Technology Drives Advances in Newborn Screening

Advisory committee recommends routine screening of 29 of the 78 conditions that it analyzed.

BY MARK S. LESNEY
Associate Editor

WASHINGTON — Rapid expansion and new technologies are changing the face of newborn screening in the United States, experts and policy makers agreed at the annual meeting of the American Association for the Advancement of Science.

Michael Watson, Ph.D., of the American College of Medical Genetics outlined the rapidity of change. In 2002, around 30% of newborns in the United States were screened for fewer than 5 conditions, and only 5% for 20 or more. By October 2004, closer to 20% of newborns were screened for fewer than 5 conditions, and 27% were being screened for more than 20 conditions.

Only three conditions—phenylketonuria, galactosemia, and congenital hypothyroidism—are screened universally.

Tandem mass spectrometry in particular is driving an increase in the number and kind of diseases tested for, and it is changing the nature of state-run testing facilities, as outlined in a series of presentations at the meeting.

"There are very few things in medicine that are universal. Newborn screening is one of them. Hardly anyone slips through this net. But states vary enormously in what they screen for," said Duane Alexander, M.D., director of the National Institute of Child Health and Human Development.

Because of this disparity, and the conviction that newborn screening has achieved only a fraction of its potential, the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children was established in 2003.

The committee was authorized by Section 1111 of the Children's Health Act of 2000 to provide recommendations for a uniform panel of screening, and decision-making tools for the states to use in evaluating the future development of screening. Part of the committee's charge is to provide advice and recommendations on the funding of grants to the states for the improvement and/or expansion of newborn screening, as outlined in Section 1109 of the act.

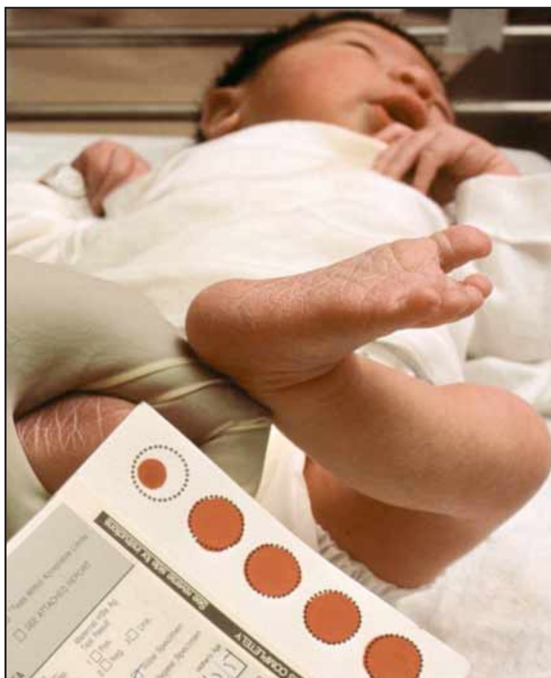
The final report of the American College of Medical Genetics committee on the 78 conditions analyzed by the panel is available for public comment, and the secretary of Health and Human Services will evaluate it. Overall, the committee is recommending routine screening of 29 of the disorders considered, Dr. Watson said

According to Dr. Watson, the committee has already been having an effect on the evolution of newborn screening by its preliminary meetings, and its recommendation for screening 20 metabolic disorders with tandem mass spectrometry (MS).

Currently, more than half the states have instituted mandatory tandem MS screening, with several more having pilot studies or optional testing available, he said.

Tandem MS provides a complex profile of the metabolic status of an infant's blood sample. It is capable of simultaneously analyzing most compounds in a sample, giving both identification as well as concentration information. This can include information for diseases not mandated for screening by a particular program, as well as information on conditions that have no treatments, or ones that may have privacy implications.

"Having already determined that tandem MS ought to be part of a newborn screening program, we now are left with all of those other conditions that fall out of an MS profile," Dr. Watson said. The committee has advocated the release of all clinically relevant data to practitioners and patients, regardless of the purpose of the initial screening.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Ultimately, newborn screening is likely to move into the genomics age, said James Hanson, M.D., of the NICHD. He suggested that, in future, DNA chips, proteomics, nanotechnology, and a variety of biophysical approaches will likely become part of this process. Luckily, for the easy evolution of screening, the standard heel-stick blood spots currently obtained are also appropriate for the majority of the new technologies.

