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The Utilization of Genetic Counselors Within A Newborn Screening Program

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Genetic counselors have participated in the Michigan Newborn Screening Program on a contractual basis since 1988. Their role includes newborn screening education and training, newborn nursery site visits, and monitoring newborn screening in hospitals. Their impact has been to improve the quality of newborn screening services by reducing errors and increasing completion of data fields on newborn screening cards, improving hospital nursery cooperation and problem solving, and enhancing health department response to specific problems.

KEY WORDS: newborn screening coordination; role of genetic counselors in newborn screening; regional hospital training; new role for genetic counselors.

INTRODUCTION

On July 1, 1987, the Michigan Department of Public Health assumed responsibility for implementing an expanded newborn screening program in response to a bill enacted by the State legislature. Public Act 14 mandated testing all infants born in the state for six disorders: phenylketonuria (PKU), hypothyroidism, galactosemia, biotinidase deficiency, maple syrup urine disease (MSUD), and sickle cell anemia. The law required that all filter paper specimens be submitted to a centralized laboratory at the Department of Public Health. It also authorized collection of a fee for each newborn tested. Originally set at $18, the fee was raised to $20 in 1989. Revenue from the fee covers the cost of laboratory testing, administration, follow-up, and medical management of affected children. These requirements

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were in contrast to Michigan's previous newborn screening program, started in 1965 for PKU, which permitted decentralized testing through individual hospital laboratories, charged no fee for specimens sent to the state laboratory, and did not assure comprehensive follow-up. We have identified a novel strategy to preserve state-funded genetic counseling positions while using genetic counselors to enhance and improve the newborn screening program.

HISTORY OF THE NEWBORN SCREENING PROGRAM

As the eighth most populous state, Michigan welcomes about 150,000 new babies each year. Under the law, the health professional in charge of the care of a newborn infant must assure that a specimen is obtained and that the screening tests are performed. The Department of Public Health oversees this process and reports all positive or unsatisfactory test results back to the proper health care provider. More than 99% of all births occur in approximately 140 maternity hospitals throughout the State. Thus, for the majority of babies, hospital nursery and laboratory staff play a key role in obtaining the heelstick specimen before discharge and sending the newborn screening filter paper card to the laboratory. (Efforts have also been made to identify and educate midwives attending home births about the newborn screening process.) Prior to 1987, however, no instruction about the newborn screening process had ever been provided to hospitals. For 22 years, the program was exclusively laboratory-based, with only limited efforts to assure comprehensive testing, follow-up, and appropriate medical management.

FOLLOW-UP METHODS

Currently, the laboratory receives about 600 specimens each weekday. A database has been established to keep track of pertinent information on each test card received. Negative results are reported to the referring hospital in writing, while positive test results are reported to the primary health care provider by telephone and/or letter, depending on the urgency of the situation. Infants with presumptive positive screening results are referred to a medical management system for diagnostic confirmation and specialized medical follow-up. Children's Special Health Care Services has contractual agreements with three centers to locate babies in need of follow-up and provide comprehensive care for diagnosed children. These include: the Pediatric Neurology Metabolic Disease clinic at the University of Michigan in Ann Arbor, the Sickle Cell Detection and Information Program in Detroit with nine regional sites, and the Neonatal Thyroid

Screening, Coordinating, and Follow-up Program at the University of Michigan which coordinates treatment through 11 pediatric endocrinology practices statewide.

PROGRAM ADMINISTRATION

Shortly before expansion of the newborn screening program, the Bureau of Community Services had revised its administration of the Department of Public Health Genetics Program. This reorganization eliminated state funding for four clinic-based genetic counselors. At the end of 1987, there appeared to be little possibility of the genetic centers supporting those pediatric genetic counseling positions. Even if hospital-based funding could have been found, public health initiatives in genetics such as education and outreach, and the statewide network for referral and reimbursement, would have been eliminated.

Meanwhile, the need to provide hospitals with regional coordination and education about the state-mandated newborn screening process had become apparent during the first few months of operating the expanded program. Public health administrators concerned about the future of the Genetics Program as well as Newborn Screening were able to develop a strategy which would meet both needs, that of maintaining the statewide Genetics Program as well as providing newborn screening training to hospitals: genetic counseling positions would be funded with newborn screening revenue.

ROLE OF REGIONAL GENETIC COUNSELORS

In December 1987, the Department of Public Health issued its first Request for Proposals for "Regional Newborn Screening and Genetic Service Coordinating Centers." The state had been divided previously into regions for the provision of genetic education and field clinics, with clinical geneticists generally agreeing on and respecting the geographic boundaries of those regions; genetic counselors successfully coordinated those efforts. With their communication and education skills, as well as knowledge of the screening disorders and other birth defects, genetic counselors were identified as appropriate professionals to work with hospital nurseries. An added benefit of working with nursery staff would be to create awareness of genetic referrals for infants with other conditions.

Since October 1, 1990, the Michigan Genetic and Newborn Screening Services Network has consisted of a central office at the Department of Public Health in Lansing with five regional centers. These are located at the University of Michigan (Ann Arbor), Wayne State University-Children's

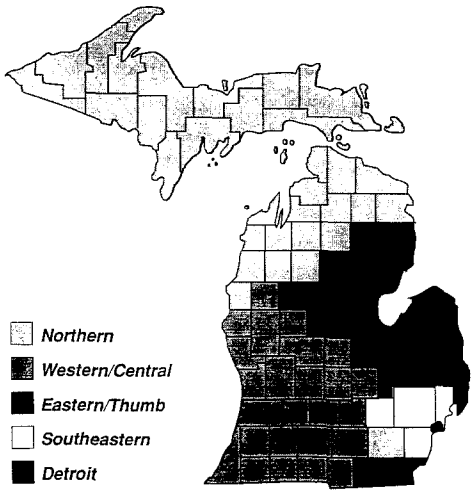

Fig. 1. Geographic regions for newborn screening coordination and genetic outreach.

