe disease (Nominated twice, not added to panel)
- Carnitine palmitoyl transferase 1A (added 1/2020)
- Spinal muscular atrophy (Added 1/2020)
- Pompe disease (Added 5/2/2020)
- X-linked adrenoleukodystrophy (Added 8/1/2025)
- Mucopolysaccharidosis 1 (Added 8/1/2025)

Newborn Screening: Nominate a Condition for the Newborn Screening Panel. Wisconsin Department of Health Services. November 11, 2025. Accessed January 6, 2026.
Nominate a Condition. HRSA. July 2024. Accessed January 6, 2026.
Newborn Screening Program: Committee Structure. Wisconsin Department of Health Services. November 11, 2025. Accessed January 6, 2026.

Assessment Question #4

Which of the following statements is true regarding state-specific screening practices?

a. All states must adopt the complete RUSP and may add additional disease states to their panel
b. All states have identical screening panels
c. States are required to repeat newborn screening 1-2 weeks after birth
d. Each state develops its own panel used for screening

Special Situations

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NBS in the NICU[3]

- Optimal timing remains 24-48 hours after birth
- Use of PN and lack of early enteral feeding can alter NBS results
  - Namely amino acid disorders
- Corticosteroids may also lead to false-negative results
  - Includes those given to the mother
- Most NICUs have their own protocol based on state guidance

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Avades and Kokhanov; Neoreviews December 2025; 26 (12): e809–e819

Primary Literature: Effect of Steroids on Screening

- "Effect of Single and Multiple Courses of Prenatal Corticosteroids on 17-Hydroxyprogesterone Levels: Implication for Neonatal Screening of Congenital Adrenal Hyperplasia"
- Purpose: Assess effect of antenatal steroid administration on false-negatives with CAH screening
- Results: Significant negative correlation between blood 17-hydroxyprogesterone levels and cumulative betamethasone dose ($r = -0.19$, $p < 0.05$)
  - Only statistically significant difference between group whose mother received multiple courses of betamethasone and those without treatment ($p < 0.05$)

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Gatelais *et al. Pediatr Res* **56**, 701–705 (2004)

Blood Spot Screening Examples with Pharmaceutical Considerations

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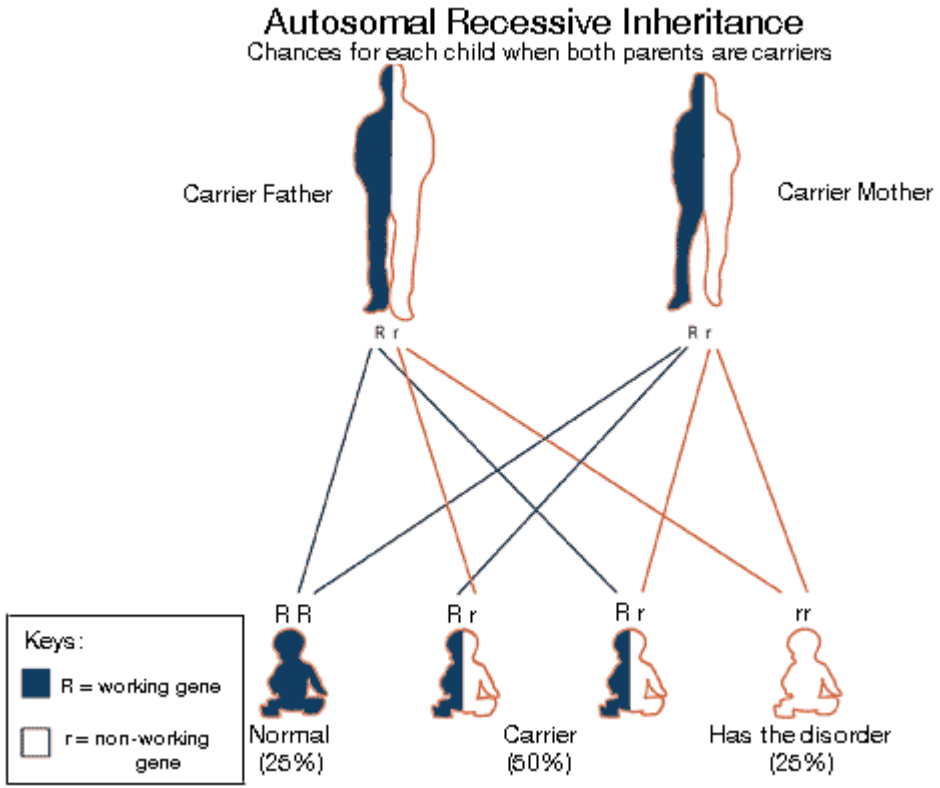

CBS (homocystinemia/cystathionine beta-synthase deficiency).newbornscreening.info. March 1, 2023. Accessed January 6, 2026.

