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Newborn Screening for Congenital Cytomegalovirus: A 1 Year Pilot

Sarah Bradley, MS, CGC

Follow-up Supervisor

New York State Newborn Screening Program

Outline

- Overview of NYS Newborn Screening Program
 - Background
 - NBS process
- Responsibilities of hospitals and pediatricians re: newborn screening
- Congenital Cytomegalovirus (cCMV)
 - Why screen for cCMV?
 - Statewide screening from late September 2023 – September 2024
 - Opt-out mechanism
 - Role of hospitals
 - Possible results
 - Follow-up of babies who screen positive
- Questions?



Newborn Screening in New York State



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Newborn Screening (NBS)

- Public health program
- Screens all newborns in a given population for certain medical disorders.
- Identifies a subset of newborns that require more specific testing.
- NYS Public Health Law § 2500-a (and Title 10 NYCRR Subpart 69-1) mandates screening for all newborns.
 - NBS is mandated in all 50 states.



New York State NBS Facts

- We screen ~215,000 babies per year.
 - ~3% are abnormal (presumptive positive or referral).
- We test for 50 different conditions, including:
 - Endocrine disorders: congenital hypothyroidism and congenital adrenal hyperplasia (CAH)
 - Inherited metabolic disorders
 - Hemoglobinopathies
 - Exposure to human immunodeficiency virus (HIV)
 - Severe Combined Immunodeficiency (SCID)
 - Other genetic conditions: Cystic Fibrosis, Pompe Disease, X-linked Adrenoleukodystrophy, Mucopolysaccharidosis Type 1 (MPS 1), Spinal Muscular Atrophy
- Newborn screening is a **free** public health service in New York State!



The Testing Process

- Specimen collection
 - Dried blood spots collected on filter paper.
 - Ideal timeframe is 24 – 36 hours after birth.
 - Collected by birth facility or birth attendant.
- Specimens are dried and sent to the lab via UPS, USPS, FedEx or courier ASAP.



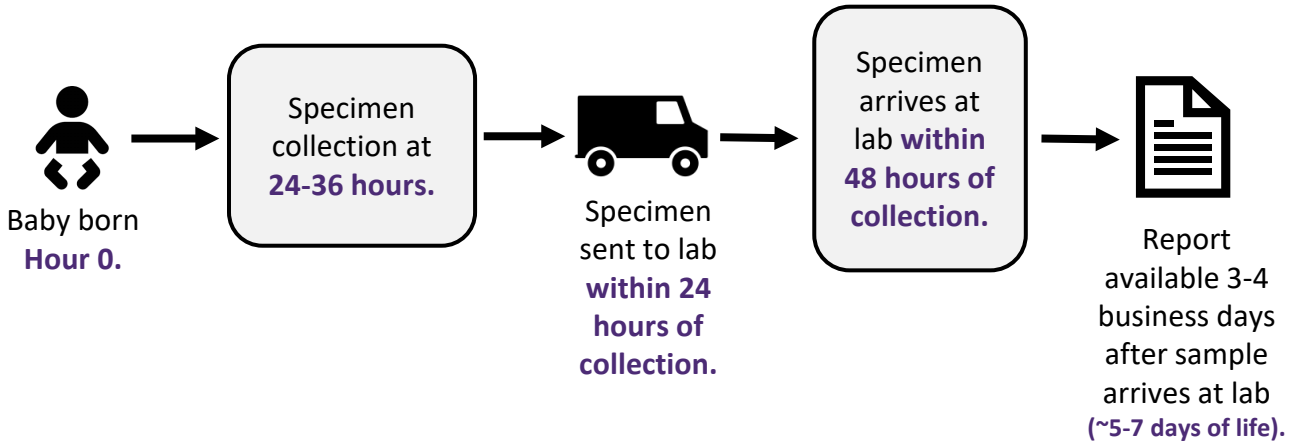
The Testing Process

Overview:

- Specimens arrive and are sorted.
- Accession numbers are assigned.
- Data entry staff enter demographic data.
- Specimens are punched, cut and sent to each lab for testing.
- Sub-lab sections:
 - Inherited Metabolic Diseases
 - Lysosomal Storage Disorders
 - Immunoassay
 - Hemoglobin/HIV
 - DNA
- Testing completed 3-4 days after sample arrives.

