

THE COMPUTERWORLD HONORS PROGRAM

CASE STUDY

LOCATION:
*Sacramento, California,
United States*

YEAR:
2006

STATUS:
Laureate

CATEGORY:
*Government and
Non-Profit Organizations*

NOMINATING COMPANY:
Deloitte

ORGANIZATION:

California Department of Health Services

PROJECT NAME:

Genetic Disease Screening Information System

Summary

The California Department of Health Services' Screening Information System – A Miracle Life-Saving System

“Austin is, as you can see, very healthy... right in line with his development... We're thankful for the newborn screening that he had. We believe that it saved his life. We all sleep better now knowing that he sleeps well and that he will wake up in the morning.” - Linda Fernandez, speaking about her grandson, Austin, at a July 26, 2005 press conference announcing the kick-off of California's expanded newborn screening system.

The Screening Information System (SIS) is the critical cornerstone of California's comprehensive prenatal and newborn genetic screening program, which identifies rare but treatable genetic disorders in babies before and immediately after birth. SIS enables physicians to diagnose and treat a wider range of genetic disorders in children at an earlier age than previously possible.

The California Department of Health Services (CDHS) implemented SIS in July 2005. SIS allows the state to better manage test results and reporting and to achieve more efficient communications between the entities involved in genetic screening, diagnosis and treatment. Using the system, newborns throughout the state are now screened for 75 inherited and congenital disorders. Once identified, those babies and their families receive extraordinary follow-up until the disorder is fully diagnosed and treatment is initiated. Undetected, these rare disorders can cause devastating disabilities. But if caught quickly, these disorders are treatable. Changes as simple as altering an infant's diet can mean the difference between a normal life or mental retardation or even early death. 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 can live a healthy life.

Introductory Overview

Prenatal screening is a voluntary program offered to all women in California who obtain prenatal care prior to 20 weeks of pregnancy. Approximately 400,000 families are served each year. Newborn screening is a mandatory program and serves approximately 550,000 newborns



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annually.

In 2000, the California Legislature authorized a massive enhancement of the existing rudimentary genetic testing information system. Before that could be accomplished, in 2004, the California Legislature mandated an expansion of newborn genetic testing. 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. CDHS leaders immediately set out on a long and complex journey to develop an innovative solution.

Today, SIS provides enhanced computer support for both 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.

Using SIS, the number of disorders that are detected and treated in California's newborns has increased from 39 to 75. In addition, the system was designed so that tests for additional disorders can be added to it quickly and easily.

Benefits

"I think it's a miracle that parents can find out this information ahead of time..." - Sue Warner, speaking about her infant son, Bradley, at a July 26, 2005 press conference announcing the kick-off of the state's expanded newborn screening system.

There is no doubt SIS is helping those it was designed to help. Using SIS, health care workers more effectively track and more readily address the needs of babies born with rare but often treatable diseases. During the pilot phase of SIS, an infant girl was diagnosed with a metabolic disorder that interferes with the proper digestion of a certain amino acid, which means she must have a protein-free diet. At the time, only 32 cases of this disease had been documented in the world. Had this baby girl not been born and tested during the pilot project, she probably would not be alive today.

Through the pilot phase of SIS, 24 newborns were diagnosed with rare genetic disorders that otherwise would not have been diagnosed in the first days of life and 12 infants were identified as having Congenital Adrenal Hyperplasia. With statewide implementation of this new system in July 2005, an additional 80 to 100 children with these disorders will be identified each year.

California laboratories, case coordinators, counselors and the staff of the CDHS Genetic Disease Branch all use SIS. Prior to SIS, they utilized a 20-year-old legacy system that was operating well past its expected lifetime. The existing system had virtually no capacity to incorporate new screening programs or meet enhanced screening requirements. CDHS 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



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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.

SIS, on the other hand, uses cutting-edge technologies and is a highly modular, scalable, and expandable application. Because it is Internet-based, it is easily accessible from any computer with an Internet connection. In addition, the integration of functionality and the pre-population of data in several screens help eliminate redundant data entry and streamline business processes.

SIS also incorporates Optical Character Recognition (OCR) technology, providing CDHS clients and staff significant time and efficiency savings. Original test results are stored in the system in portable document format (PDF) form, making them easily accessible by doctors, lab technicians or whoever else may need to verify data, a common process that previously involved numerous phone calls and manual location of paper forms.

A data warehouse was built along with SIS and is populated on a daily basis to support over 150 operational and analytical reports. In addition, an industry strength business intelligence and analytical reporting tool allows ad-hoc analysis and creation of myriad reports, allowing CDHS to meet state and federal reporting requirements on an ongoing basis.

SIS was designed to be easily adaptable to support an increasing number of disorders and a growing population. In addition, it was purposely built in a generic fashion so other states, if so inclined, could use it as a model for building their own system. Already, California has had inquires from numerous states and even other countries, including China, Australia and the Philippines, that are interested in finding out more about the system and how they might develop something similar.

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

The Importance of Technology

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.

Technology also allowed the project to be extended to include tandem mass spectrometry testing and testing for Congenital Adrenal Hyperplasia. This expansion added screening for many more metabolic disorders and could not have been accomplished using the legacy system.

