



American College of Medical Genetics

Medical Genetics: Translating Genes Into Health®

Position Statement on Importance of Residual Newborn Screening Dried Blood Spots

State newborn screening programs are highly valued by the public for their ability to detect newborns that are at high risk for developing diseases with high morbidity and mortality. Newborn screening provides early detection and, thereby, allows for timely treatment with proven clinical interventions that are effective in minimizing disease development. The great majority of the conditions for which newborns are screened are genetic. The dried blood spot card that is collected from the vast majority of the 4.2 million U.S. newborns each year is central to such public health-based newborn screening activities. The American College of Medical Genetics (ACMG) believes that these are invaluable resources for the improvement of newborn screening and, therefore, the health of our children. In addition to their immediate use in screening babies, dried blood spots have considerable additional value. They are essential for quality improvement of newborn screening tests and are critical in the development of new screening tests.

A newborn screening test cannot be introduced into the general population until pilot studies are done in the population. The full spectrum of a specific genetic disease cannot be known until it has been assessed in a general population. This permits determination of the range of severity of the disease, its incidence and genetic etiology in the general population and in subpopulations, as well as the performance characteristics of both the screening and diagnostic tests and the response to interventions.

The only source of material available to carry out such pilot studies and answer many of these questions is the dried blood spot. Many of these questions can be answered by use of either anonymized (no individual identifying link is retained) or deidentified (individual identity link retained and privacy and confidentiality maintained under the stewardship of the public health programs) dried blood spots. When the identity of the individual is needed, as occurs when it is necessary to test a dried blood spot to determine if a disease for which an individual has been diagnosed might be amenable to newborn screening, investigators seek typical informed consent from those involved.

A very small but very vocal minority has begun to argue for the destruction of residual newborn screening dried blood spot filter cards after screening has been completed. Their arguments are based on unsubstantiated and highly exaggerated privacy concerns. Such destruction of dried blood spots would significantly and negatively impact the quality and development of newborn screening programs.

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Accordingly, it is the ACMG's position that:

- Residual newborn screening dried blood filter spots are a valuable national resource that can contribute significantly to the health of our children.
- Newborn screening blood spots are stored with rigorous control and respect for privacy and confidentiality to protect the public.
- If a state decides that newborn screening blood spots should not be retained or used for anything more than the screening test, it is critical that individuals have the option of having their children's dried blood spots deposited in a national repository which will allow for necessary studies under appropriate privacy and confidentiality protections.

Approved by the ACMG Board of Directors on April 29, 2009