THE IMPORTANCE OF RESIDUAL NEWBORN SCREENING DRIED BLOOD SPOTS

HIGHLIGHTS FROM AN ACMG POSITION STATEMENT

Overview

Newborn screening (NBS) identifies serious conditions in infants before symptoms appear, preventing complications that can lead to permanent disability or death. During screening, a small blood sample is collected and tested. A portion of that sample — called a **residual dried blood spot (RDBS)** — is often stored by state programs and used to improve testing, monitor program quality, and support research.

Why RDBS Matter

Residual dried blood spots:

- Support **test development** and validation for new screening conditions
- Enable **quality control** and improvements to existing tests
- Help **confirm diagnoses** when conditions are identified after the newborn period, understand unexpected infant deaths, or to help diagnose conditions in older siblings not screened for the condition at the time of their birth.

Because NBS is broadly implemented, RDBS can also **provide population health insights** as one of the only specimen sources that reflect the general US population. These samples can be used to study disease incidence, infections, or exposures.

Safeguards and Responsible Use

State newborn screening programs take multiple steps to store RDBS securely and ethically. While practices vary by state, safeguards generally include:

- **Secure storage** with time and temperature protocols
- **Deidentification** of specimens when placed in long-term storage and when used for research or test development
- **Retention policies** aligned with state regulations and policies
- **Retrieval options** for parents, including releasing specimens for clinical use or requesting destruction

Policy Considerations

Legal concerns have emerged over how RDBS are stored and used — especially in research or by law enforcement without consent. While HIPAA protects identifiable health information that may be revealed by RDBS, it does not cover the actual specimens. Further, HIPAA allows certain disclosures to public health authorities and law enforcement. **No federal law governs ownership of RDBS**.

ACMG Position

It is the ACMG's position that:

- NBS RDBS are a valuable national resource that significantly contribute to the health of all children and families.
- RDBS are necessary for test development, quality control, and quality assurance. Federal and state policies should allow for storage, retention, and use of RDBS for these purposes and other key goals of their programs.
- RDBS should be stored with rigorous control and respect for privacy and confidentiality to protect the public.
- Federal and state policies should allow for the appropriate clinical and research uses of deidentified RDBS relevant to child health and include consent requirements for uses in which RDBS will be or could be reidentified.
- States should strengthen policies and laws regarding use of RDBS by law enforcement.
- As states continually revise policies affecting retention of RDBS, careful consideration must be given to the irreplaceable value to child health provided by these specimens.

Did You KNOW?

About 3.6 million newborns are screened annually in the US, improving or saving over 12,000 lives each year.

RDBS repositories are uniquely valuable resources because they represent the full US population.

For more information, see: Rose, N.C., et al (2025). The importance of residual newborn screening dried blood spots, 2025 revision: A position statement of the American College of Medical Genetics and Genomics (ACMG). *Genetics in Med*. 27(8):101433.

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