



The California Expanded Newborn Screening Program

What is newborn screening?

The American Academy of Pediatrics defines screening as: “The early detection of conditions for which early and timely interventions can lead to the elimination or reduction of associated mortality, morbidity, and disabilities.” Newborn screening is one of the largest disease prevention programs in the United States, reaching four million babies each year.

California’s current mandatory newborn screening program tests the approximately 530,000 children born each year for four groups of disorders: sickle cell disease and other hemoglobin diseases, phenylketonuria (PKU), congenital hypothyroidism, and galactosemia. PKU can cause irreversible mental retardation if children eat foods high in protein, while galactosemia can cause death from consuming dairy products. Fortunately, with early detection, these disorders can be easily treated with special diets or medication.

In the United States, newborn screening policies and decisions are left up to individual states. This has resulted in a patchwork of standards for newborn screening. Infants across the country do not have equal access to newborn screening and its potential to prevent morbidity and mortality. While other states have improved their efforts of early detection in newborns, California has lagged behind. Forty-two other states test for more diseases than California, and 26 other states have implemented tandem mass spectrometry, a new technology that greatly expands the number of disorders that can be detected at one time, using blood from a single heel prick. This disparity led to the passage of California legislation AB 2427 (Kuehl) in 2000.

AB 2427 (Kuehl): Genetic Diseases and Genetic Screening

This bill mandated the DHS Genetic Disease Branch to conduct a pilot project to study a state

newborn screening (NBS) expansion using tandem mass spectrometry (MS/MS) and appropriated \$3.9 million from the Genetic Disease Testing Fund. The goals of the project were to find out which of the more than 30 disorders detectable by MS/MS have clinical significance, respond to treatment, and therefore warrant inclusion in the mandatory NBS program.

The Tandem Mass Spectrometry (MS/MS) Pilot Project was to be implemented with the help of a grant authorized by the federal Children’s Health Act of 2000 (Kennedy/Frist). Title XXVI of this act requires states to enhance, improve, or expand screening, counseling, or health services to newborns and children having or at risk for heritable disorders. However, no appropriations have been allotted for the Children’s Health Act of 2000. The pilot project lasted 18 months (January 7, 2002 – June 13, 2003) and ended due to state budget constraints.

Maternity Hospital Participation in California MS/MS Pilot Project

Several task forces such as the Institute of Medicine, the Task Force on Genetic Screening, and the American Academy of Pediatrics Newborn Screening Task Force recommend informed consent for population-based newborn screening, particularly for research studies.¹ About half the state’s maternity hospitals decided not to participate in the MS/MS Pilot Project because of restrictions or requirements of individual hospital Institutional Review Boards and concerns about staff time required for the consent process, among other reasons. A study conducted by Faulkner, et al, concluded that written informed consent for newborn screening may not be necessary and may not enable participants to make informed decisions.²

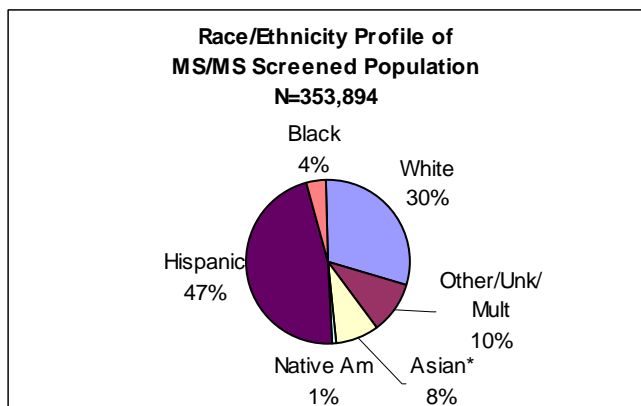
Written informed consent may be a barrier in the newborn screening program that warrants further exploration.

Participants

Families who gave birth at a hospital participating in the pilot project were offered the expanded testing at no additional charge. All parents received a copy of the brochure *Important Information for Parents about the Newborn Screening Test*, the consent form, and the *California Research Participant's Bill of Rights* at the hospital. The *Important Information for Parents about the Newborn Screening Test* brochure is also available in seven other languages besides English and Spanish. Please visit the DHS website: www.dhs.ca.gov/pcfh/gdb/html/NBS/EducationMaterial.htm. Participation was voluntary with signed consent from the parent.