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Carrier Screening

- Genetic testing that tells you if you carry a gene for a genetic disorder
  - Provides chances of having child with a genetic disorder
- This testing focuses on recessive disorders
  - No symptoms of disease
- Can be performed prior to or during pregnancy
- Two types
  - Targeted carrier screening
  - Expanded carrier screening

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American College of Obstetricians and Gynecologists. Carrier Screening. www.acog.org. Published December 2020. Accessed January 27, 2026.

Metabolic Disorders

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Phenylketonuria

- First disorder for which NBS was implemented
- Autosomal recessive disorder resulting from deficiency of phenylalanine hydroxylase
- Cardinal screening feature: increased level of phenylalanine
  - Causes CNS injury in excess
- Symptoms:
  - Seizures, irritability, eczema, musty body odor, lighter hair/skin than family members, intellectual disability
- Reliably identified as early as the first day after birth

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Gleason CA. Avery's Diseases of the Newborn - E-Book. Elsevier Health Sciences; 2023.

Pharmaceutical Considerations

- Avoid medications with phenylalanine
- Phenylalanine listed in the package insert
- Non-prescription products must list phenylalanine on the outside label
  - Phenylalanine content is equal to half the amount of aspartame
- Common products: chewable/ODT
- Sapropterin dihydrochloride (Kuvan)
  - Initial dose: 10-20 mg/kg PO once daily

Gleason CA. Avery's Diseases of the Newborn - E-Book. Elsevier Health Sciences; 2023
What are common treatments for phenylketonuria (PKU)? Eunice Kennedy Shriver National Institute of Child Health and Human Development. January 16, 2024. Accessed January 6, 2026.
Drug products containing phenylalanine. National PKU News. 2007. Accessed January 6, 2026.
Sapropterin. Lexi-Drugs. UpToDate Lexidrug. UpToDate Inc. https://online.lexi.com. Accessed January 6, 2026.

Maple Syrup Urine Disease

- Rare autosomal recessive inborn error of amino acid metabolism cause by branched-chain amino-ketoacid dehydrogenase complex deficiency
- Primary indicators: increased leucine level
- Screening can miss mild variants
- Can present acutely in the first week of life
  - All high probability positive screens require immediate action
- Symptoms:
  - Maple syrup smell in earwax, sweat, or urine; poor feeding; lethargy; seizures

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HRSA. Maple Syrup Urine Disease | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Avoid branched chain amino acids (leucine, isoleucine, valine)
  - Caution with products such as protein supplements, amino acid infusions, and parenteral nutrition formulas
- Caution with drugs that promote catabolism
  - Corticosteroids, vasoactive catecholaminergic agents
- Increased risk for ADHD, depression, anxiety
- Thiamine supplementation for thiamine-responsive disease
  - Start with 4-week trial of enteral thiamine (50-100 mg/day PO divided twice daily)

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HRSA. Maple Syrup Urine Disease | Newborn Screening. September 2025. Accessed January 6, 2026.
Strauss et al. 2006 Jan 30 [Updated 2020 Apr 23]. In: Adam MP, Bick S, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025.

Homocystinuria

- Rare autosomal recessive inherited disorder where methionine is not broken down due to issues with cystathionine beta-synthase
- Two types
  - B-6 responsive
  - B-6 non-responsive
- Symptoms: poor growth, developmental delays

HRSA. Homocystinuria | Newborn Screening. September 2025. Accessed January 6, 2026.
CBS (homocystinemia/cystathionine beta-synthase deficiency). newbornscreening.info. March 1, 2023. Accessed January 6, 2026.

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Pharmaceutical Considerations

- Low methionine diet
- Vitamin B6
  - Dose: 10 mg/kg/day PO (up to 500 mg/day)
- Betaine
  - Dose: 50 mg/kg/dose PO twice daily
- Vitamin B12
- Folic acid
- L-cysteine
- Pregnancy increases risk of blood clots, stroke, and heart disease in women with homocystinuria – blood thinner during last few months of pregnancy until 6 weeks after delivery

HRSA. Homocystinuria | Newborn Screening. September 2025. Accessed January 6, 2026.
CBS (homocystinemia/cystathionine beta-synthase deficiency). newbornscreening.info. March 1, 2023. Accessed January 6, 2026.
Betaine. Lexi-Drugs. UpToDate Lexidrug. UpToDate Inc. https://online.lexi.com. Accessed January 26, 2026.
Gerrard and Dawson. Journal of Clinical Pathology 2022;75:744-750.

