Long-term Effects of a Community-wide Hemoglobinopathy Screening Program

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Methods

For more than two decades, Queens Hospital Center, a large municipal hospital with a predominantly minority patient population, had a community-based hemoglobinopathy education, screening, and treatment program. These services, which were simultaneous and ongoing throughout the years of the study, included the education of more than 250,000 community residents, the screening of more than 60,000 individuals, and genetic counseling of more than 10,000 patients. Education consisted of one-on-one sessions with counselors or educators, lectures to school and community groups, and pamphlets. Program staff scheduled educational sessions in many public elementary and high schools throughout Queens and Brooklyn. Thousands of at-risk clients, defined as either African American or Caribbean American ethnicity or having a positive family history of sickle cell disease, and other clients were thus reached and offered education and screening. Screening consisted of alkaline electrophoresis of peripheral blood in children and adults and of dried blood on filter paper from newborns. Those who tested positive for a hemoglobinopathy were then offered genetic counseling. Almost exclusively, African-American counselors with deep roots and professional and personal roots in the community delivered the services, thereby making these services culturally attractive. A universal, state-run newborn screening program reported all newborn screen results, regardless of race, directly to our program, because we were responsible for the medical follow-up of neonates identified with hemoglobinopathies. Other services included family counseling and family member screening. The program also included informal linkages with community-based social and religious organizations which afforded opportunities for education in churches, community centers and the like, as well as formal agreements with other prenatal care providers in our area, including community hospitals and teenage pregnancy clinics. A large clinical service grant from New York State supported all program elements other than newborn screening. Treatment consisted of medical treatment of sickle cell disease and its complica-
tions at our sickle cell clinic. We chose three objective criteria to evaluate the efficacy of our comprehensive program. We compared the number of infants born with a hemoglobinopathy over a five year period (1987-1991) with the expected number based upon population norms. We reviewed the level of prenatal diagnosis utilization over the same time interval. Prenatal diagnosis, in the form of amniocentesis, was offered to all at-risk pregnant women, regardless of the father's carrier status. Finally, we compared the number of hemoglobinopathies diagnosed prenatally with the number diagnosed as a result of universal newborn screening.

Results

Among sickle cell diseases, HbSS, HbSC, and Hb/β-Thal are the most common sickling disorders among African-Americans. HbSS is found in approximately 1/400, HbSC in 1/1000 and HbS/β-Thal in 1/5000 African American newborns (Serjeant, 1985). These incidences are used only as an approximation, because a more accurate population estimate would have to take into consideration the different incidences in our ethnically diverse African American sample. In the five calendar years, 1987-1991, the total number of births at Queens Hospital Center was 14,051 of which 7,714 were African American. Newborn race was based upon the parents' self-report in response to an open-ended question. No further questions were asked about other races or mixed racial origins. Using the estimates above, the calculated number of African American births with the three aforementioned hemoglobinopathies was 29 and the observed number was 36 (23 were HbSS and 13 HbSC). An exact binomial test was utilized to assess the consistency of the observed and expected numbers of newborns with hemoglobinopathies. From general population incidence figures, the probability of a hemoglobinopathy in an African American newborn is 1/400 + 1/1000 + 1/5000 = 0.0037. The exact binomial probability of 36 or more cases among 7,714 births is therefore 0.099, showing no significant difference between observed and calculated cases. The estimated number of at-risk pregnancies during that same period was 144 (4x36). However, only 55 at-risk pregnant women presented early enough in pregnancy (before twenty weeks) for us to offer prenatal diagnosis. Thirty-three affected babies were born to the remaining 7,695 at-risk women who were not identified early enough to have amniocentesis. Since 1990, prenatal diagnosis has been offered to women, regardless of whether or not the father's carrier status was known. It is offered irrespective of the patient's interest in terminating an affected pregnancy. Only 19 of the 55 couples that were identified to be at-risk and were offered prenatal diagnosis elected to undergo the procedure. Three of these 19 pregnancies were found to be affected and resulted in 2 terminations of pregnancy and 1 continuation to term. There were no fetal losses among the seventeen pregnancies that went to term. Therefore, only 19/144 or approximately 13% of couples at-risk received prenatal diagnostic testing and only 3/36 or approximately 8% of children with hemoglobinopathies were diagnosed prenatally during a five year period.

Discussion

Neonatal screening programs for hemoglobinopathies have been adopted because of the evidence that prophylactic penicillin lowers mortality and morbidity when given within the first few months of life (Grover, 1989). Early advocates of prenatal diagnosis for hemoglobinopathies suggested that the effectiveness of such programs depends upon community-wide education and counseling efforts. Driscoll et al. (1987) reported that as a result of the lack of such services, only approximately 5% of pregnancies at-risk in the New York metropolitan area received these services in 1987 (3). By 1989, Rowley (1989) had concluded in a survey of prenatal programs in the United States and Canada, that these programs should not be judged solely upon the number of prenatal diagnoses and the number of terminations of affected pregnancies. The 272 prenatal diagnoses made in 1987 in the United States constituted only 4.1% of the estimated pregnancies at-risk for hemoglobinopathies. The pregnancy termination rate in this same study for HbSS was 39%. Schoen et al. (1993) demonstrated that prenatal diagnosis was not a cost-effective method of identification when compared to newborn screening alone. We agree with Rowley that programs such as ours should not be assessed solely on the number of terminations of pregnancy following prenatal diagnosis of an affected fetus. Rather, the benefits of our activities in the community may be a better educated population, more informed decisions by couples at-risk, and additional options for those availing themselves of prenatal diagnosis. Future studies might incorporate a means to evaluate some of these benefits. In addition to providing at-risk women with prenatal diagnoses, we also offered a comprehensive integrated program utilizing neonatal screening, education, and outreach. The subjective benefits of such a comprehensive program are advantageous as a supplement to our goal of early diagnosis. Newborn screening programs, even if mandatory as in New York State, are not designed to encourage parents to opt for prenatal diagnosis. The low prenatal diagnosis rate in our sample may be due to several factors: patients register late for prenatal care, object to prenatal diagnosis for personal reasons or would not terminate the pregnancy even if the fetus is affected. Thus, we believe that the main benefits of our program are the availability of prenatal testing for those women who want it, the education of a large population of African and Caribbean Americans about hemoglobinopathies, and the counseling of clients who were identified as having a hemoglobinopathy trait about reproductive possibilities and options. We were able to quantify the level of
utilization of prenatal diagnosis in our patient population. We were unable to quantify the community's level of knowledge about hemoglobinopathies.

References


