thalassemia is recognized by the World Health Organization as the most prevalent genetic blood disorder in the world. The thalassemias are a family of inherited blood disorders that affect an individual’s ability to synthesize hemoglobin, the protein in red blood cells that transports oxygen and other nutrients throughout the human body. Although treatments exist for the disorder, it is not curable and is often fatal. Currently, there are more than 300,000 thalassemia patients worldwide. In the United States, an estimated 2 million people carry the genetic trait for the disorder, and more than 100 children with severe thalassemia syndrome are born every year, 10 to 12 of them in Minnesota.

Because the incidence of thalassemia is particularly high among people of Asian descent, thalassemia is emerging as a public health problem of increasing importance in Minnesota, where the number of Asian immigrants continues to grow. Minnesota health regulations require newborn screening for a number of diseases and disease traits, including thalassemia, and place on the primary care provider the responsibility for communicating to parents the results of such tests. Unfortunately, the newborn screen remains an underutilized tool in the detection of the trait for thalassemia and other hemoglobin-related disorders due to inadequate follow-up by physicians.

To identify ways in which the newborn screen can be used more effectively in the prevention of thalassemia, it is necessary to identify the strengths and weaknesses of the current screening system in Minnesota. With support from CURA, we developed and conducted a survey of primary care physicians in the state that focused on their procedures for following up on newborn screens. This article reports our findings. First, we describe thalassemia and the treatment options available for the disorder. Then, we discuss the benefits and drawbacks of successful newborn screening programs for thalassemia and other diseases, and describe the screening system in Minnesota. Next, we present the results of the physician survey we conducted. Finally, we offer recommendations for strengthening and improving the newborn screening system in Minnesota and other states.

Thalassemia: Epidemiology and Clinical Presentation

The thalassemias constitute a tremendous economic and social burden for affected populations and for society as a whole. The increasing incidence of the disease in the United States and the nature of the disease itself constitute a public health problem of growing concern.

Epidemiology. The name thalassemia comes from the Greek root thalassa, meaning “the sea,” because the first reported cases were Mediterranean in origin. Today, the thalassemias are distributed across the Mediterranean region, the Middle East, the Indian subcontinent, and throughout Southeast Asia in a line stretching from southern China down the Malaysian peninsula to the Indonesian islands (Figure 1). In many of these countries, gene frequencies for the different forms of thalassemia are high.

In the United States, an estimated 2 million people carry the genetic trait for thalassemia, and each year, more than 100 children are born with the disease, 10% of whom live in Minnesota. As the number of people of Asian origin continues to grow in Minnesota, thalassemia is emerging as a public health problem of increasing importance.

Clinical Presentation. Although there are many variations of thalassemia, for the purposes of this discussion, the disease can be divided into two main types: beta (β) thalassemia and alpha (α) thalassemia.

1 The number of Asians living in Minnesota grew from 52,000 in 1990 to 130,000 in 1999, a 66% increase. This number is expected to continue to grow in the coming decades.
Thalassemia is inherited in a Mendelian recessive fashion. This means that parents who carry the trait and are symptom free have roughly a one in four chance of giving birth to a severely affected child.

**Beta Thalassemia.** Beta thalassemia, also known as Cooley’s anemia, is a fatal genetic blood disease that results in the failure to produce normal hemoglobin, the oxygen-carrying component of red blood cells. To remain alive, children born with beta thalassemia must undergo blood transfusions every two to three weeks. Over time, iron from the transfused blood cells builds up and becomes toxic to tissues and organs, particularly the liver and heart. Untreated, this condition leads to early death, typically through heart failure. To help remove excess iron, patients must undergo a daily infusion of the drug Desferal, a difficult and painful process that takes up to 12 hours. Although Desferal infusions have increased the life span of thalassemia patients, many find the daily treatments so burdensome that they abandon therapy altogether, which almost invariably leads to accelerated health problems and early death.

Successful bone marrow transplants can eliminate the need for blood transfusions and Desferal infusions in some cases of thalassemia, but because of the risks involved, transplants are considered an option only for the minority of patients who have a matched sibling donor.

Although many beta thalassemia patients are living longer as a result of these new therapies, longer life expectancy has itself lead to new problems. Many beta thalassemia patients who continue to undergo treatment end up struggling with secondary medical problems resulting from their condition such as heart disease, hepatitis, liver cancer, osteoporosis, and fertility problems.

**Alpha Thalassemia.** Alpha thalassemia shows several important differences from beta thalassemia. Because alpha chains, which are a component of hemoglobin molecules, are shared by fetal and adult hemoglobin, the disease is manifest in both fetal and adult life. The disorder results from the encoding of alpha globin by four genes (two from each parent) instead of the typical two genes (one from each parent). The clinical features of alpha thalassemia differ in severity depending on how many alpha globin genes are not functioning properly. The most severe form of the disorder (hydrops fetalis) is caused by four nonfunctioning genes, while the least severe form (no symptoms) results from one nonfunctioning gene.

