
Nearly every baby is screened at birth for more than two dozen serious diseases.

BIOMEDICINE

# *Newborn screening collides with privacy fears*

## Informed consent requirement may change decades-old program

*By* **Jennifer Couzin-Frankel**

The wrinkled heel of nearly every baby in the United States is pricked at birth, and a few drops of blood are dabbed on filter paper and shipped off for analysis. Started in the 1960s, this newborn screening program tests for more than 30 rare and serious diseases that are treatable if caught early in life.

Now, many public health experts who help run or advise the program are worried what the future holds. A new law shaped by a coalition of privacy advocates and conservative politicians requires consent for federally funded research on newborn blood spots, which include DNA but no names. Seeking consent sounds innocuous, even welcome. But experts are concerned that the law, which took effect in March, could hamstring not just fundamental research but also the kind of studies that routinely improve screening.

Efforts to improve newborn testing often require studies on hundreds of thousands of stored blood samples; seeking consent for each one would be prohibitive and impractical. When California researchers sought informed consent to test a cutting-edge screening technology on blood spots from 400,000 newborns, for example, overworked hospital staff did not contact nearly half of eligible families, hampering the study. "Do you want genetic privacy at the expense of everything else?" asks David Orren, chief legal counsel of the Minnesota Department of Health in St. Paul.

When it began lumbering through Congress, the Newborn Screening Saves Lives Reauthorization Act of 2014 was unremarkable; it simply updated an expiring 2007 law that provided federal support for state-run newborn screening programs. In early 2014, the bill passed in the Senate—unanimously, and "in about 30 seconds," says Cynthia Pellegrini of the March of Dimes in Washington, D.C., who advocated for the bill.

The controversy began a few days before the House of Representatives voted on the bill last June, when a nurse named Twila Brase, who runs the Citizens' Council for Health Freedom, a nonprofit in St. Paul that presses for medical privacy, reached out to the office of Michele Bachmann, a tea party icon whose district included the northern suburbs of Minneapolis-St. Paul until she retired from Congress earlier this year. Brase, who also opposes the federal mandate for electronic health records and the Affordable Care Act, had been fighting storage and research on newborn blood spots for years.

Brase's contact had its desired effect: When the bill reached the House floor, Bachmann delivered an emotional speech. "This legislation presumes that every parent of every newborn in the United States of America pre-agrees that the government can have their baby's blood sample, which contains their DNA code," she said. "Americans should not see the death of privacy, especially of the most sensitive private information that every American can have."

Bachmann's speech came too late to affect the House vote. The bill passed. But because legislators had added some minor tweaks to the language before voting, the bill had to return to the Senate, so that the two chambers were passing identical text. That gave time for Bachmann's qualms to catch the attention of members of the conservative Senate Steering Committee, including Rand Paul (R–KY) and Patrick Toomey (R–PA). They sought input from her, as well as from officials from the March of Dimes, the National Institutes of Health (NIH), and other research and advocacy groups. After much discussion, the senators settled on the clause mandating informed consent when newborn blood spots were used in federally funded research. It passed both chambers and was signed into law by President Barack Obama a week before Christmas.

At the crux of scientists' and public health advocates' concerns is what fits under the umbrella of "research," which federal regulations define as investigations that "develop or contribute to generalizable knowledge." Does testing a new screening technology qualify as research? What about studies of a test for a disease not currently on a screening panel, to determine whether it should be added?

"There are public health functions that are mixed up with" what might be considered "pure" research, says Logan Spector, an epidemiologist at the University of Minnesota, Twin Cities. And some research that seems unrelated to newborn screening might not be: Probing leukemia's origins, as research-

ers studying blood spots have done, could also represent nascent steps toward a test for leukemia risk.

Jeffrey Botkin, a pediatrician and bioethicist at the University of Utah in Salt Lake City, who is part of a federal advisory panel on newborn screening, worries about the impact of mandating informed consent. But he's sympathetic to its appeal. "It's good to be the subject of much more public dialogue and scrutiny," Botkin says. Many acknowledge that screening programs could do a far better job of educating parents and doctors, ideally before a baby's birth rather than in the distracted hours afterward.

The Office for Human Research Protections is drafting guidelines on the law and plans to define what qualifies as research. In the meantime, scientists and state health departments are trying to anticipate the law's effects. "We've essentially frozen" our repository, says Michael Watson, the executive director of the American College of Medical Genetics and Genomics in Bethesda, Maryland, which runs a virtual bank of dried blood spots. None of the four participating states plans to provide information from blood spots collected after March, when the law took effect. A pilot study to develop a test for detecting Duchenne muscular dystrophy "has been slowed down tremendously," Watson says.

There's also a big question about whether the law is an early jolt of a larger seismic shift in how deidentified samples are handled. Until now, studying such samples, which carry no names or addresses and are not linked to an individual's health records, hasn't required informed consent. But in January, NIH began expecting grantees on genomic research to seek consent before using deidentified samples. The newborn screening law is turning that recommendation into a national requirement, at least for blood spots. (A handful of states already mandate consent.)

Other samples, like tumor tissue or deidentified blood samples from adults, could be next. The Department of Health and Human Services is rewriting its "Common Rule" governing human subject research. An upcoming draft will reveal whether it wants consent for all deidentified samples. Once those regulations are finalized, perhaps within a couple of years, the newborn screening requirement for consent will be subsumed by the Common Rule.

The Common Rule is Brase's next frontier. She plans to comment on the proposed draft rules when they're released, to urge that all deidentified samples be subject to informed consent before scientists can access them. "When researchers decide we're theirs, that sets people up to oppose what's happening in research," she says. ■

AVIAN INFLUENZA

# Enigmatic bird flu strain races across the U.S. Midwest

## Virologists baffled by the potent strain's continued spread as more than 30 million poultry hit

*By* **Mara Hvistendahl**, *in Minneapolis, Minnesota*

The United States' largest avian flu outbreak in decades is decimating poultry flocks in the heartland and shaking up old certainties about how highly pathogenic avian flu viruses spread. "All the old dogma about high-path influenza transmission has just gone out the window," says Michael Osterholm, director of the Center for Infectious Disease Research and Policy here at the University of Minnesota (UMN), Twin Cities. "We're in totally uncharted territory."

More than 30 million poultry, mostly chickens and turkeys, have been affected, either infected directly by the lethal H5N2 virus or marked to be sacrificed in massive culls. Three states—Iowa, Minnesota, and Wisconsin—have declared a state of emergency, and the federal government has earmarked at least $330 million in emergency funds. So far, the virus appears to pose no threat to people, unlike some other avian flus. But its transmission route is a mystery, and infection control measures have failed to contain it. "This is an unprecedented outbreak in which influenza doesn't seem to follow the rulebook," says Jeff Bender, an epidemiologist at UMN Twin Cities.

The current outbreak, which has affected 147 backyard and commercial flocks since December, is the third arrival of high-pathogenicity H5N2 to the United States: Different strains walloped Virginia and Pennsylvania in 1983, affecting 17 million poultry, and a single flock of 6600 chickens in Texas in 2004. The latest one evolved from an H5N8 virus found in a migratory bird in Russia in September 2014. The same H5N8 strain later cropped up in Germany, Japan, the Netherlands, and the western United States (*Science*, 6 February, p. 616). Along the way, it swapped genes with a North American avian influenza virus to

PHOTO: USDA PHOTO

David Swayne (*left*), director of USDA's Southeast Poultry Research Laboratory in Athens, Georgia, is heading up efforts to develop a vaccine that would potentially stop H5N2's spread in poultry.