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 family members' tests can be linked to the 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.

Overall, SIS is demonstrating how an investment in technology, when done right, can make an enormous difference in the lives of children affected by rare genetic disorders.



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Originality

California's genetic disease screening program is one of the largest and most comprehensive in the world. 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.

SIS is also advanced in its ability to protect sensitive medical information. It was designed and built to be fully compliant with the requirements of the Health Insurance Portability and Accountability Act of 1996 (HIPAA). Its multi-tiered security structure provides the highest level of application and data security.

Success

The Screening Information System is fully operational and has exceeded its goals. It 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.

Through the pilot phase of SIS, 24 newborns were diagnosed with rare genetic disorders that otherwise would not have been diagnosed in the first days of life and 12 infants were identified as having Congenital Adrenal Hyperplasia. As a result of this new system an additional 80 to 100 children with such disorders will be caught through the expanded screening each year. 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.



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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.

Most users across the state, including CDHS staff, laboratories, case coordinators, and follow up specialists, have adopted SIS quickly and easily.

Difficulty

The SIS project was almost defeated on a number of occasions for a variety of reasons. Despite the problems that might have undermined its success, through the project team's perseverance, creativity, dedication, and old fashioned hard work, it succeeded spectacularly.

California information technology procurements are best described as an obstacle course where a wide variety of stakeholders can delay or reject a contract award at any time during the procurement process. The SIS procurement began in 1999 but a contract was not awarded until 2003. In the interim, the contract award was delayed by a change in procurement rules just as the contract was about to be awarded. Then there was a bid protest which took significant time to resolve.

Next, the State Legislative Analyst's Office requested that an independent legal review by a private legal firm be undertaken of the standard contract terms and conditions in the proposed SIS contract. It took six months to procure the independent legal firm and have it complete its analysis of the contract terms. Finally, after the contract was awarded to Deloitte Consulting, there had to be a renegotiation of certain contract terms based on the independent legal review. Still other provisions of the contract had to be renegotiated because the technology existing when the procurement RFP was written had changed substantially over time. In addition, by the time Deloitte Consulting was ready to begin work, the key staff which had been proposed for the contract had already been assigned to other projects because of the procurement delays, and a new group of key staff people had to be put together quickly.

Next, the California Legislature mandated expanded screening within a timeframe viewed by most as unattainable. In addition, coordinating among a large number of diverse stakeholders within CDHS, including Genetic Disease Program staff; Information Technology Services Division staff, Primary Care and Family Health program staff, as well as several contracted external oversight management teams, made the task extremely complicated.

Staff reductions added to the complexity on the project. After SIS was first proposed, the Genetic Disease Branch lost 37 positions as a result of statewide budget cuts. The Genetic Disease Branch proceeded with a severe resource shortage that put an extreme burden on the remaining staff. The remaining staff was required to, in effect, work two jobs for long stretches of time.

In addition, the two key senior Information Services Technology Division staff associated with



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this project retired and their replacements joined the project midstream. Information Services Technology Division support was crucial for the success of this project in a number of ways and the loss of this staff was initially a setback for the project. Similarly, the Assistant Deputy Director of the Primary Care and Family Health program, who had taken an active role in overseeing the project, was transferred to a new job and her replacement joined the project in midstream.

Moreover, CDHS was forced to fire the external project manager in the middle of the project. The project oversight committee saw this as an enormous risk. They insisted that the learning curve was too steep for someone new and predicted that CDHS had just introduced failure into the project. But based on the situation at hand, CDHS leadership saw it as the only way to get them where they needed to go by the deadline. CDHS hired a new project manager who was able to learn quickly. Complicating the situation further, the remaining staff was loyal to the previous project manager. It took some time for the new project manager to win them over.

Perhaps most challenging was the fact that CDHS was approximately half way into the design phase of the project when the legislature introduced a second piece of legislation which added the requirement to include tandem mass spectrometry testing for new congenital disorders. Suddenly, CDHS had to switch gears. Much of the work they had already done was now obsolete. This forced CDHS to make a critical decision: Do they design another system as a parallel effort or put all their eggs in one basket and integrate the two systems into one innovative system that would essentially require them to start all over again? CDHS decided to go for broke and design an innovative, integrated system, despite warnings from the State Legislative Analyst's Office and the Department of Finance that the approach was fraught with risk.

Ultimately, it was the right decision. SIS was delivered slightly ahead of schedule and has proven to be an innovative and effective system for managing expanded genetic testing in California. Some of the keys to achieving this included getting leadership involved on a daily basis, introducing a structured process, dealing with issues before they got out of control, coupling IT and program areas tightly, and partnering with an effective and professional application development contractor (Deloitte Consulting).

Today, SIS is allowing a higher number of California infants to be effectively screened for a wider range of genetic abnormalities, giving affected children and their families a greatly improved quality of life. Again, to quote David Swift: "Had Giana not been born during that pilot program...and had we not had such great follow-up from the State and different agencies...she probably wouldn't be alive here today. That's our miracle."