Hydrops fetalis is found primarily, although not exclusively, among couples of Southeast Asian origin, and is encountered in increasing numbers in North America and elsewhere. These pregnancies inevitably result in fetal death during the third trimester of gestation or shortly after birth, and often are associated with serious maternal morbidity and even mortality. Hydrops fetalis may be detected by ultrasound during the second trimester of pregnancy. Frequently the diagnosis is made only after the birth of one or more hydropic newborns. The correct diagnosis of these hydropic fetuses is sometimes missed at autopsy. After experiencing the horrendous obstetrical, medical, and psychosocial problems associated with such pregnancies, many couples decide against becoming pregnant again, and some even request sterilization.

**Newborn Screening and Counseling for Thalassemia**

Advances in technology now permit newborn screening for an increasing number of genetic and nongenetic conditions. Routine screening for hemoglobin-related disorders began in 1987, after a National Institutes of Health (NIH) conference on sickle cell disease and other hemoglobin-related disorders recommended that every newborn child be screened to prevent the potentially fatal complications of sickle cell disease during infancy. As a result, newborn screening for sickle cell disease, thalassemia, and other hemoglobin-related disorders is now mandatory in 47 states, including Minnesota.

![Figure 1. Worldwide Distribution of Thalassemia, 1999](image-url)
counseling for any genetic disease, there are potential benefits and risks to screening newborns to detect their status as a trait carrier for thalassemia (Figure 2). The objective of the newborn screen for hemoglobin-related disorders is the early detection of infants with diseases such as sickle cell or thalassemia. However, the screen also detects infants who, although without a disorder themselves, are carriers of the gene for a hemoglobin-related disease. As many as 1 in 32 infants tested may be identified as a carrier of the trait, and neonatal screening may identify 17 to 100 times more carrier infants than infants with a full-blown disorder. However, the majority of parents of infants identified as genetic trait carriers are not notified of their child’s carrier status, and are not offered the option of confirmatory testing or counseling. This is particularly true in the case of alpha thalassemia. Indeed, several large-scale newborn screening programs nationwide do not report—either to the patient’s parents or to the physician of record—when alpha thalassemia trait has been detected in a child, and counseling programs currently do not exist for this disorder.

Although some may argue against follow-up and counseling in cases where a child is identified as a trait carrier, on the grounds that this is not the primary purpose of the newborn screen, these views ignore the right of parents to full access to genetic information about their child. Indeed, withholding such information violates legislative guidelines in many states that screen for genetic disorders. When parents are not informed of the carrier status of their child, they may wrongly conclude that the result of the newborn screen did not reveal any genetic anomalies.

In addition, the opportunity for parents to make informed reproductive choices is effectively lost when the initial identification of a newborn as a trait carrier is not followed by confirmatory testing for the infant, testing of other family members, and appropriate counseling. Experience with community-based thalassemia education campaigns and counseling programs in the Mediterranean has shown that such programs frequently influence parents’ subsequent reproductive choices. Since the late 1970s, voluntary screening of adults of child-bearing age, genetic counseling, and prenatal diagnosis have been introduced among populations in the Mediterranean region considered at risk for thalassemia. These programs have been marked by intensive educational campaigns (via mass media, schools, workplaces, and local community gatherings) about beta thalassemia and the methods for its prevention. The high level of acceptance of the program by the public has produced a dramatic reduction in the number of newborns with severe forms of thalassemia. In Sardinia, for example, the incidence of beta thalassemia major has dropped from 1 in every 250 births in 1975 to 1 in every 4,000 births in 1996, a prevention rate of 94%. In continental Greece, the number of newborns with thalassemia dropped from a high of 120 a year to only 15 cases in 1992. There is only limited experience with such programs in the United States. However, in a study conducted by Dr. Peter Rowley in Rochester, New York, 88% of Southeast Asian women offered prenatal counseling for thalassemia accepted it, and 90% brought their partners in for testing.2

The cost of establishing and maintaining thalassemia control programs in the Mediterranean has already been greatly outweighed by the economic and social benefits of the reduced incidence of the disease among newborns. The cost of transfusion and iron chelation for a child with beta thalassemia ranges from $30,000 to $100,000 per year. Although the advantages of a thalassemia control program are obvious in high-prevalence areas, even the cost of treating patients with thalassemia for life. In weighing the ultimate costs and benefits of such a program, one must consider not only the economic benefits, but also the numerous social benefits that accrue from preventing serious chronic illness and offering reproductive choice to families. Despite the obvious benefits of thalassemia screening and counseling programs, there are potential risks associated with such programs as well. Failed efforts to institute sickle cell screening in the United States during the 1970s illustrate some of these problems. Between 1970 and 1972, 12 states and the District of Columbia enacted mandatory sickle cell screening for all African-American citizens. More often than not, however, these laws were written and passed without adequate attention to the risk of stigmatizing patients—not only those with the disease, but also those who simply carried the sickle cell trait. Many African Americans identified as trait carriers were denied health and life insurance, employment opportunities, and even acceptance into the U.S. Air Force Academy. Thus, while screening and counseling programs can potentially be very effective at reducing the incidence of disease, it is important that community members are given input during the creation of such programs, and that the programs are accompanied by extensive education of healthcare providers, policy makers, and the community at large.