Unfortunately, part of the long-term problem with the evolution—and cost—of newborn screening techniques involves competition from other forms of screening, such as for prostate cancer in adult men, and breast cancer in women. At some level, all these new screening methods inevitably compete for resources, Dr. Hanson said.

"Truth of the matter is, there is not enough money in the health care system the way it's applied for children at the present time to allow us to spend all we would like for every patient or child with a rare disorder," Dr. Hanson said. "It is deplorable from an ethical standpoint but is a practical reality at the present time."

Such cost considerations are likely to increase as screening proliferates. According to Piero Rinaldo, M.D., a pediatric geneticist at the Mayo Clinic, Rochester, Minn., there are also moves to expand screening beyond the newborn stage to capture disorders such as Wilson's disease—a hereditary disorder that causes copper to build up to toxic levels—and congenital disorders of glycosylation, none of which are detectable until later in an infant's life. ■

Public comment can be mailed to Maternal and Child Health Bureau, Health Resources and Services Administration, 5600 Fishers Lane, Parklawn Building 18A-19, Rockville, MD 20857; faxed to 301-443-8604; or e-mailed to screening@hrsa.hhs.gov.

Health Savings Accounts Not the Answer, CalPERS Chief Says

BY JOYCE FRIEDEN
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WASHINGTON — Despite their growing popularity, health savings accounts are not a good solution to the problem of rising health care costs, at least not for California state employees and retirees, Fred Buenrostro said at a health care congress sponsored by The Wall Street Journal and CNBC.

Mr. Buenrostro is chief executive officer at the California Public Employees' Retirement System (CalPERS), the second largest health care purchaser in the country. CalPERS, based in Sacramento, provides health benefits to more than 1.2 million employees, retirees, and family members.

In California, out-of-pocket health care premiums have nearly tripled in 5 years, and Gov. Arnold Schwarzenegger (R) is seeking to cut the amount of premium assistance the state gives to employees and retirees. So "CalPERS, like other employers, is hearing the call of consumer-driven health care," including health savings accounts, Mr. Buenrostro said.

"We are resisting it because we don't want our highway workers, our police officers, our firefighters, our office workers, to switch from our defined benefits health care model to a defined contribution model. We oppose putting our members at risk in such a complex, broken market," he said.

Under a defined benefit plan like those that CalPERS offers, employers agree to pay for a particular level of benefits, no matter what the cost of the plan is. But under a defined contribution plan, the employer pays only a certain amount toward the cost of an insurance policy; any additional costs must be paid by the enrollee.

So CalPERS is trying other ways to cut health care costs. One technique is to avoid doing business with providers that the plan perceives to be too high cost. "Two years ago, we dropped two big HMO partners because their prices went over the top," Mr. Buenrostro said.

The plan is also using generic drugs in 95% of cases, and giving members incentives to buy mail-order drugs. CalPERS has extended the length of its PPO contracts to improve its negotiating position, and is encouraging members to use "centers of excellence" for various procedures.

CalPERS also is talking with other purchasers about price inequities of health care in local markets, and plans to convene a conference of purchasers on this issue later in the year, Mr. Buenrostro said.

A big part of controlling CalPERS' costs has been getting the best price for hospital services. Between 2001 and 2003, hospital prices rose 60%, which was "just unaffordable," he said. CalPERS partnered with California Blue Shield to analyze the costs.

"Blue Shield came up with what was then a shocking discovery: In many cases there was no correlation between price and quality," he continued. "I thought they were kidding." For example, they found that the cost of chemotherapy could range from $135,000 to $300,000.

As a result of the analysis, CalPERS notified 38 hospitals and 17 physician practices that they were in danger of being dropped from CalPERS' provider network unless they dropped their costs and agreed to undergo performance assessments. The proposed change would have saved the plan $36 million in the first year and $50 million for the next few years.

After negotiations with the hospitals and scrutiny from the state insurance department, CalPERS ended up dropping 24 hospitals and several physician practices as of January, forcing 32,000 members to switch their primary care physicians. Although the move resulted in complaints from members as well as the California legislature, Mr. Buenrostro has no regrets.

"It will save tens of millions of dollars for our members and the taxpayers [who pay our salaries], and the decision helped us keep our HMO and PPO premium increases for members under 65 at 9.9% without any takeaways or any increases in copays or deductibles," he said. "We're pretty proud of that."

Despite CalPERS' success, the state of California, like other employers, can't solve the long-range health care cost problem by acting on its own, Mr. Buenrostro said. "We can only solve this problem if we get a national solution." ■