2006 NASCIO

Recognition Award Submission

State of California

Office of the State CIO

Cross-Boundary Collaboration and Partnerships

Executive Summary

The Screening Information System (SIS) implemented by the California Department of Health Services (CDHS) is the critical cornerstone of California's prenatal and newborn genetic disease screening program, one of the largest and most comprehensive genetic screening programs in the world.

CDHS implemented SIS in July 2005 with two major goals in mind: to enhance their existing outdated information technology system and to expand the number of rare genetic diseases they screen for. Today, SIS enables physicians to diagnose and treat a wider range of genetic disorders than previously possible. Using the system, newborns throughout the state are now screened for 75 inherited and congenital disorders rather than the previous 39. Undetected, these rare disorders can cause devastating disabilities. But if caught quickly, they are often treatable. Changes as simple as altering an infant's diet can mean the difference between a normal life versus mental retardation or even early death.

SIS also allows the state to better manage test results and reporting and to achieve more efficient communications and collaboration between the multiple public and private entities involved in genetic screening, diagnosis and treatment. Once identified as having a genetic disease, SIS helps facilitate extraordinary follow-up for affected babies and their families until the disorder is fully diagnosed and treatment is initiated. This process involves an extensive amount of cross-boundary collaboration between labs, case coordinators, counselors, physicians and staff of the CDHS Genetic Disease Branch.

Ultimately, SIS allows CDHS to intervene earlier with more effective treatment of children with a wider range of genetic disorders, thereby radically increasing the chances a baby born with a genetic abnormality in California can live a healthy life.

A. Concise description of the business problem and solution, including length of time in operation

In 2000, the California Legislature authorized a massive enhancement of the state's prenatal and newborn genetic testing information system. The existing 20-year-old legacy system was operating well past its expected lifetime and had virtually no capacity to incorporate new screening programs or meet enhanced screening requirements. The California Department of Health Services (CDHS), which manages the prenatal and newborn genetic screening program for the state, was unable to take advantage of newer, lower-cost computing resources because the current system precluded the introduction of innovative tools, methodology, and system software. End-users faced unacceptable system response times and the list of required system enhancements continued to grow.

At the same time, medical technology was advancing rapidly. The existing system would soon have been unable to interface with new medical technology, which had become increasingly prevalent among screening partners. In addition, CDHS faced the risk that it would not be able to respond quickly to a system failure due to the proprietary nature of the existing system. This risk was life threatening for newborns with genetic diseases that might go undetected if the legacy system went down.

In 2004, before replacement of the information technology system could be accomplished, the California Legislature also mandated an expansion of newborn genetic testing. The existing system and technology could only screen for 39 genetic diseases. The Legislature wanted to expand that number dramatically. It was then up to CDHS to determine how to design and build both expansions under short timeframes, as well as accomplish the difficult task of collecting and managing a much larger amount of data.

Working with Deloitte Consulting, CDHS leaders set out to develop an innovative solution. In July 2005, SIS was launched. SIS is a web-based information system that allows CDHS to better manage its prenatal and newborn genetic screening program. SIS enables physicians to diagnose and treat a wider range of genetic disorders in children than previously possible; allows the state to better manage test results and reporting; and enables more efficient communications between the multiple entities involved in genetic screening, diagnosis and treatment.

B. Significance to the improvement of the operation of government

SIS provides enhanced computer support for both prenatal and newborn screening programs. The prenatal screening program screens for genetic and congenital disorders (e.g., Down syndrome, anencephaly, spina bifida, and neural tube defects). The newborn screening program provides screening of newborns for other genetic and congenital disorders (e.g., phenylketonuria and other metabolic disorders, galactosemia, primary congenital hypothyroidism, sickle cell anemia, and other hemoglobin diseases, and congenital adrenal hyperplasia). As a result of both programs, babies are spared lifelong disabilities (including severe retardation) and even premature death.

SIS facilitates collaboration among California laboratories, case coordinators, counselors, physicians and staff of the CDHS Genetic Disease Branch. SIS was designed to transform congenital disease screening and treatment business processes by providing solutions to several problems the legacy system was not capable of solving. Online access by multiple end users was required along with a system capable of processing California's enormous volumes of data. The system had to allow ease of expansion for new types of testing and new disorders and a myriad of complex reporting requirements had to be supported. The system had to provide enhanced security and privacy protections and be HIPAA compliant. SIS has addressed all these issues through a highly modular, scalable and expandable system that includes a data warehouse that supports over 150 operational and analytic reports and ad-hoc reporting. In addition, SIS incorporates the latest generation of specialized congenital screening equipment, called Tandem Mass Spectrometers. This technology has allowed a dramatic expansion in the number of genetic disorders that can be screened for.

The data in SIS also serves as a vital source to support state-required registries of genetic abnormalities, and provides data to meet myriad state and federal reporting requirements, thus allowing state and federal leaders to target public health intervention and education more appropriately.

Technology is a critical component of SIS, allowing for an improved user interface, direct access to on-line reporting capabilities, easier access to necessary information by appropriate medical providers, and improved security and privacy protection. Cutting-edge technology also made it possible to add a matching capability to SIS, which means connections can now be made between prenatal and newborn cases, and all family members' tests can be linked to each patient. This was impossible with the legacy system because it is a highly complicated function due to the nature of medical data, its sources, and formats. Such matching allows creation of a complete genetic profile of the family's risk, thereby increasing the quality and comprehensiveness of the services the state provides to its citizens.

