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Pathways to Genetic Screening:
Patient Knowledges - Patient Practices

TECHNICAL REPORT OF RESEARCH PROGRESS II

This study is designed to clarify the integration of genetic knowledge into the lived experience of high-risk family members. A major focus is elucidation of the social and cultural barriers and bridges to the use of genetic information to increase reproductive options and improve the quality of family life. The study was originally planned to span three years and focus on families at risk for cystic fibrosis and sickle cell disease. These two disease groups were selected because they are among the most common potentially lethal genetic diseases and because while each has a similar pattern of inheritance and raises similarly serious bio-medical challenges and issues of information management, they primarily affect different racial and ethno-cultural groups permitting a "naturally occurring experiment" in the variable penetration and meaning of genetic medicine in two populations.

SUMMARY OF OVERALL PROGRESS

Our first year of funded fieldwork and interviewing ended May 31,1993. From June 1, 1993 to March 4, 1994, we were forced to cut our operations to a minimum due to a funding hiatus. However, the assistance of a supplementary grant from DOE enabled us to keep the project going at a minimal level. Funding has now been resumed at a level that permits us to continue data collection and move into the preliminary stages of analysis.

DISCLAIMER

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We have maintained our ties to Children's Hospital Sickle Cell Program, Oakland (CHO) and to the California Pacific Medical Center's Cystic Fibrosis Treatment Program, and the Alta Bates Medical Center's Adult Sickle Cell Program to the present, attending a variety of medical, genetic and psycho-socially oriented meetings and seminars to maintain an up-to-date understanding of clinical issues related to sickle cell and cystic fibrosis. These institutions have continued to serve to identify high-risk families and as sources of index cases.

As of August 1994 we have conducted intensive interviews with more than 300 individuals in approximately 88 families. We have attempted to balance the effort equally between the two groups, but have found we are able to penetrate the family systems of the cystic fibrosis families more easily than the sickle cell families. This is indicated by the smaller proportion of CF as opposed to SC families (25:63) but larger number of CF than SC respondents (170:132).

PROBLEMS AND FAVORABLE DEVELOPMENTS

Confidentiality. We have encountered no new serious problems related to sickle cell or cystic fibrosis families. We have encountered a number of potentially sensitive methodological issues with regard to interviewing many members of one family. The most serious is the necessity of taking special precautions not to inadvertently divulge information from one family member to another. Turnover of graduate student interviewers means that it is necessary to spend substantial amounts of time training on sensitivity to issues of privacy and reinforcing that training. These same issues of confidentiality are arising as we prepare reports on family dynamics. Since each family is unique and would be recognizable to its members if described comprehensively, and since

secrets within families from other family members are common, reporting on family dynamics is potentially dangerous to family relationships. We are currently developing strategies for dealing with these challenges.

Extending our sampling frame. Our collaborative ties to health care settings and the involvement of clinical geneticists as consultants on this project have made it clear that the cultural strains in the application of genetic knowledge to the clinic setting are even more pressing than we had anticipated. The influx of immigrants from Southeast Asia has increased the clinical significance of cultural differences. While thalassemia (Cooley's anemia) is most commonly associated with Southern Europeans (Greeks and Italians), in California they are increasingly associated with Southeast Asians (15.9/100,000 for beta thalassemia major and 31.8/1000 for E-beta thalassemia in California's Southeast Asian population). As with cystic fibrosis and sickle cell disease, the thalassemias are also autosomal recessive genetic disorders, transmitted in a similar autosomal recessive pattern and resulting in serious illness. Thus, in order to highlight more sharply the theoretical issues related to culture and genetic testing we have begun to conduct some fieldwork and interviews related to the thalassemias. This extension of our sampling procedures is consistent with the methodological approach of "grounded theory" in our original proposal. Inclusion of families from Vietnam, Laos and Cambodia will increase our power to illuminate the central issues of how cultural values and behaviors affect receptivity toward genetic testing and will assist us greatly in formulating feasible strategies to counsel families of various cultural backgrounds.