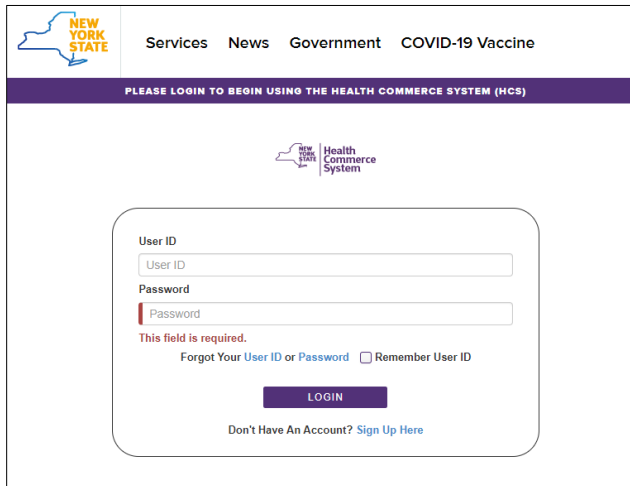


Newborn Screening Timeline



Newborn Screening Process

Reports are available on the NBS app on the Health Commerce System (HCS) as soon as the final report is generated, usually within 4 business days.



The screenshot shows the login interface for the Health Commerce System (HCS). At the top left is the New York State logo. To its right are navigation links: Services, News, Government, and COVID-19 Vaccine. Below these is a purple banner with the text "PLEASE LOGIN TO BEGIN USING THE HEALTH COMMERCE SYSTEM (HCS)". The main content area features the "New York Health Commerce System" logo. Below the logo is a login form with the following elements:

- A "User ID" label above a text input field.
- A "Password" label above a password input field.
- A red asterisk and the text "This field is required." below the password field.
- Links for "Forgot Your User ID or Password" and a checkbox for "Remember User ID".
- A purple "LOGIN" button.
- A link for "Don't Have An Account? Sign Up Here" at the bottom.

Tip: If you do not have access to look up NBS reports, email nbsinfo@health.ny.gov with your HCS username to request access.

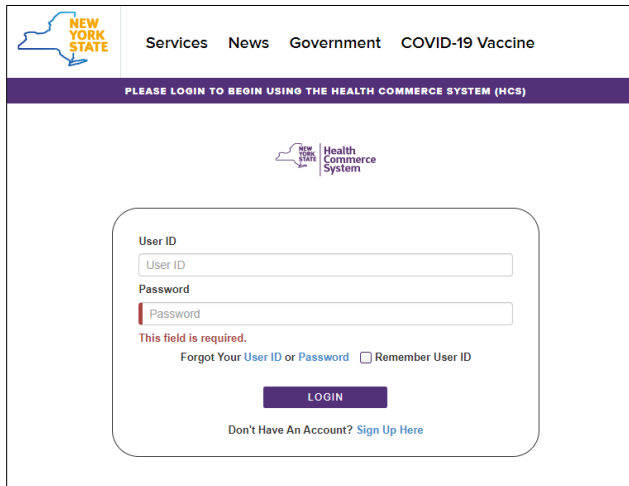


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Newborn Screening Process

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- User ID**: A text input field with "User ID" as a placeholder.
- Password**: A text input field with "Password" as a placeholder.
- Validation**: A red error message "This field is required." is positioned below the password field.
- Links**: A blue link "Forgot Your User ID or Password" and a checkbox "Remember User ID".
- Button**: A purple "LOGIN" button.
- Footer**: A blue link "Don't Have An Account? Sign Up Here".

Note: As of June 1, 2023, accessing the NBS app requires multi-factor authentication.