Not only does SIS screen for a battery of prenatal and newborn genetic disorders, but the system also provides extraordinary support to the families in the form of extensive, individual follow-up, counseling, and definitive diagnostic testing. Each

baby identified with a positive screen is carefully followed until necessary treatment is initiated. No other state has a system that combines prenatal and newborn genetic screening with case-specific, individualized follow-up and counseling to ensure that each newborn baby who screens positive for a genetic disorder receives definitive diagnostic tests and immediate therapeutic services.

SIS has significantly improved data sharing between all end-users (CDHS staff, laboratories, case coordinators, and follow up specialists). It has allowed real-time, online access to follow-up specialists who, in the past, had received delayed information via faxes and mail. To do this in any state is an accomplishment. To do it in a state as large and complex as California is truly a feat.

C. Benefits realized by service recipients, taxpayers, agency or state

SIS allows health care workers to more readily address the needs of babies born with rare but often treatable diseases. During the pilot phase, an infant girl was diagnosed with a metabolic disorder that interferes with digestion of a particular amino acid. At the time, only 32 cases of the disease had been documented worldwide. Had the girl not been born and tested during the pilot project, she probably would not be alive today.

SIS provides significant benefits to not only the 400,000 pregnant women who are tested each year and the 550,000 newborns tested annually, but also to other important stakeholders, including the State of California, the citizens of California, the genetic disease screening communities, and health care professionals.

In the first year of expanded testing, 105 newborns were diagnosed with rare genetic disorders that otherwise would not have been diagnosed in the first days of life and 20 infants were identified as having Congenital Adrenal Hyperplasia. With early intervention and treatment (sometimes involving nothing more than a specialized diet), these babies will be spared severe disabilities and even premature death. Instead, they will be able to live healthy lives.

As David Swift put it when he spoke about the call he received from his doctor telling him that his daughter, Giana, tested positive for a genetic disorder: "I gasped. I thought at the time it was the worst phone call I'd ever received in my life. I think in hindsight it was the best phone call I've ever received in my life, because...that was the gift of life.... Had that test not caught this, had the screen not provided for the opportunity to know she had this disorder, chances of her dying were probably better than 80%. Had she not died, she probably would have been severely ...ill or ...retarded. And today we have a precocious, happy, healthy, beautiful little girl."

Every California mother benefits from SIS in that it can, in most cases, eliminate worry that an unborn child has a genetic abnormality. In the cases where test results are positive, educational efforts can be undertaken so the parents understand the disease and are prepared, along with their doctors, to manage it.

The child, the family, the community and the taxpayers of California all benefit through significant cost savings, future cost avoidance, and improved service delivery. Cost savings will be realized through increased efficiency, and reduction or elimination of redundant processes. Future cost avoidances will be achieved through early detection of genetic abnormalities in newborns whose disorders would have gone undetected in the past, resulting in extraordinary lifetime medical costs. Without detection and early treatment, each child born with these disorders would suffer mental retardation, other handicapping conditions or premature death. To illustrate the cost avoidance, considering mental retardation alone, the lifetime costs of care of a single affected newborn have been documented at $300,000 to $1 million. Of course, the emotional cost for the family is incalculable.

D. Realized return on investment, short term/long term payback (include summary calculations)

Annually the expansion will detect on average an additional 125 cases of metabolic and endocrine disorders that, if undetected and untreated, would lead to mental retardation, other handicapping conditions or death. Roughly, 105 of these cases are metabolic disorders and approximately 20 are cases of congenital adrenal hyperplasia (CAH). It is conservatively estimated that at least 60 of these cases (50 metabolic and 10 CAH) have disorders where interventions could be life saving and prevent serious handicaps. The costs of interventions for these 60 cases have been estimated to average $100,000 over a lifetime and $6,000,000 in program treatment costs. However, state metabolic specialists estimate that it is more likely that as many as 92 cases may be detected that would involve significant interventions.

Without the expansion's detection and intervention, 20 would result in severe mental retardation, 20 in mild to moderate retardation, and 20 in death.

The lifetime costs of care of individuals who are mentally retarded have been documented and average $1,000,000 for severely mentally retarded and $300,000 for mild to moderately mentally retarded per newborn. The net average in averted costs would be $26 million (20 x $1,000,000 = $20,000,000; 20 x $300,000 = $6,000,000).

In addition, 20 deaths can be prevented each year. If the economic value of $5,000,000 is assigned to each life saved, an additional $100 million in benefits will be obtained.

The net benefit of this expanded testing statewide is approximately $126 million. The net incremental benefit of $126,000,000 achieved divided by the annual screening cost of $9,079,000 gives an overall cost benefit ratio of $13.88 saved for each dollar spent on expanded screening, or $8.36 if the treatment costs are included. However, if the higher estimate of 92 rather than 60 preventable cases is used, these values would be significantly higher.

COSTS

Annual Screening costs:	$9,079,000
Treatment costs	$6,000,000
Total costs	$15,079,000

BENEFITS	20 lives saved
Lives saved	$100,000,000
Treatment costs averted	$26,000,000
Total	$126,000,000