Figure 2. Benefits and Risks of Neonatal Detection of Carrier Status for Genetic Disorders

<table>
<thead>
<tr>
<th>Benefits of neonatal detection of carrier status:</th>
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</thead>
<tbody>
<tr>
<td>• Informed future reproductive choice of parents</td>
</tr>
<tr>
<td>• Informed future reproductive choice of child</td>
</tr>
<tr>
<td>• Opportunity to educate families about clinical implications of carrier status</td>
</tr>
<tr>
<td>• Social and psychological empowerment of families from ownership of genetic information</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Risks of neonatal detection of carrier status:</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Anxiety caused by misdiagnosis or misinterpretation of test</td>
</tr>
<tr>
<td>• Exposure of nonpaternity</td>
</tr>
<tr>
<td>• Social and psychological aftereffects</td>
</tr>
<tr>
<td>• Discrimination by employers or insurers</td>
</tr>
</tbody>
</table>


2
Newborn Screening in Minnesota.
In 1999, the Minnesota Department of Health (MDH) screened for five different conditions: congenital adrenal hyperplasia, congenital hypothyroidism, phenylketonuria, galactosemia, and hemoglobin-related disorders such as sickle cell disease and thalassemia. The department screens approximately 65,000 newborns per year. Since 1991, there has been a steady increase in the number of newborns who have hemoglobin-related disorders, or who carry the trait for such disorders (see Table 1). This increase is most likely due to the increase in the Southeast Asian population during the 1990s. According to the U.S. Census Bureau, the Asian population in Minnesota increased 66% during the last 10 years, mainly due to the increased emigration of Hmong people to Minnesota.

The State of Minnesota mandates follow-up for all genetic abnormalities detected by newborn screening. The specific procedures for follow-up care are dependent on the condition that is detected. All normal newborn screening results are mailed to the sample submitter, which is usually the hospital or clinic where the child was born. For samples that are unsatisfactory (e.g., due to an inadequate sample or a borderline result), the submittter is contacted and asked to resubmit the sample. Children whose test results indicate inborn errors of metabolism or major hemoglobin-related disorders such as sickle cell disease are tracked by MDH to verify that confirmatory testing is done to corroborate the initial screening results.

In contrast, MDH does not require tracking of or follow-up for children whose test results indicate they are genetic trait carriers for a hemoglobin-related disease such as thalassemia. In such cases, responsibility for patient follow-up lies with the primary care physician, who must order appropriate confirmatory tests, seek the opinion of an appropriate specialist regarding follow-up care, notify parents of test results, and provide (or refer parents to) appropriate genetic counseling. Because there is no centralized agency responsible for overseeing physician follow-up care, it is uncertain how frequently further testing is performed to confirm carrier status, or how often the parents of newborns are informed of the screening or confirmatory test results.

Evaluation of Parental Notification of Abnormal Newborn Screens
Because confirmatory testing and follow-up in cases of an abnormal newborn screen is the responsibility of the primary care provider, we sought to evaluate the follow-up care actually provided in such instances.

Method. A survey questionnaire was developed to solicit information from physicians involved in patient follow-up for newborn screening. Questions asked about such things as the timing of follow-up care, the procedure used for follow-up testing, what information was disclosed to the patient's family, the physician's opinion of his or her own experiences with patient follow-up, the timing of reporting of results, and the usefulness of the information on newborn screens provided by MDH. Space was also included for general comments regarding respondents' answers or the process of newborn screening in Minnesota.

Under Minnesota law, the newborn's place of birth is required to provide a screening sample to the Minnesota Department of Health. MDH recommends that the sample be collected between 24 and 72 hours after birth, and requires that the sample be clearly and accurately labeled with the newborn's name, date of birth, time of birth, specimen date, and collection time. The mother's name and other identifiers are included on the newborn screening card that accompanies the sample. To allow for accurate and efficient follow-up, all samples also require complete physician information, and the physician indicated on the lab slip is considered the “physician of record.”

We obtained from MDH a list of 126 physicians or clinics listed as the physician of record for newborns who had confirmed abnormal screen results for phenylketonuria, galactosemia, sickle cell disease, congenital hypothyroidism, congenital adrenal hyperplasia, or hemoglobin-related disorders. In cases where only a clinic was listed, the clinic was eliminated from the list. From the original list of 126, a total of 112 physicians were identified. We were ultimately able to locate the addresses and phone numbers of 76 of these physicians, either by contacting directory assistance or by referencing the AMA website (http://www.ama-assn.org). The remaining 36 physicians (32%) were either not listed, or had relocated and left no forwarding address.

