

MOUNTAIN STATES GENETICS REGIONAL COLLABORATIVE CENTER
Final report
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PROJECT: Implementation of Genetic Counseling Services Through Community Public Health Clinics:
Sickle Cell Trait Pilot Project

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RECIPIENT ORGANIZATION: Colorado Sickle Cell Treatment and Research Center

BACKGROUND SUMMARY:

In the course of screening for sickle cell disease, many more newborns are identified with sickle cell trait; in Colorado alone, approximately 400 are identified each year. Sickle cell trait is a generally benign carrier state that has no immediate health consequences. However, the recognition of sickle cell trait in an infant raises potentially important genetic counseling implications. Parents of an infant with sickle cell trait may be at risk for a subsequent child with sickle cell disease, given that at least one parent is presumed to have a sickle cell gene, and the other parent may carry a sickle cell or other hemoglobinopathy gene. Depending on the ethnicity of the parent, up to 10% may be silent carriers of a hemoglobin trait that may contribute to sickle cell disease in an offspring. In most states in the Mountain States Region, newborn screening follow-up for sickle cell trait is generally limited to notification of the infant's primary care provider indicated on the newborn screening form. In Colorado, educational materials intended for the family are included in a letter sent to the provider along with confirmatory test results. Discussion of health care issues and genetic counseling regarding this benign carrier state are left to the primary care provider. Currently there is no system in place to offer appropriate specific laboratory testing or genetic counseling information to these families in the context of the newborn screening program, as in done for families of infants with sickle cell disease. It is not clear how many families would be interested in such a program if it could be offered in a relatively convenient location and at little or no direct cost.

On a national level, there has been recent emphasis placed by the sickle cell community on sickle cell trait counseling. Based on our experience in Colorado, the extent of community interest in genetic counseling, supported by appropriate laboratory testing, is unclear. We propose to create a system which would reduce financial and access barriers to these services in an effort to determine the actual community interest in sickle cell trait counseling in the Mountain States Region. This project would initiate our approach in Colorado, where there is a large enough population to potentially warrant services, but not so large as to practically preclude implementation. By piloting programs through the public health community clinics, educating and integrating local public health professionals and trained lay community members, we hope to develop self-sustaining models of genetic counseling for sickle cell trait and other genetic carrier states that may be applicable to other states in the region.

PROJECT GOALS:

As a demonstration project in Colorado, we created a sickle cell trait counseling outreach program, utilizing existent community-based public health clinics in Denver, and Colorado Springs services

were offered to families identified by newborn screening as carrying sickle cell trait. These outreach programs were initiated in each city by the project staff, including newborn screening follow-up personnel and trained sickle cell trait counselors from the lay community. This pilot project provided:

1. Free laboratory testing, including complete blood count and hemoglobin electrophoresis, for persons identified by newborn screening to be potentially at risk for offspring with sickle cell disease
2. Consumer education and genetic counseling for at-risk individuals
3. Professional education of public health professionals in community public health clinics regarding sickle cell trait, sickle cell disease, and their genetic counseling implications
4. A model for the implementation of genetic counseling which is potentially applicable in all states with a similar public health system and for a variety of genetic disease and carrier states.

Ultimately, our hope was these programs would be sustained by the local community health clinic staff and community members on a periodic basis as indicated by the specific needs of the each community.

Project Accomplishments:

1. Advertisement to families of infants recognized to have sickle cell trait by newborn screening begin in August, 2006. Notification of outreach programs occurred by:
 - a. Letter accompanying the newborn screening confirmatory information sent to the infant's provider.
 - b. Direct phone call from the newborn screening follow-up program (Shannon Gillette and Nina Accornero) during routine follow-up contact.
2. Completion of training of two lay community members as sickle cell trait counselors at the national training course offered in New Britain, Connecticut in May, 2006.
3. Outreach programs were held in Colorado Springs on October 3, 2006 and Denver on November 16, 2006.
 - a. The Colorado Springs program was held at the Memorial Hospital Outreach Clinic, which was facilitated by Kathy Winder, RN, MSN; no facility fee was incurred
 - b. The Denver program was held at the Tri-County Health Department, Iliff WIC Office, which was arranged by contact through Colleen Doherty, WIC Education Office, Denver Health Medical Center declined to participate referred us to Aurora WIC who then referred us to Denver WIC.
 - c. Arrangements were made for programs in Weld County at the Weld County Public Health Bldg. in Greeley and Larimer Counties, but only a single family from this area of Colorado expressed an interest in attending; this family was invited to attend the Denver program.