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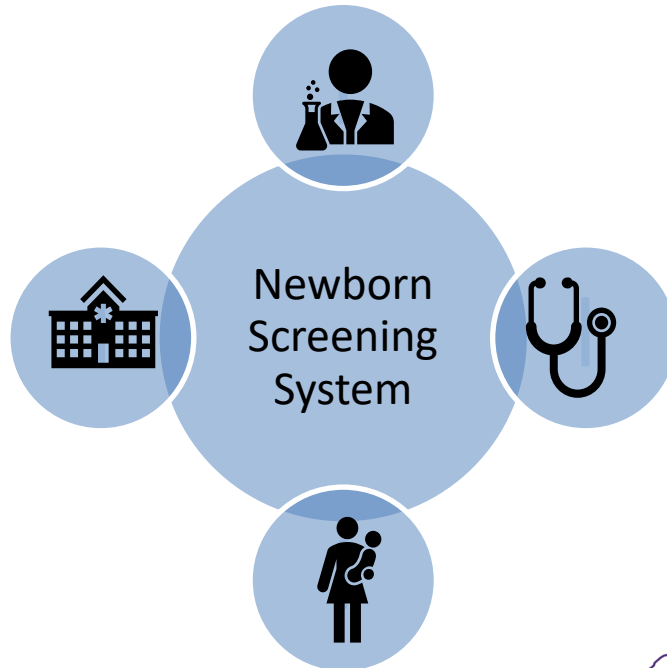
Hospital & Pediatrician Responsibilities Regarding NBS



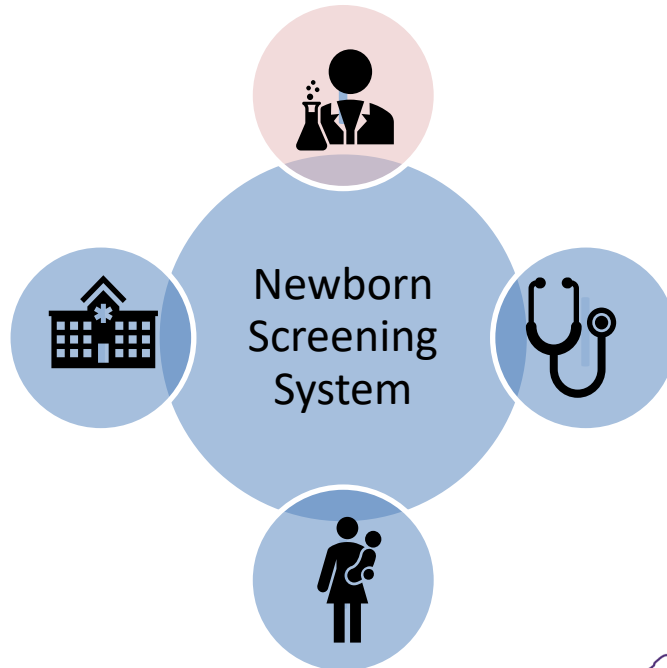
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Newborn Screening System Stakeholders