Hospital (Detroit), Henry Ford Hospital (Detroit), William Beaumont Hospital (Royal Oak), and Michigan State University (East Lansing). Each center is responsible for providing newborn screening coordination, genetic services, and educational outreach in a specific geographic area. (Fig. 1)

After an initial orientation to state newborn screening policies and procedures, the counselors meet quarterly with staff to receive updates and coordinate efforts. The role of regional coordinator includes assuring proper specimen collection and monitoring hospital newborn screening procedures by serving as telephone resource for hospitals in the region, identifying newborn screening contacts within hospitals, maintaining up-to-date mailing lists, and working with hospitals to solve problems related to the newborn screening process, as illustrated in Table I. Another role is to provide on-site training to hospital staff. Typical in-service topics are included in Table II.

DISCUSSION

From October 1, 1989 through September 30, 1990, regional coordinators continued to monitor hospitals by making 66 visits to 45 different hospitals in all parts of the state. They have enhanced communication within

Table I. Issues Often Addressed by Genetic Counselors During Hospital Site Visits

1.	Internal hospital policies on newborn screening
2.	High frequency of late specimens
3.	Infants discharged before testing
4.	Incomplete demographic information on the card
5.	Large number of unsatisfactory specimens

hospitals as well as between hospitals and the State Newborn Screening Program. With just one state employee devoted full-time to newborn screening follow-up issues, feedback from the regional coordinators has improved the Program's ability to respond to hospital concerns, and to initiate appropriate policy changes. The success of the program is monitored in part by the use of quarterly hospital reports generated from the database. Criteria such as the number of cards from each hospital with no ethnic code or sample date, and the number of unsatisfactory or late specimens, provide an indirect method of assessing each coordinator's progress in identifying and solving problems related to the newborn screening process (Table III). Quarterly evaluation of the time period from birth to treatment for affected infants provides another way to identify factors which have delayed diagnosis; those that are hospital related can then be addressed by the regional coordinator.

The Genetics and Newborn Screening Program is currently highly regarded within the Division of Children's Special Health Care Services at

Table II. Topics Presented During Hospital In-Service Training Sessions

1.	History and background information on newborn screening in Michigan
2.	Heelstick instruction
3.	Interpretation of state policy
4.	Medical information about the disorders
5.	Explanation of follow-up protocols
6.	Clarification of newborn screening under special circumstances such as Transfusions Critically ill infants Hyperalimentation Discharge before 36 hours of age

Table III. Comparison of Newborn Screening Specimens Received Before and After the First Series of Workshops and Site Visits to Hospitals by Genetic Counselors

	Before (10/1/87–12/31/87)		After (4/1/89–6/31/89)	
Total initial newborn screens	33,957		36,546	
Specimens with no ethnic code	13,680	(40.29%)	3,737	(10.23%)
Specimens received over 5 days old	1,627	(4.79%)	1,301	(3.56%)
Specimens with no birthdate	94	(0.28%)	85	(0.23%)
Specimens with no sample date	3,458	(10.18%)	2,005	(5.49%)
Unsatisfactory specimens	598	(1.76%)	318	(0.87%)

the Department of Public Health. It is often showcased as a model for casefinding and prevention of disability. Because of the connection with newborn screening which is mandated by state law, funding for regional coordination of genetic services is expected to continue. Ongoing changes in the program as well as hospital staff turnover insure a continuing need for regional genetic coordinators to provide quality control, training and problem-solving resources to nurseries. Additional objectives for the future include more direct methods of evaluating each coordinator's services to hospitals, production of a newborn screening information manual for hospitals, development of a uniform training program with audiovisual materials, and increased physician education.

CONCLUSION

The Michigan Department of Public Health has identified a useful strategy to assure the continuation of state funding for genetic counselors, while improving the state newborn screening program. While 21 states and the District of Columbia monitor hospitals to ensure proper screening of all newborns, most employ a computerized system of matching birth certificates to specimens received or checking hospital discharge lists, without emphasis on improving quality or timeliness of the specimens being collected (Illinois Department of Public Health, 1988). Of the 29 states which charge a fee for newborn screening, California's system appears to be most similar to the one used in Michigan. Regional coordinators are utilized for hospital monitoring and education as well as follow-up of presumptive positive infants (CORN, 1991). Revenue from the fee supports these

activities, which are all directly related to the newborn screening program. In contrast to our program, California's coordinators are nurses by training, with one or two having become only recently ABMG certified as genetic counselors.

In Michigan, newborn screening activities have tied pediatric genetic centers in with all hospital nurseries and neonatal units, and the network of genetic counseling, outreach and education services has been strengthened. Most importantly, some 160 children benefit every year from the advantages of early detection and treatment, due to a more efficient newborn screening program.

REFERENCES

The Council of Regional Networks for Genetics Services (CORN) CORN Data and Evaluation Committee 1990 Newborn Screening Report, Draft No. 1, September 1991.

Illinois Department of Public Health (1988) Genetic Diseases Section Newborn Screening — An Overview of Newborn Screening Programs in the United States and Canada.