During the pilot project, approximately 354,000 (47%) of the 756,000 California babies who received mandatory newborn screening also received supplemental newborn screening. Only 42,000 (6%) of parents refused supplemental screening and 360,000 (47%) were not offered it. Among families offered the voluntary test, 90% chose to have it.² The main reason for low participation among California babies is that only half of the maternity hospitals offered the supplemental screening to 50% or more of the babies born in their facilities.

The racial/ethnic distribution of participants is as follows³:



*Asian includes: Japanese, Chinese, Filipino, Korean, Vietnamese, Cambodian, Laotian, and Southeast Asian.

Medi-Cal eligible newborns represented 19% of all those screened and 35% of diagnosed and affected newborns.⁴

Cost-Benefits of MS/MS Screening

According to an evaluation of the MS/MS pilot project, for every dollar invested in newborn

screening, the state saves \$2.59 in medical care, special education, developmental services, and physical, speech, or occupational therapy services to children with untreated disorders.⁵ In addition, the annual incremental lifetime costs averted is \$1.7 million per case, and the net treatment costs savings (lifetime costs of not screening minus the lifetime costs with screening) is over \$4.5 million.

Prenatal Care Provider and Pediatrician Surveys – Results of the Pilot Project

The state sent more than 18,000 letters to obstetricians, pediatricians, and maternity hospitals encouraging them to inform expectant parents about the expanded newborn screening program. As part of a three-year evaluation process supported by a Health Resources Services Administration grant, a survey was mailed in June 2003 at the conclusion of the pilot project to 6,197 California prenatal care providers which assessed knowledge of the MS/MS screening pilot study, how often providers discuss screening program information with patients, and barriers to providing screening program information to patients.

700 (11%) prenatal care provider surveys were returned, with about 25% of responses from providers who practice in Los Angeles County.⁶ When asked how they first learned of the MS/MS pilot project, about 1/3 of providers just found out from the survey, about 1/3 learned about it through the *Newborn Screening News*, and about 1/3 learned about it from a letter from the state. Many prenatal care providers did not consider discussing screening with their pregnant patients: 36% said it was the responsibility of the maternity hospital or nursing staff, while 38% said it was the responsibility of the pediatrician. **In addition, nearly 30 percent reported that they never distribute the *Important Information for Parents about the Newborn Screening Test* brochures that they are required by law to hand out to all pregnant women.**⁶

Another survey was mailed between July and August 2003 to 325 pediatricians who had a newborn with an abnormal test result and required follow-up services to either confirm or rule out a diagnosis. A greater percentage of pediatricians were knowledgeable about the MS/MS screening: 48% learned about it through the *Newborn Screening News* and 23% through the state's letter.

These survey results indicate that provider compliance is spotty and that many pregnant

women are not getting information about newborn screening before birth. A significant amount of work needs to be done to ensure access to timely information.

Pilot Project Process

The MS/MS screening and follow-up process took place after the mandatory NBS was completed on the same blood specimen. Thus, the turnaround time for the MS/MS screening results, 28 days, was longer than it was in the mandatory program. Once the MS/MS screening is incorporated into the California NBS program, it is anticipated that the turnaround time will be approximately three days for both processes.⁷

Eight private laboratories contracted with the state to do supplemental testing. Specimen results were sent to the state lab and maintained in a separate database. The state lab and program staff provided follow-up on abnormal results for the MS/MS specimens (e.g., made referrals to centers, contacted physician of record, hospital of birth, etc.). All cases with positive results were reported to one of seven Newborn Screening Area Service Centers (NBS-ASC):

NBS Area Service Center Contact Information

Kaiser Permanente - No. Cal.	(510) 752-6192
Kaiser Permanente - So. Cal.	(626) 564-3322
Stanford Medical Center	(650) 812-0353
Children's Hospital Central Cal.	(559) 353-6416
UCLA	(310) 826-4458
Harbor/UCLA	(310) 222-3751
UC San Diego	(858) 300-1081

Area Service Centers follow up on all positive and inadequate newborn screening tests; provide training, consultation, and technical assistance to NBS providers, primary care providers and public health departments; and provide assistance, referral, and education to families of newborns.⁸ The NBS-ASC Coordinator assists providers in referring families to a California Children's Services (CCS)-Approved metabolic center for confirmatory tests, evaluation, diagnosis, and medical treatment for the newborn.

