

Pressure mounts to expand screening of US newborns

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A US health advisory task force is recommending that all children born in American hospitals be screened for a standard range of 29 genetic diseases — a policy that would put an end to what some call "**newborn** roulette."



Figure. **The US government wants newborns in all states to be screened for 29 medical conditions.** Photo by: Telefocus

Currently, each state sets **newborn** screening guidelines, deciding which metabolic conditions to track in health department laboratories. That has led to huge differences in the number of diseases for which hospitals test — and cries of outrage from parents.

The Advisory Committee on Heritable Disorders and Genetic Diseases in **Newborns** and Children, set up by the US Department of Health and Human Services (HHS), recommended in May that all states screen **newborns** for the same conditions. The task force

consisted of geneticists, laboratory directors, pediatricians, parents, federal agencies and advocate groups.

HHS Secretary Mike Leavitt is expected to make final recommendations later this summer. As with childhood vaccinations, Leavitt's decision will not bind individual states but is expected to influence their choice of screening tests.

For the past decade, organizations of parents whose children have died or suffered because they were born in the "wrong" state have pushed for a uniform screening standard.

"In one state you can live and lead a normal life, and in another state, you can die or be mentally retarded," says Tera Mize of Winston, Georgia.

For example, Arkansas screens for just 4 conditions, while Connecticut mandates screening for 30, including cystic fibrosis.

Mize and her husband, Dallas, started the advocacy group Save Babies Through Screening after their 9-day-old son, Tyler, died in 1998 because of mistakes in the test processing. "It's like **newborn** roulette," Mize says.

Current **newborn** screening identifies conditions in 4000–5000 babies annually in the US. If all states adopted standard screening, the task force estimates at least another 1000 babies with treatable metabolic and endocrine disorders would be detected annually.

Throughout the 40-year history of **newborn** screening, delayed results, false positives and human error have marred the overall success of the program, which is estimated to have spared at least 30 000 children worldwide from mental retardation, other cognitive disorders and death.

The practice of screening **newborns** was started in order to detect phenylketonuria (PKU). Once the leading cause of mental retardation, it is now detected in about 1 of every 12 000 babies.

Over the years, as researchers and physicians came to understand more metabolic conditions, **newborn** screening evolved into one of the most effective prevention tools.

Tandem mass spectrometry, or TMS, can screen for up to 40 conditions at once, using drops of blood collected through pricking a **newborn's** heel. But not every state can afford the TMS machines (US\$300 000) and the staff to run them.

In addition, physicians, researchers and parents are often at odds over the tests because of the lack of clear-cut intervention for some of the diseases and chemical imbalances that expanded screening may identify. Some physicians are concerned that wider screening will increase false positives, leading to unnecessary treatment and anxiety. Advocacy groups, on the contrary, argue that until patients are identified, treatment for the conditions will not be developed.

In the past, WHO's criteria governed which disorders the states chose to screen patients. WHO standards held that established treatment should be available before screening.

The advisory committee's recommendation to test for 29 core diseases is based on treatment protocols. About 20 states currently test for all the conditions on the committee's list.

"The 29 conditions all have a treatment according to the experts that reviewed the material," says Dr. Peter van Dyck, associate administrator of HHS's Maternal and Child Health office. The technology can identify another 25 conditions but there are no efficacious treatments for them.

The incidence of conditions range from 1 in 4000 (hypothyroidism) to 1 in 60 000 (biotinidase deficiency) to 1 in 200 000 (homocystinuria).

Federal agencies already provide technical assistance and funding for regional **newborn** screening centres and the March of Dimes also provides grants to states to purchase equipment.

"[**Newborn** screening] was uniform for years and years. But the explosion of science and technology changes that," says van Dyck. "We do not have the authority to regulate it. But we feel the guidance will be appropriate and it will do what we all want — which is providing the best care for mothers and babies."

Canada is lagging dramatically behind the US in screening of **newborns**. See article on page [23](#).

Footnotes

Health writer Patricia Guthrie's father, the late Dr. Robert Guthrie, developed the **newborn** screening blood-spot filter paper test more than 40 years ago in his Buffalo, NY, laboratory.

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