Specific Activities and Results:

1. 158 families were contacted and invited to attend one of the outreach programs
 - a. 54 families expressed interest when contacted by phone and were provided additional information about the program locations and times over the phone and by mail
 - b. 9 families (2 in Colorado Springs and 7 in Denver) attended the clinics
 - c. 21 family members (5 in Colorado Springs, 16 in Denver) had blood samples taken for hemoglobinopathy testing (CBC and hemoglobin electrophoresis) and received genetic counseling during the programs

- d. 8 families considered themselves of non-African American heritage and 3 families were Spanish-speaking only. Services, including genetic counseling, were offered in Spanish through a medical translator.
2. All 9 families who participated in the programs received follow-up phone contact (Denver families) or in-person contact by the community sickle cell trait counselor (Colorado Springs families) to discuss laboratory test results.
 - a. 7 additional family members sickle cell trait carriers were identified, 1 with co-existent α thalassemia
 - b. 13 family members had normal test results
 3. Follow-up survey (see Appendix) was applied to participating families approximately 2 months following the programs to assess client satisfaction with the program and to answer any questions.
 - a. All families contacted felt the services provided by this outreach program were helpful and that their questions were answered.
 - b. All expressed they now understand how trait is passed from mother or father (or both in the case of sickle cell disease) to the child.
 - c. All families contacted felt the services provided by these outreach programs were of significant benefit and should be continued.
 - d. The Denver families felt the facilities used for the program at the Iliff WIC Clinic were inconvenient and would have preferred a different location.

PROJECT OBJECTIVES: Through the completion of this project, a number of objectives were achieved and prompt the following conclusion:

Objective 1: Establish the extent of consumer interest in specific genetic counseling information regarding sickle cell trait in this at-risk population.

Over the course of the project we contacted the families of infants with sickle cell trait identified through newborn screening over 6 months (01/06-06/06). These families were offered outreach sickle cell trait counseling and testing services. Of the 158 families contacted, 54 (34%) expressed interest and 9 families (6%) utilized the services of the programs. All family member that attended utilized free laboratory testing and received counseling services, and expressed .

Our experience suggests there is a small but significant portion of families with infants identified to carry sickle cell trait by newborn screening who would avail themselves of genetic counseling and laboratory testing when provided at no cost. This interest and utilization appeared to be higher in non-African American communities, and may reflect the “unexpected” nature of sickle cell trait in these populations. Outreach programs provide the opportunity to increase education and awareness of sickle cell disease in these at-risk communities as well as to the more traditionally affected community.

Objective 2: Based on the utilization of these services, determine the financial support necessary to sustain continued availability of laboratory testing at no or low cost to the consumer.

Laboratory testing for hemoglobinopathies included a complete blood count and hemoglobin electrophoresis, at a cost of approximately \$5,620 for 21 persons. Based on the apparent level of interest detected by this project, up to 40 families (10% of those identified with sickle cell trait by newborn screening) might take advantage of laboratory testing each year. If an average of 2 members in each family unit underwent testing, than approximately \$21,440 would be needed each year to test these 80 persons.

Our preliminary work to establish sites of outreach programs uncovered a paucity of public health and community clinics that offer phlebotomy services, especially for children. During the project, our team members obtained blood samples and University Hospital provided laboratory services. Other routes of sample procurement and testing might be explored if the volume of clients in these public health clinic settings warranted the establishment of a system to do so.

Objective 3: Develop a model in which the public health system is engaged in the provision of genetic counseling services for various disease states.

Based on impressions from contact with the public health clinic system, there appear to be few resources available for the incorporation of sickle cell trait counseling. Community clinic staff members seem significantly stretched to provide their current services, which do not include sickle cell trait counseling or capacity for blood sample procurement or easy access to appropriate laboratory testing. Nonetheless, there is still optimism that if outreach programs were held twice a year within this setting, initially supported by project team members, that the local staff would eventually develop an appreciation for and comfort with sickle cell trait, and may serve as a resource for future clients. This project also explored the establishment of outreach programs in other practice settings, including established genetics clinics (Weld County) and pediatric multi-specialty clinics (Colorado Springs), which may present additional opportunities for outreach.

Objective 4: Empower trained community members to serve with public health professionals in consumer education and genetic counseling.

Two lay community members received sickle cell trait counseling training through this project. These persons can now serve the community in this capacity in many different settings, in addition to supporting future formal outreach programs. Given the interest expressed by families from non-African-American communities, future training opportunities need to include members of these communities, especially native-speakers.