A survey and cover letter were sent to all 76 physicians. A total of 33 completed surveys (43%) were returned. Of the respondents, 16 (48%) were pediatricians, 15 (45%) were family practice physicians, and 2 (6%) were neonatologists. Twenty-five of the respondents (75%) practiced in the Minneapolis–St. Paul area or surrounding suburbs, while 8 respondents (25%) practiced in greater Minnesota. Although the respondents were demographically similar to those who were initially sent surveys, their responses may not be representative of the opinions and practices of the majority of physicians in Minnesota.

Results. Results from the physician survey are presented below. It should be noted that the level of physician interest and participation in newborn screening is most likely overemphasized in this study because those who were more interested were more likely to complete and return the survey. Thus, whatever percentage of these respondents indicated difficulty notifying children’s parents of their newborn screen results or providing follow-up care, there is probably a correspondingly larger percentage of parents of newborns who are not notified of the results of their child’s screen or offered follow-up care.

Table 1. Number of Abnormal Newborn Screen Results in Minnesota, 1991–1998

<table>
<thead>
<tr>
<th></th>
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<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital adrenal hyperplasia</td>
<td>—</td>
<td>—</td>
<td>35</td>
<td>48</td>
<td>64</td>
<td>67</td>
<td>37</td>
<td>35</td>
</tr>
<tr>
<td>Congenital hypothyroidism</td>
<td>15</td>
<td>24</td>
<td>54</td>
<td>79</td>
<td>83</td>
<td>93</td>
<td>33</td>
<td>49</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td>7</td>
<td>6</td>
<td>4</td>
<td>7</td>
<td>9</td>
<td>17</td>
<td>9</td>
<td>10</td>
</tr>
<tr>
<td>Hemoglobin-related disorder</td>
<td>33</td>
<td>24</td>
<td>18</td>
<td>21</td>
<td>16</td>
<td>23</td>
<td>26</td>
<td>23</td>
</tr>
<tr>
<td>Hemoglobin-related disorder carrier</td>
<td>899</td>
<td>1,099</td>
<td>960</td>
<td>1,012</td>
<td>1,202</td>
<td>915</td>
<td>1,094</td>
<td>1,524</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>19</td>
<td>15</td>
<td>29</td>
<td>15</td>
<td>24</td>
<td>4</td>
<td>8</td>
<td>9</td>
</tr>
</tbody>
</table>

Source: Dr. David Jinc, Director, Newborn Screening Department, Minnesota Department of Health
Type of Abnormal Newborn Screen.
Of the 33 respondents, all reported having had a patient with an abnormal newborn screen during the last five years for one of the five conditions screened for by MDH. Twenty of the respondents (60%) reported having had two or more patients with an abnormal newborn screen within the last five years. A breakdown of responses by type of abnormal screen is shown in Table 2.

Of the respondents, 31 (94%) reported having had a patient with an abnormal screen for a hemoglobin-related disorder such as thalassemia, 18 (55%) had a patient with an abnormal screen for congenital hypothyroidism, 5 (15%) had a patient with an abnormal screen for phenylketonuria, 10 (30%) had a patient with an abnormal screen for galactosemia, and 7 (21%) had a patient with an abnormal screen for congenital adrenal hyperplasia.

Table 2. Number of Abnormal Newborn Screen Results Reported by Physicians

<table>
<thead>
<tr>
<th>Genetic disorder</th>
<th>Reported incidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hemoglobin-related disorder</td>
<td>31</td>
</tr>
<tr>
<td>Congenital hypothyroidism</td>
<td>18</td>
</tr>
<tr>
<td>Galactosemia</td>
<td>10</td>
</tr>
<tr>
<td>Congenital adrenal hyperplasia</td>
<td>7</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td>5</td>
</tr>
</tbody>
</table>

Reasons cited for not discussing screen results with parents included the following:
- long delay in reporting of normal results by MDH/information not available to discuss with parents (5 out of 33, or 16%)
- time constraints (4 out of 33, or 13%)
- disinterest of parents (4 out of 33, or 13%)
- unavailability of parents (2 out of 33, or 6%)
- language barriers (1 out of 33, or 3%)

Timing of Follow-Up Care and Reporting of Results. Fourteen respondents (42%) indicated that the age of the newborn at the time parents were informed of an abnormal newborn screen was generally between three days and one week. Eleven respondents (33%) reported that the age of the newborn was generally one to two weeks, while the remaining respondents stated that the newborn was either two to three weeks old (7 out of 33, or 21%) or more than three weeks old (3 out of 33, or 9%). One respondent noted that the timing was dependent on the condition for which the patient had an abnormal screen. Those respondents whose patients were more than two weeks of age when parents were notified indicated that the condition for which the newborn had an abnormal screen was generally a hemoglobin-related disorder.