The Two Zacharys

On February 24, 2004, the March of Dimes held an informational hearing at the State Capitol on the pilot project results. The Wyvill family heard about the meeting and came to testify during the

public comment period. Meanwhile, a March of Dimes lobbyist asked several families whose children's illnesses were detected during the pilot project to testify, including the Black family. The Wyvill and Black families sat side by side in the audience. Both have a son named Zachary, with the exact same metabolic disorder and born only a month apart. The difference is that Zachary Black was born in a hospital that offered the expanded testing and is living a healthy, normal life. Zachary Wyvill was not offered expanded screening under the California expanded newborn screening pilot project and continues to suffer from irreversible brain damage.

On March 11, 2004, the March of Dimes held a legislative summit at the State Capitol to update legislators on newborn screening across the country. At the end of the day, Senator Dede Alpert of San Diego announced that she was immediately introducing legislation that would screen all babies in California for all detectable disorders.

SB 142 (Alpert): Genetic Testing

This legislation, sponsored by the March of Dimes and others, was signed by Governor Schwarzenegger last August 2004. Under this bill, which takes effect August 1, 2005, all newborns will be screened for amino acid, fatty acid oxidation, and organic acid disorders and congenital adrenal hyperplasia in addition to the four categories of disorders California now tests. Until August, families can pay for supplemental screening prior to delivery by contacting one of three out-of-state private laboratories:

Baylor: 1-800-4BAYLOR

www.bhcs.com/MedicalSpecialties/MetabolicDisease

Mayo: 1-800-533-1710

www.mayoreferenceservices.org/mml/mml-sns-intro.asp

Pediatrics: 1-866-463-6436

www.pediatricscreening.com

The laboratory will mail a screening kit and instructions to families. Families must bring this kit with them to the delivery and tell hospital staff that they would like to have an extra blood sample drawn at the time of their baby's mandatory newborn screening. The labs will send the results to the physician of record in about two to four days.

Cost Issues

Last year, the cost of California's mandatory newborn screening program was \$60, covered by Medi-Cal, health plans, and most private health insurance. However, effective January 1, 2005, this fee was increased to \$78 through emergency rulemaking to finance the newborn screening expansion. The \$18 increase covers additional lab equipment, reagents, and personnel in preparation for the August 2005 expansion and is also covered by insurance programs.

Medi-Cal and other health plans will cover the expanded newborn screening when it is incorporated into the existing NBS program. Until then, families will have to pay out-of-pocket to cover the expense of supplemental testing through private labs. Testing fees range from \$25-\$89.

Medi-Cal is funded by 50% matching federal funds and 50% General Funds. The state share for tandem mass screening would be \$20 per baby, or \$2 million. Medi-Cal will save four times as much over the course of the lifetime of what we detect in 30 years.⁹ Therefore, there will be no net cost to the state.

The Future: HHS Recommends Uniform Panel of Newborn Screening Tests for All States

In October 2004, a federal advisory committee recommended that states screen for at least 30 genetic disorders for which there is available treatment. According to Peter Van Dyck, director of the Maternal and Child Health Bureau within the HHS Health Resources and Services Administration (HRSA), a major push for the guidelines is the wide disparity in the number of conditions now included in state-mandated newborn screening programs. The federal advisory committee seeks to promote standardization in newborn screening across the country by encouraging newborn screening programs to become aligned and uniform in their screening.¹⁰

The California newborn screening program is an essential public health program that prevents catastrophic health consequences through early detection, diagnosis, and treatment. California births represent one in eight of all children born in the United States; therefore, the expansion has the potential to reach a vast majority of newborns. All children deserve the same basic standard of care, no matter what state they are born in.

Maternal and Child Health Access asks that you promote expanded newborn screening within your clinic, your family, and your neighborhood and help women who can't pay for the test until Medi-Cal or insurance does so later this summer.

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