Endocrine Disorders

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Congenital Adrenal Hyperplasia

- Autosomal recessive condition
- Several Types
  - Type detected in NBS alters CYP21A2 gene, which means sufficient cortisol and aldosterone are not made
- Affect function of adrenal glands – makes the adrenal gland grow very large
- Most often have salt-wasting
- Symptoms:
  - Poor feeding, vomiting, lethargy, weight loss

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HRSA. Congenital Adrenal Hyperplasia | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Hydrocortisone to replace missing cortisol
  - Initial dosing: 8-15 mg/m^2/day PO in 3 divided doses
- Fludrocortisone to maintain proper salt and fluid levels
  - Dosing: 0.05 to 0.2 mg PO daily in 1 or 2 divided doses
- Sodium chloride
  - Normal daily maintenance sodium requirements: 2 to 5 mEq/kg/day PO

HRSA. Congenital Adrenal Hyperplasia | Newborn Screening. September 2025. Accessed January 6, 2026.
Hydrocortisone. Lexi-Drugs. UpToDate Lexidrug. UpToDate Inc. https://online.lexi.com. Accessed January 6, 2026.
Fludrocortisone. Lexi-Drugs. UpToDate Lexidrug. UpToDate Inc. https://online.lexi.com. Accessed January 6, 2026.
Sodium chloride. Lexi-Drugs. UpToDate Lexidrug. UpToDate Inc. https://online.lexi.com. Accessed January 26, 2026.

Congenital Hypothyroidism

- Thyroid gland absent/small or just does not produce enough thyroid hormone
- In most cases, the condition is random (not genetic)
- Inherited in 15-20% of cases
- Many babies with down syndrome also have congenital hypothyroidism
- Symptoms:
  - Slow growth, floppy arms/legs, lethargy, difficulty feeding, constipation, jaundice

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HRSA. Congenital Hypothyroidism | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Replace thyroid hormone
  - Levothyroxine dose for 1-3 months: 10 to 15 mcg/kg/dose PO once daily
- Limit soy
- Limit iron

HRSA. Congenital Hypothyroidism | Newborn Screening. September 2025. Accessed January 6, 2026.
Levothyroxine. Lexi-Drugs. UpToDate Lexidrug. UpToDate Inc. https://online.lexi.com. Accessed January 6, 2026.

Other Disorders

Cystic Fibrosis

- Inherited condition that causes thick/sticky mucus buildup in the body
- Caused by change in CFTR gene (controls chloride channels), unable to properly balance salt
- Leads to fluid-filled sacs and scar tissue in organs
- Caused by salt imbalance (excess fluid some places, absent in others)
- Symptoms:
  - Salty sweat, thick mucus, frequent cough/wheeze, diarrhea

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HRSA. Cystic Fibrosis | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Pancreatic enzymes
- Vitamin supplements
- Antibiotics
- Anti-inflammatories
- Mucus thinners
- Bronchodilators
- CFTR gene targets

HRSA. Cystic Fibrosis | Newborn Screening. September 2025. Accessed January 6, 2026.
Gene therapy for cystic fibrosis. Cystic Fibrosis Foundation. Accessed January 6, 2026.

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Sickle Cell Disease

- Inherited condition (*HBB* gene) that affects hemoglobin
- Normal hemoglobin is replaced with hemoglobin S
- In extreme conditions, sickle hemoglobin turns RBCs into sickle/crescent shapes
- Difficult time carrying oxygen to body
- Symptoms:
  - Difficulty breathing, anemia, large spleen, serious infections

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HRSA. S,S Disease (Sickle Cell Anemia) | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Hydroxyurea
  - Initial dosing: 15-20 mg/kg/dose PO once daily
- Daily antibiotics
- Analgesics
- Blood transfusions

HRSA. S,S Disease (Sickle Cell Anemia) | Newborn Screening. September 2025. Accessed January 6, 2026.
Hydroxyurea. Lexi-Drugs. UpToDate Lexidrug. UpToDate Inc. https://online.lexi.com. Accessed January 6, 2026.
Yates et al. Section on Hematology/Oncology, AMERICAN SOCIETY OF PEDIATRIC HEMATOLOGY/ONCOLOGY; Health Supervision for Children and Adolescents With Sickle Cell Disease: Clinical Report. Pediatrics August 2024; 154 (2): e2024066842. 10.1542/peds.2024-066842

Severe Combined Immunodeficiency

- Group of genetic conditions where baby is born without working immune system
- Usually inherited, can spontaneously develop
- T, B, and NK cells absent/not working correctly
- X-linked = bubble boy disease
- Symptoms
  - Many severe infections or atypical infections, infections that don't improve with antibiotics, chronic diarrhea

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HRSA. Severe Combined Immunodeficiencies | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Isolation
- Antibody treatment (immunoglobulin replacement)
- Antimicrobial therapy
- Bone marrow transplant
- Thymus transplant
- Gene therapy

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HRSA. Severe Combined Immunodeficiencies | Newborn Screening. September 2025. Accessed January 6, 2026.