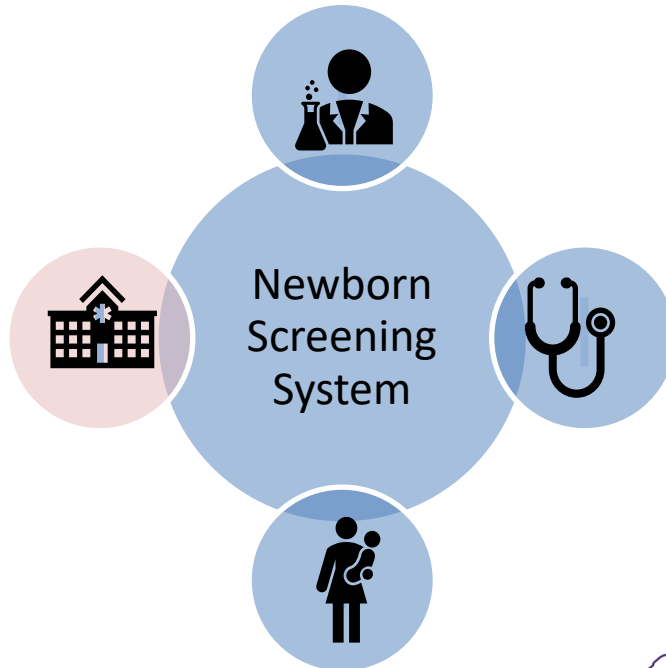
Newborn Screening System Stakeholders



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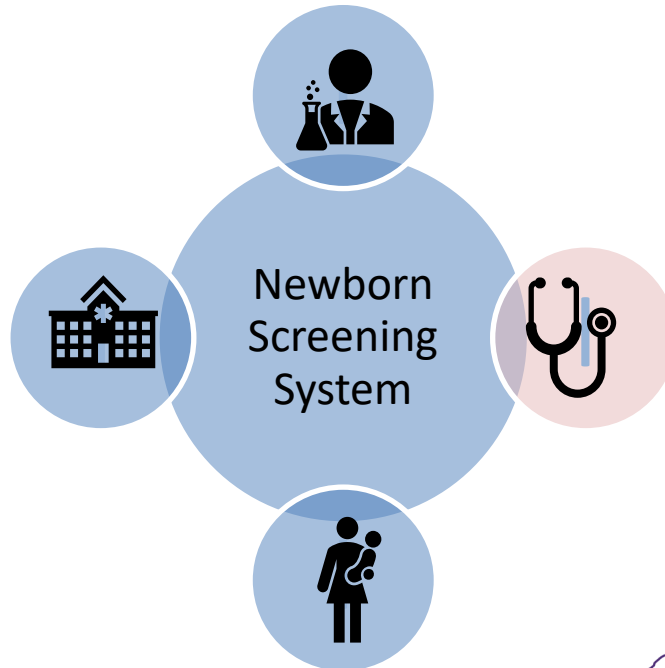
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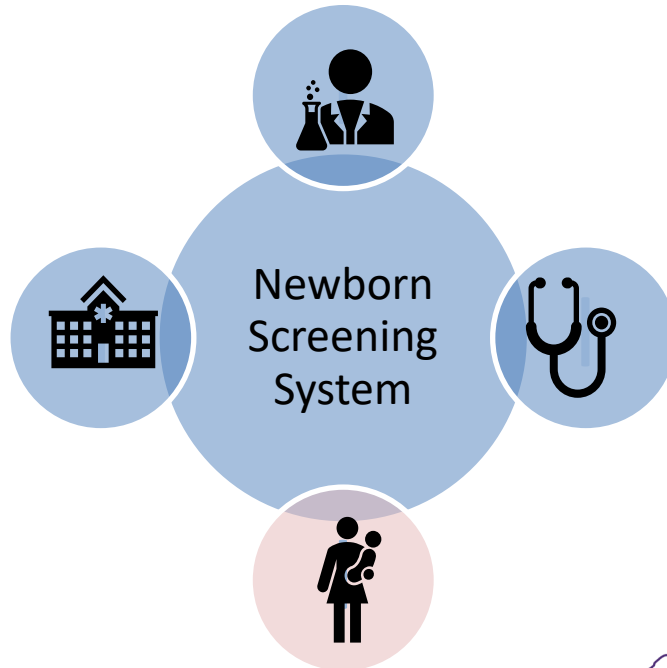
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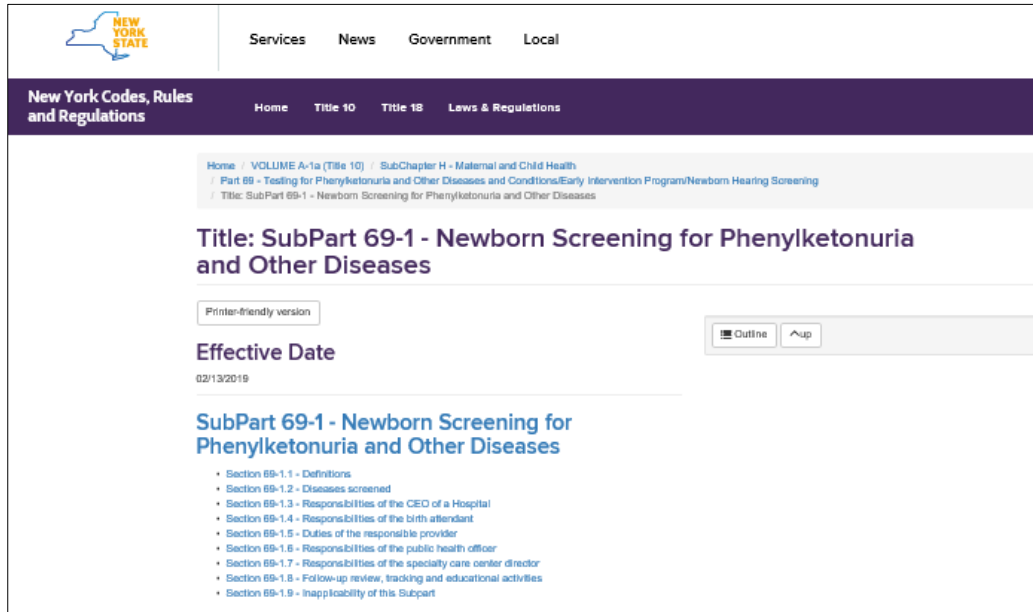
Newborn Screening System Stakeholders