Members of our group have presented preliminary findings and papers related to this project at a number of professional meetings during this current funding period, including the following:

Pacific Sociological Association (Beeson and Doksum)

"Emotions and Genetic Testing" April, 1994

Society for the Study of Social Problems (Duster and Beeson)

"Molecular Genetics Meets the High-risk Family: Close Encounters of Divergent Worlds" August, 1994

National Sickle Cell Disease Program (Yamashita, et al)

"Social and Cultural Factors in Sickle Cell Disease" March, 1994

International Bioethics Institute Fourth Annual Congress (Duster)

"Individual Autonomy versus Collective Interests in Genetic Information." April 1994.

The following publications by members of the research team have been published recently:

Duster, Troy. "Human Genetics, Evolutionary Theory, and Social Stratification," in Albert H. Teich and Mark S. Frankel, eds., *The Genetic Frontier: Ethics, Law and Policy*, American Association for the Advancement of Science Press, 1994.

Duster, Troy. "Depistage Genetique et Resurgence de L'Eugenisme," *Review Quadrerni*, Paris, France, Fall, 1993.

Beeson, Diane. "Social and Ethical Issues in the Prenatal Diagnosis of Fetal Disorders" in *Health Care Ethics: Critical Issues*, ed. John F. Monagle and David C. Thomasma, Aspen Publishers, 1994.

PRELIMINARY FINDINGS

We have continued to expand and elaborate our understanding of family dynamics related to the acquisition, transmittal and use of genetic knowledge. Patterns previously identified (see previous progress report) such as gender differences, generational differences and class differences are being elaborated.

A major focus of our analytical sessions is distinguishing class issues from ethno-cultural issues. This is particularly complex in the sickle cell families.

A key theme that is emerging from our data is the shared cultural values that cross-cut ethnic groups, but contrast sharply with the shared meanings and values of the world of molecular genetics. Increasingly we see evidence that the world of clinical medicine serves as a cultural borderland mediating between the social worlds of science on the one hand, and family life on the other. This means that while the clinician must be concerned with cultural differences between families of various ethnic groups, policy makers should take note of some of the sharp contrasts between the social world of molecular genetics and the social worlds of family life in general. The central issue here is the contrast between the probe to the molecular level that eventually strips away other contexts (with the corresponding hope that this information will enhance the health and the lives of those who receive it), and the vigorous insistence on the re-contextualization of decontextualized genetic diagnosis. It is the importance of the latter process, and the difficulty of carrying it out under many social conditions, that often results in a devaluation of, or even complete disregard for genetic knowledge in families we have studied. The implications of this tension for medical practice, health-seeking behavior, intra-familial communication and health policy will be drawn out in our final year of work.

SUMMARY OF WORK TO BE PERFORMED DURING NEXT TWO YEARS

During the final year of the project we plan to continue interviewing family members with emphasis on moving more deeply into the sickle cell family networks. Our cases increasingly will be selected on theoretical grounds rather than simply to build our numbers of respondents. We have found that obtaining access to each cultural group requires developing different strategies. Unlike

either our African-American or European -American respondents, our Asian respondents appear less willing to meet our interviewers outside the clinic setting. They are also much more circumspect in how they talk about their problems. Candid discussion requires multiple contact in the clinic setting. We will commit an increasing number of hours in the field to establishing contact with these subjects.

We plan to carry out our original plan to convene focus groups to elaborate and evaluate key findings of our work. These groups will be culturally specific, containing six to ten respondents. They will be selected in order to achieve stratified homogeneity around key social dimensions we have determined to be relevant to responses. This method, involving reinforcement of the perspectives associated with specific social location, should facilitate clearer articulation of non-dominant perspectives on sensitive issues.

We will conclude our computer-assisted narrative analysis and continue to prepare articles and books reporting our findings for publication.