Spinal Muscular Atrophy

- Inherited autosomal recessive condition that affects the motor neurons (SMN1 gene deletion)
- Body cannot make enough survival motor neuron protein which leads to motor neuron death
- Eventually results in muscle atrophy
- Symptoms
  - Weakness, hypotonia, poor growth

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HRSA. Spinal Muscular Atrophy | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Nusinersen (Spinraza)
  - Loading dose: 12 mg IT once every 14 days for 3 doses then 12 mg IT 30 days after the 3rd dose
  - Maintenance dosing: 12 mg IT once every 4 months
- Risdiplam (Evrysdi)
  - Dosing: 5 mg PO once daily
- Gene therapy
- Clinical trials

HRSA. Spinal Muscular Atrophy | Newborn Screening. September 2025. Accessed January 6, 2026.
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G-6PD Deficiency

- Inherited condition with low levels of G-6PD
- Protects RBCs from reactive oxygen species
- Causes RBCs to break down too quickly
- X-linked recessive pattern
- Symptoms
  - Jaundice, fatigue, shortness of breath, tachycardia

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HRSA. Glucose-6-Phosphate Dehydrogenase Deficiency | Newborn Screening. September 2025. Accessed January 6, 2026.

Pharmaceutical Considerations

- Phototherapy
- Blood transfusion
- Avoidance of certain medications
  - Includes:
    - Nitrofurantoin, ciprofloxacin, moxifloxacin, ofloxacin, sulfamethoxazole/trimethoprim, sulfasalazine, methylene blue, dapsone, doxorubicin, and others

HRSA. Glucose-6-Phosphate Dehydrogenase Deficiency | Newborn Screening. September 2025. Accessed January 6, 2026.
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Assessment Question #5

What medications put someone with G6PD deficiency at increased risk of hemolytic anemia (select all)?

a. Moxifloxacin
b. Doxycycline
c. Indomethacin
d. Dapsone

Summary/Conclusion

- Newborn screening is an essential part of early childhood healthcare to detect disease and prevent serious complications
- Screening now includes blood spot screening, critical congenital heart disease screening, and hearing screening
- There are many examples of diseases on the newborn screening panel with pharmaceutical considerations ranging from medication avoidance to up-and-coming medication treatments

Future Direction of NBS

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G-6PD Deficiency Testing

- Only New York, Pennsylvania and Washington DC included G-6PD deficiency DNA screening in their routine panels
  - Mandated in New York in 2022 for high-risk scenarios
- Can take up to a week to result
- Performed in other countries which has reduced rates of acute bilirubin encephalopathy and kernicterus

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Avades and Kokhanov; Neoreviews December 2025; 26 (12): e809–e819

Primary Literature: G-6PD Screening

- “Outcomes of Universal Newborn G6PD Deficiency Screening in a Large Urban Cohort”
- Purpose: Explore outcomes of newborns impacted by G6PD during
- Results: G6PD-deficient infants were more likely to require phototherapy during hospitalization following birth
  - Well nursery: 21.3% for G6PD-deficient infants vs 9.1% for G6PD-sufficient infants ($p < .001$)
  - NICU: 90.9% for G6PD-deficient infants vs 36.4% for G6PD-sufficient infants ($p < .001$)
  - G6PD-deficient infants were also more likely to be readmitted for phototherapy
    - 5.7% for G6PD-deficient infants vs 1.4% for G6PD-sufficient infants ($p = .04$)

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Katherine Dalldorf et al.; *Pediatrics* January 2026; 157 (1)

Next Generation Sequencing (NGS) Technology

- Increased use of NGS technology to screen for wider range of genetic disorders
- Earlier detection of difficult-to-diagnose conditions
- Advantages
  - Faster sequencing, broader sequencing range, higher sensitivity, greater precision
- Disadvantages
  - Increased cost, ethical considerations, availability
- Possible complement to traditional screening

Avades and Kokhanov; Neoreviews December 2025; 26 (12): e809–e819
Carli et al. JAMA Netw Open. 2025;8(10):e2538198

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Questions?

Linden Young, linden.young@aah.org