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Regulatory Responsibilities of NBS Stakeholders



The screenshot displays the New York State Department of Health website. At the top left is the New York State logo. A navigation bar includes links for Services, News, Government, and Local. Below this is a dark purple header with the text "New York Codes, Rules and Regulations" and a secondary navigation bar with links for Home, Title 10, Title 18, and Laws & Regulations. The main content area shows a breadcrumb trail: Home > VOLUME A-1a (Title 10) > SubChapter H - Maternal and Child Health > Part 69 - Testing for Phenylketonuria and Other Diseases and Conditions/Early Intervention Program/Newborn Hearing Screening > Title: SubPart 69-1 - Newborn Screening for Phenylketonuria and Other Diseases. The main title is "Title: SubPart 69-1 - Newborn Screening for Phenylketonuria and Other Diseases". There is a "Printer-friendly version" button and a "Outline" button with an upward arrow. The "Effective Date" is listed as 02/13/2019. Below the title is a list of sections: Section 69-1.1 - Definitions, Section 69-1.2 - Diseases screened, Section 69-1.3 - Responsibilities of the CEO of a Hospital, Section 69-1.4 - Responsibilities of the birth attendant, Section 69-1.5 - Duties of the responsible provider, Section 69-1.6 - Responsibilities of the public health officer, Section 69-1.7 - Responsibilities of the specialty care center director, Section 69-1.8 - Follow-up review, tracking and educational activities, and Section 69-1.9 - Inapplicability of this Subpart.

<https://regs.health.ny.gov/volume-1a-title-10/1384929965/subpart-69-1-newborn-screening-phenylketonuria-and-other-diseases>



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Regulatory Responsibilities of Birth Hospitals & Birth Attendants

1. Inform parents of each newborn of the purpose and need for newborn screening and provide educational materials.
2. Specimen collection between 24-36 hours after birth; specimens submitted to NBS Lab within 24 hours of collection.
3. Notify parents and responsible provider when a repeat specimen is required or a result is abnormal.
4. If unable to obtain a repeat specimen, notify the NBS Lab of steps taken to notify parents and responsible provider.
5. Record the lab ID from the collection form, the date and time of sample collection and all screening results in the baby's health record.
6. Establish written policies and procedures re: newborn screening sample collection, storage, shipping, tracking and disposition of results.



Regulatory Responsibilities of Pediatricians

1. Obtain initial screening results within 14 days of birth.
2. When notified a repeat specimen is required, will notify parents and submit repeat specimen to NBS Program.
3. Specimens should be air-dried on flat surface for 3 hours prior to shipment. Specimens should be sent within 24 hours of collection.
4. Record the newborn's Lab ID, the date and time of sample collection, and the screening result in infant's record.
5. Notify parents of the result.
6. Report results of diagnostic evaluations to the NBS Program.



Screening for cCMV



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Congenital Cytomegalovirus (cCMV)

- Most common congenital infection
 - It is estimated that 1 in 200 newborns are born with cCMV
- Of infants with cCMV:
 - 10% are symptomatic at birth.
 - Symptoms include rash, jaundice, microcephaly, low birth weight, hepatosplenomegaly and hearing loss.
 - Of the remaining 90% who are asymptomatic at birth, 10-15% will have long-term health consequences such as sensorineural hearing loss.
- Babies and children with hearing loss are at risk for speech and developmental delays.