Most respondents (31 out of 33, or 94%) reported that they made efforts to perform confirmatory testing that same day for the condition for which their patient had an abnormal screen. Those who did not provide confirmation the same day were likely to provide results within one week. Those who exceeded one week in confirming the results explained that it is customary to confirm results for hemoglobin-related disorders after six months of age.

A majority of the physicians (27 out of 33, or 82%) indicated that screen results were provided to them by MDH in a timely manner. Six respondents (18%) felt they did not receive results in a timely manner, while four (12%) indicated that both the hospital and MDH were slow to report normal results. Two respondents (6%) indicated delays in abnormal newborn screen reporting, and noted that newborns are often symptomatic with congenital adrenal hyperplasia or congenital hypothyroidism by the time the respondent is notified of screen results. This appeared to be a particular problem in the hospital setting, where the attending physician is generally notified of screen results, even though he or she may not be responsible for the newborn’s primary care and therefore may not document the results in the newborn’s chart in a timely manner.

Information Provided to Parents. According to most respondents (29 out of 33, or 88%), parents were generally aware that newborns were screened for genetic disorders. Only four respondents (12%) indicated that most parents were unaware their child had undergone newborn screening. One respondent noted that most parents were aware of screening for phenylketonuria exclusively.

Table 3 provides a breakdown of the type of information respondents typically provide to parents of a newborn who has had an abnormal test result for one of the conditions screened for by MDH. Most respondents (30 out of 33, or 91%) said they discuss what steps need to be taken to obtain diagnostic confirmation. Additionally, most physicians said they discuss clinical information regarding the condition (28 out of 33, or 82%) and referral to a specialist (22 out of 33, or 67%). Other topics discussed include genetic inheritance of the condition (12 out of 33, or 36%), trait carrier testing for parents and other family members (8 out of 33, or 24%), and availability of community and support groups.

Table 3. Information Provided to Parents of Newborns with a Positive Newborn Screen Result

<table>
<thead>
<tr>
<th>Type of information provided</th>
<th>Pct. of respondents</th>
</tr>
</thead>
<tbody>
<tr>
<td>Information about obtaining diagnostic confirmation</td>
<td>91</td>
</tr>
<tr>
<td>Clinical information about the condition</td>
<td>82</td>
</tr>
<tr>
<td>Referral to a specialist</td>
<td>67</td>
</tr>
<tr>
<td>Information about inheritance of the condition</td>
<td>36</td>
</tr>
<tr>
<td>Information regarding carrier testing for parents/other family members</td>
<td>24</td>
</tr>
<tr>
<td>Information regarding community groups and support groups</td>
<td>12</td>
</tr>
</tbody>
</table>
support groups (4 out of 33, or 12%). A majority of physicians stated that their clinic had available—or had access to—patient information on the conditions screened for (31 out of 33, or 94%), patient information on support groups (27 out of 33, or 82%), carrier testing facilities (27 out of 33, or 82%), and a genetic counselor (29 out of 33, or 88%). The two respondents (6%) whose clinics do not have patient information on the conditions also said they do not have access to this information. The six respondents (18%) whose clinics do not have support group information stated they were unaware of where to access this information. The six respondents (18%) whose clinics do not provide carrier testing for family members indicated that they had to refer family members for such testing due to difficulties in billing. Finally, the four respondents (12%) whose clinics do not provide genetic counselors stated that they refer parents to similar specialists, such as a geneticist.

Evaluation of Minnesota Department of Health. All of the respondents reported that the screen results provided by MDH were easy to understand. Most respondents (29 out of 33, or 88%) felt that the information allowed them to efficiently provide testing to confirm the initial diagnosis. Those who did not agree (12%) stated that the results do not clearly indicate the best next test, or provide only partial direction. Most respondents (29 out of 33, or 88%) stated that they could readily locate an appropriate specialist based on the information provided. Those who did not agree (12%) stated that they could not locate the name or number of a specialist. Additionally, one respondent indicated that specialists for hemoglobin-related disorders were easy to identify, but that it was more difficult to locate specialists for other conditions. Finally, two respondents (6%) indicated that it was difficult to find a specialist in the provider insurance network. A majority of respondents (27 out of 33, or 82%) stated that the information provided by MDH allowed them to readily identify resources available to the patient, such as supplemental insurance and support groups. Those respondents who felt they could not identify patient resources (18%) indicated they were not aware of any resources available to them through the Minnesota Department of Health.

Improving Follow-Up Care Based on Newborn Screens
As with any genetic disease, there are potential benefits and risks to screening newborns to detect their status as a trait carrier for thalassemia. Accordingly, newborn screening, confirmatory testing, parental notification, and counseling should be carried out in the context of a carefully designed and structured program. Involvement of parents and consumer groups, providers, and policy makers is important in designing models of delivery of such services. At minimum, a comprehensive program to improve identification and counseling of individuals with thalassemia should require and establish standard reporting procedures for universal notification of thalassemia screen results, offer genetic counseling to the families of all children with an abnormal newborn screen, create public educational campaigns about thalassemia and the methods for its prevention, and make healthcare providers aware of the importance of newborn screening and parental notification for thalassemia.