Why Screen for cCMV?

- CMV is a very common virus.
 - By the age of 40, more than 50% of adults in the U.S. have been infected with CMV.
- Symptoms are mild so most people do not know they have it.
- Pregnant people may unknowingly pass the virus to their unborn babies.



Should There be Nationwide Screening for cCMV?

- Advisory Committee on Heritable Disorders in Newborns & Children
 - Reports to Secretary of Health and Human Services
 - Recommended Uniform Screening Panel
 - cCMV was nominated and reviewed in 2022, but not approved due to two remaining questions:
 - Can cCMV be detected by dried blood spot newborn screening?
 - Is catching and diagnosing cCMV at birth helpful?
- **Screening in a diverse population like New York will help determine true incidence.**



Statewide Screening for cCMV

- All samples received between late September 2023 and September 2024 will be tested for cCMV (exact start date TBD).
- An extra blood sample is NOT needed for the cCMV screen.
- There is no charge for this extra test (the newborn screen itself is also free).
- CMV screening will be performed using a real-time polymerase chain reaction (PCR) to amplify and detect the genome of the virus in dried blood spots.
 - FDA-approved assay



Opt-out Mechanism

- Brochure to be given to new parents after birth
- **Opt-out options:**
 - QR code on front page can be scanned to go to opt-out portal.
 - Tear-off page:
 - Completed tear-off pages can be sent in with NBS samples.
 - Tear off pages are to be paperclipped to the NBS samples or included in the mailing package.
 - **DO NOT staple the tear-off page to the NBS samples.**
 - Parents can email a picture of completed opt-out page to cmvnbs@health.ny.gov
 - Completed opt-out forms can be mailed to the NBSP.
- Parents can opt out by calling the NBSP: 518-473-7552 **and select option 5 on the phone menu.**
 - Language interpretation available upon request.

Opt-Out Form (tear off)

Please fill out parts A and B below ONLY if you do NOT want your baby's cCMV results recorded. After you complete the form, carefully remove this section from the brochure. Hand this section to your nurse or birthing staff. Ensure the nurse attaches this form to your baby's newborn screen. You may also send this form to the NBS Program (see address on page 4).



Complete Part A and Part B to opt out:

A. Enter information into the boxes:

Newborn screen Lab ID (from pink slip):
Infant's first name:
Infant's last name:
Infant's date of birth:
Birth mother's first name:
Birth mother's last name:
Birth mother's date of birth:
Hospital of birth:

B. Initial, sign and date to confirm you opt out:

I (initial here) _____ am choosing to opt out of receiving the cCMV test result for my infant, even if the result is positive.
Parent name (print):
Parent signature:
Date signed:

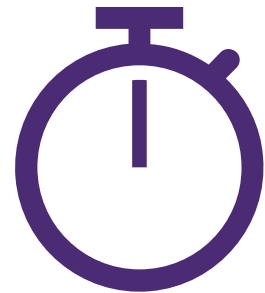


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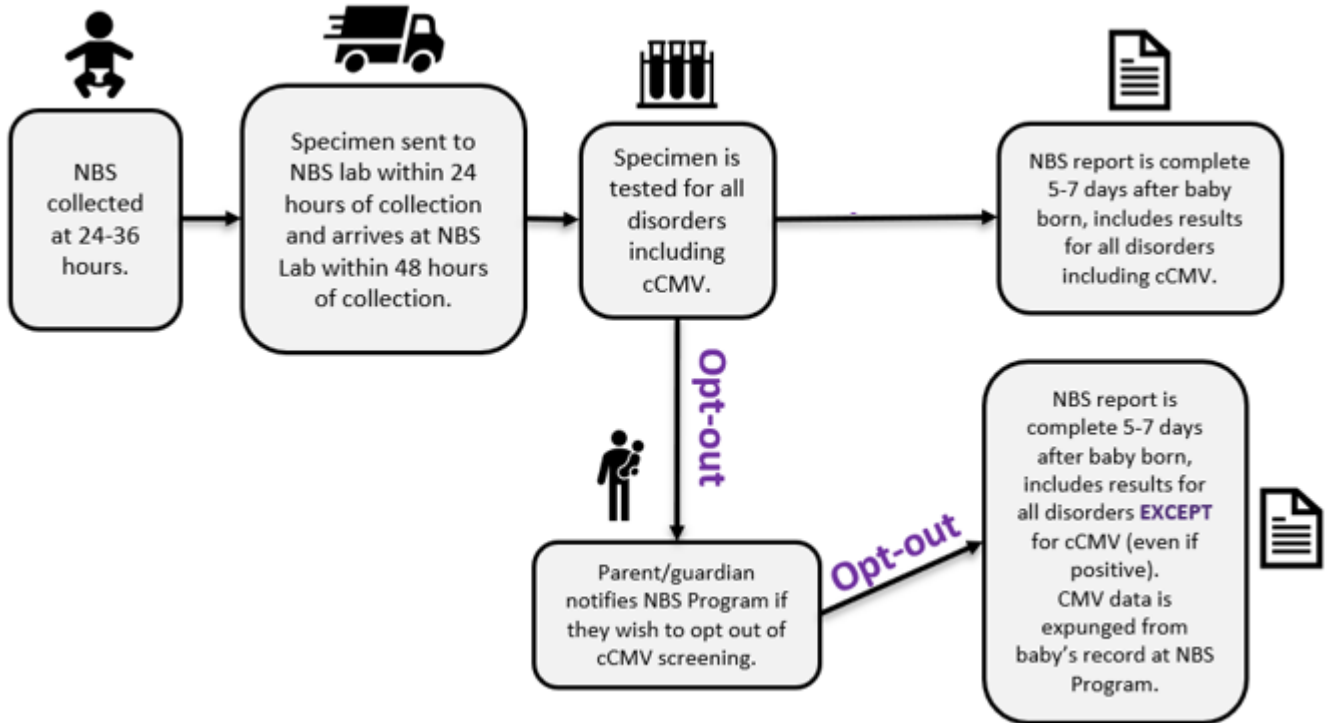
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Opt-out Mechanism

- **Parents must act quickly!**
- Newborn screen reports are available within 5-7 days of baby's birth.
- Once the NBS report is released it is too late for them to opt out of CMV reporting.



cCMV NBS Opt-Out Process/Timeline



Role of Birth Hospitals & Birth Attendants

- Provide all new parents with cCMV educational brochure produced by the NBSP.
- When requested by a parent, send in completed opt-out form with the newborn screen sample.



Possible cCMV Results

NBS Report Type	Description	Action Needed	Necessary Follow-up Actions
Negative	Normal result, patient likely does not have a cCMV infection	None	<ul style="list-style-type: none"> • Primary Care Provider (PCP) communicates results to parent(s) • If baby has any symptoms concerning for CMV, order PCR; report false negatives to the NBSP
Positive	Abnormal result, patient likely has a cCMV infection	Prompt consultation needed with Infectious Disease Specialty Care Center (SCC)	<ul style="list-style-type: none"> • NBSP staff notifies PCP and SCC of the result • PCP or SCC notifies parent(s) of the result • SCC or PCP orders PCR for confirmatory testing as soon as possible • SCC discusses whether treatment is needed with parent(s), notifies PCP • SCC and/or PCP completes diagnosis form and returns it to NBSP