Identifying and Locating the Physician of Record. One of the most common difficulties with follow-up for newborn screens is incomplete or illegible information accompanying the sample, including misspellings or misinformation regarding the primary care physician. In a previous study, for instance, J. M. Tuerck found that 25% of samples were missing essential information such as name and address of patient or the primary care provider. In our study, insufficient information about the physician of record on the list of names provided by MDH frequently made it difficult to identify the primary care provider. More than 30% of the names on the list were insufficient or inaccurate, making it impossible to locate the appropriate physician in those cases. Common problems included incomplete physician names, or names of physicians who appeared on the newborn screening samples received by MDH, but who no longer practiced medicine. MDH offers no guidelines for indicating the physician of record on the newborn screen sample. Because MDH does not directly notify parents of the results of the newborn screen, if the physician of record cannot be located, parents will most likely never learn of an abnormal screen result. Educating both parents and hospitals about the importance of completing the newborn screening card may help increase compliance.

Notification of Results. Respondents indicated that they were not able to provide universal disclosure of newborn screening results because they routinely received results from MDH after the newborn’s two-week clinic visit. One possible reason for the delay may be that results have to go through the screening sample submitter (the place of birth) before being returned to the primary care physician. The submitter may also have difficulty notifying the physician of record based on incomplete or inaccurate information provided on the sample. Studies of other newborn screening programs have noted that a common cause of delay is late submission of the sample to the laboratory. The delay in submission could make it difficult to provide the results to the physician of record in a timely manner.

As previously explained, screening for hemoglobin-related disorders such as thalassemia identifies not only newborns affected with a disease, but also carriers who will not develop the condition but are at risk of having offspring with the disease. The biological parents of a child identified as a trait carrier are also at risk of having an affected newborn in the future. One survey respondent indicated that he or she did not disclose screen results to parents of newborns who are carriers for a hemoglobin-related disorder. Not disclosing such information deprives the newborn and its family of information that may have a profound effect on future family-planning decisions. In addition, all newborn screening results can potentially be placed in the newborn’s medical records, whether parents are informed of the results or not. It is clearly unjust to put a newborn trait carrier at risk for genetic discrimination by insurance companies, employers, or others without notifying the parents of the child’s trait carrier status.

Changes in notification and submission must be made to ensure that universal notification of parents is possible, efficient, and effective. To ensure universal disclosure of newborn screening results, all patients of newborns, as well as their primary care providers, should receive a copy of the child’s screening results.

Information Provided to Patients. The responsibilities of primary care providers regarding patients with genetic

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conditions include proper identification of patients who might benefit from genetic services, appropriate referral, and recognition of the psycho-social needs of families who have known genetic conditions or susceptibility to these conditions. Less than half of respondents indicated that they did provide inheritance information to the family about the genetic conditions screened for, and even fewer discussed carrier testing with or provided support group information to parents of newborns with an abnormal newborn screen result. Several respondents indicated they did not have access to or could not locate support group information.

Based on the responses of physicians in this study, we do not know the quality of the information provided. Studies have found that physicians can provide genetic counseling, but that they need adequate time with the patient and training in genetics to be effective.\(^5\) It is well known that primary care physicians in the United States are allotted very little time with their patients. In addition, the genetic knowledge of physicians has routinely been shown to be inadequate, both for communicating inheritance information to patients and for understanding genetic information.\(^6\)

Many molecular genetic testing laboratories state in their reported results whether genetic counseling is indicated for a particular result. The Minnesota Department of Health should consider including a similar statement in the case of abnormal newborn screen results, particularly in those cases where further testing has confirmed their putative diagnosis. Whatever plan is ultimately adopted, it is important that all providers involved in newborn care are educated about the notification process and the importance of reporting results to parents in a timely manner. Minnesota is one of the few states that currently does not provide formal education to primary care providers or parents. Many of the difficulties voiced by primary care physicians could be overcome with a better working knowledge of the screening tests and the resources provided by MDH.

To further ensure that appropriate follow-up care is provided by qualified individuals, children with an abnormal newborn screen result and their families should be matched with a qualified genetic expert. Many other states provide one-on-one counseling to children with a hemoglobin-related disorder or trait and their families, and several have trait counselors who provide counseling for all trait results. Currently, MDH does not provide or require such counseling. Periodic evaluation of the program should be built into the follow-up system to ensure quality care. Further research is also needed to evaluate both the consequences of a carrier-state diagnosis for infants and parents, and the cost effectiveness of different models of delivering test results and counseling.

**Conclusion**

Thalassemia and other hemoglobin-related disorders are among the most common genetic diseases in the world. They constitute a tremendous health, economic, and social burden for affected populations and the healthcare systems that serve them. With the continuing demographic change in the United States, and in Minnesota in particular, such disorders are growing in importance as public health problems. Universal newborn screening aims at early detection of severe forms of hemoglobin-related disorders. Detection of trait carriers for such disorders is an inadvertent consequence of newborn screening, but it provides a valuable tool for identifying affected families who might benefit from genetic counseling. Effective public health programs have been implemented in several countries for screening, counseling, and prevention of these diseases, and they have been well received by the affected populations. In Minnesota, the onus of confirming screen results and counseling families rests with the primary care provider. This study has shown that information regarding the genetic carrier state of a newborn for a hemoglobin-related


Because thalassemia and other hemoglobin-related disorders constitute a tremendous health, economic, and social burden, a comprehensive program to improve identification and counseling of individuals with thalassemia is needed.
Project Awards

To keep our readers up-to-date about CURA projects, each issue of the CURA Reporter features a few capsule descriptions of new projects under way. The projects highlighted in this issue are made possible through CURA’s Program for Interactive Research. All five projects involve significant issues of public policy, and include active participation with communities, groups, or organizations in Minnesota. The projects described here represent only a portion of those that will receive support from CURA and its partners during the coming year.

- James McManus (Large Lakes Observatory, UMD) will investigate the causes of eutrophication of two urban lakes in Virginia, Minnesota, and work with community members to identify ways to reclaim the lakes for recreational use.

- Judith M. Garrard and Susan B. Foote (Health Services Research Policy, School of Public Health) will collaborate with the Minnesota State Patrol to research state trends among individuals driving under the influence of drugs, as well as criminal justice outcomes for those arrested for such violations.

- Laura Kalambokidis (Applied Economics) will investigate the economic impact of alternative land use policies on Minnesota communities through case studies of Chisago and Isanti counties.

- Kathryn D. Rettig and Kerry Kriener-Allen (Family Social Science) will examine the effectiveness of the Minnesota Child Support Guidelines in meeting the fiscal needs of children of divorced, unmarried, and never-married parents.

- Melissa M. Stone (Humphrey Institute of Public Affairs) will work with the Ramsey County Community Employment Partnership to conduct a case study of Work Resource Hubs, collaborations that are used to implement social welfare reform and workforce development.

Update: The Minnesota Population Center

The Minnesota Population Center (MPC) recently celebrated its first birthday by counting its accomplishments: more than $10 million in research grants and designation as a national center for population research by the National Institutes of Health, one of only 13 such centers in the country.

MPC was officially launched in March 2000 with a $100,000 grant from the Vice President for Research under the Interdisciplinary Research, Scholarly and Creative Activities Program (IRSCA). The center is an interdisciplinary unit devoted to research and teaching on demography, the statistical analysis of human populations. Since its creation, the center has enjoyed tremendous growth, and currently supports 38 faculty and postdoctoral members and 80 support staff who are engaged in a variety of population-related research projects.

Because of its empirical orientation and emphasis on policy-relevant research, MPC has devoted considerable effort to developing the technology to share demographic data worldwide. For example, the Integrated Public Use Micro Sample (IPUMS) is a two-century-long statistical sample of raw census returns that MPC has made accessible online to researchers and analysts worldwide. Another project currently under way—the National Historical Geographic Information System (NHGIS)—is an effort to collect, standardize, and make publicly available all existing aggregate-level census data from 1790 to 2000.

In addition to its research projects, the Minnesota Population Center hosts specialized forums, workshops, and symposia on topics related to population research, including a brown bag seminar series that brings nationally and internationally recognized demographers from a variety of disciplines to Minnesota to share their research. The center is also one of three coordinating members of the Minnesota Data Center (MNDC) network, serving as the data depository and providing a fee-based custom data service.

CURA is enthusiastic about this new center because of its obvious implications for policy research and outreach relevant to Minnesota. CURA and MPC are cosponsoring the Conference on Demographics for Policy Analysts this October (see announcement on page 28), and hope to work together on other initiatives.

For more information about the Minnesota Population Center, visit http://www.pop.umn.edu, or contact MPC’s director Steven Ruggles at ruggl001@umn.edu or (612) 624-4081.
CURA’s Trade Centers of Minnesota Project Receives Award

A group of researchers from CURA and the Minnesota Department of Transportation (MnDOT) has received an award from the University of Minnesota’s Center for Transportation Studies (CTS) recognizing their contributions to the Trade Centers of the Upper Midwest project. The team of six researchers from CURA (William Casey, the late Tom Anding, and Barbara Lukermann) and MnDOT (Abby McKenzie, Cecil Selness, and Cathy Gillaspy) was honored with a special recognition award under the CTS Research Partnership program. The award is intended to recognize research projects within the research program that result in significant impacts on transportation, and reward teams of individuals who have drawn on the strengths of their diverse partnerships to achieve those results.