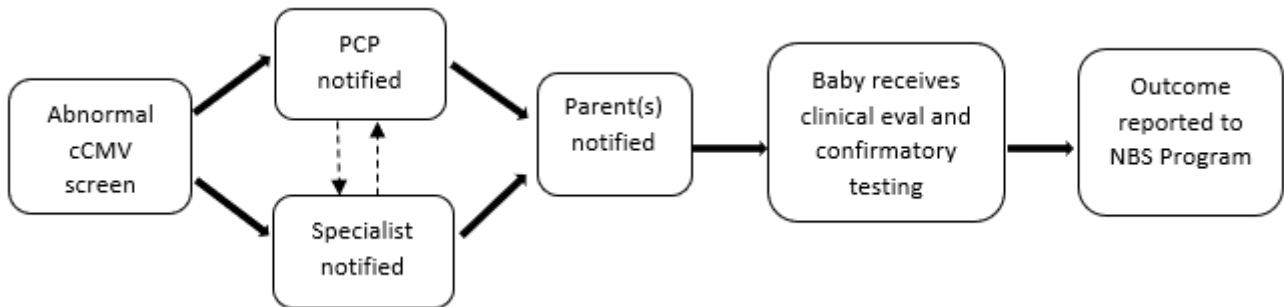


Next Steps Following Positive cCMV Result

- If a baby screens positive for cCMV the NBSP will notify that child's PCP and the closest certified Infectious Disease Specialty Care Center.
- Baby should have confirmatory testing
 - PCR (urine is preferred sample type but saliva is acceptable)
 - Must be completed within 21 days of life to distinguish congenital infection from acquired
- Assess baby for clinical symptoms such as petechiae/purpura, jaundice, microcephaly, low birth weight, hepatosplenomegaly, seizures, hearing loss and retinitis.
- Report PCR result and symptoms to NBSP using provided diagnosis form.



Next Steps Following Positive cCMV Result



Treatment of cCMV Infection

- Symptomatic babies:
 - Antiviral medications such as Ganciclovir or Valganciclovir may improve hearing and developmental outcomes.
 - Treatments last from 6 weeks to 6 months.
 - Administered orally or through IV or PICC line.
 - Ganciclovir can have serious side effects.
- Treatment has only been studied in infants with symptomatic cCMV disease.
- There is limited information on the effectiveness of antiviral medications to treat infants with hearing loss alone.
- **Caution: treatment should only be initiated after consultation with a pediatric Infectious Disease specialist!**



cCMV Registry

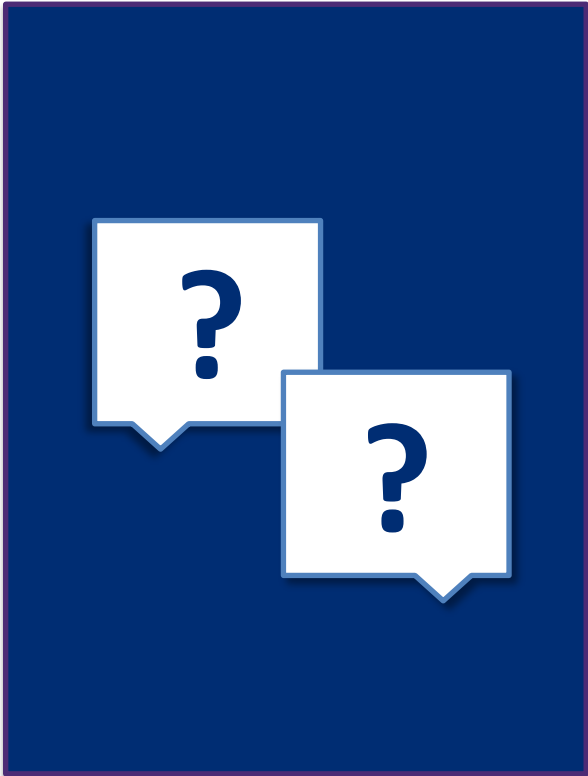
- Babies identified by the NBS may be consented by a pediatric Infectious Disease Specialist to participate in a long-term follow-up registry.
- This will follow outcomes over at least a 2 year period following diagnosis.
- This will help the Infectious Disease community learn about clinical presentation in a diverse, prospectively identified population, best treatment practices, developmental outcomes, etc.



cCMV Health Management

- Infants diagnosed with cCMV should have regular hearing and vision tests.
 - Hearing assessments every 3 months for first 3 years of life.
 - Hearing assessments every 6 months from ages 3 to 6 years.
 - Children identified with hearing loss or vision concerns should be referred to Early Intervention services.





Questions



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Newborn Screening Resources

Website: www.wadsworth.org/newborn

Contact Information:

Phone: 518-473-7552

Fax: 518-474-0405

Email: cmvnbs@health.ny.gov

Address:

David Axelrod Institute
120 New Scotland Avenue
Albany, NY 12208



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