Beginning in summer of 1998, the team from CURA and MnDOT collaborated on an update to an earlier analysis of trade centers in the region conducted by Tom Anding and others. The team’s final report, *Trade Centers of the Upper Midwest 1999 Update*, was prepared by William Casey and published by CURA in June 1999. The update focused on identifying higher order trade centers in the Upper Midwest that served relatively large geographic areas, and was used by MnDOT as the basis for allocating transportation funds to Minnesota communities.

The Trade Centers of the Upper Midwest project was originally initiated in 1963 by CURA’s first director, the late John R. Borchert, and produced a report by Borchert and Russell B. Adams titled *Trade Centers and Trade Areas of the Upper Midwest*. Borchert and Adams described an interconnected system of economic trade centers stretching across the region, and classified population centers in Minnesota, Montana, North Dakota, South Dakota, and Wisconsin into one of eight categories based on the extent to which they provided employment and services to nearby communities and surrounding rural areas. Using Borchert and Adams’ work as a starting point, Anding and others later expanded the scope of the project to include Iowa and Nebraska, and updated the analysis using computerized data sets unavailable earlier. Their report, *Trade Centers of the Upper Midwest: Changes from 1960 to 1989* (1990), described a complex economic system that had continued to evolve and change throughout the period under study.

New Publications from CURA


One of CURA’s most popular publications, the directory lists more than 600 not-for-profit associations, organizations, and mutual assistance and fraternal groups in the state of Minnesota that are controlled by people of color or primarily serve one or more communities of color. The directory provides the name and address of each organization listed and, where available, phone and fax number, email address, Web address, name of contact person, and a brief description. Organizations are indexed alphabetically by name, by the community the organization serves, and by the main activity in which the organization engages. Also included are mailing label matrices for all organizations listed.

The directory is also available on the Web as a searchable database. The online directory contains all of the information in the latest print edition, and will be regularly updated to reflect additions, deletions, or changes to the listing of organizations. Visitors can search for specific organizations by community served, major activity area, and keyword; make customized mailing labels based on search results; update or add their organization’s listing online; and limit their search to those organizations whose listings have been added or updated since the print version of the directory was published in September 2001.

The online edition of the directory can be found at http://www.cura.umn.edu/publications/nporc.html. Single copies of the print directory are available upon request. To order, contact CURA by phone at (612) 625-1551, by fax at (612) 626-0273, or by e-mail at cura@umn.edu.


This article, co-authored by CURA associate Esther Wattenberg, explores the circumstances of parents whose extreme neglect and abuse of their young children resulted in the drastic state action of termination of their parental rights. The authors review the records of 97 children in Minnesota aged 6 and under whose parents’ parental rights were terminated between 1991 and 1997, develop profiles of these children and their parents, and identify a “risk pool” of parents whose children became wards of the state. Based on this risk pool, the authors establish guidelines that may help alert practitioners to those parents who are unlikely to safely maintain their children. Questions and implications for policy and practice are highlighted. Reprints of this article are available from the University of Minnesota Center for Advanced Studies in Child Welfare by calling Anne Preston at (612) 624-4231 or sending e-mail to cascw@che.umn.edu.
Conference on Social Justice Issues, October 27

Citizens for Democracy, a Twin Cities group committed to citizen participation in democracy, will host a one-day conference in Minneapolis on Oct. 27, 2001, to examine issues of social justice, civic engagement, and current policy agendas that depend on citizen support. The conference, titled “A Citizen’s Call to Justice: Creating a New Democracy,” will focus on impending policy decisions that affect people’s lives and on creating a network of progressives dedicated to revitalizing democracy. Scheduled speakers include John a. powell, executive director of the Institute on Race and Poverty at the University of Minnesota; Peter Edelman, professor of law at Georgetown University; and Senator Paul Wellstone (DFL-Minnesota).

CURA, the Institute on Race and Poverty, the Headwaters Fund, VoterMarch Minnesota, the DFL Education Foundation, the Minneapolis Urban League, and the Minneapolis branch of the NAACP provided initial funding for this event.

For more information or to register for the conference, visit http://www.umn.edu/irp or call (612) 624-2904.

Third Upper Midwest Conference on Demographics, November 29

The Third Upper Midwest Conference on Demographics for Policy Analysts will explore the theme “The Changing Face of Demographic Data: Census and Other Data Sources for Policy Analysis.” Cosponsored by CURA, the Minnesota Population Center, and other research centers and state agencies engaged in demographic analysis, this gathering will provide a unique opportunity for both experienced data analysts and newcomers to learn about census and noncensus data available for policy analysis, recent population trends, new methods of analysis, and current trends in the data environment. The conference will be held Thursday, November 29, 2001, at the Earle Brown Continuing Education Center on the University of Minnesota St. Paul campus. Visit http://www.pop.umn.edu/midwest_main.html for more information, or contact Noreen Huntington at (612) 625-6748 or nhunting@cce.umn.edu.