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Genetics, Mental Illness, and Complex Disease: Development and Distribution of an Interactive CD-ROM for Genetic Counselors

Final Report
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NOTICE

ACCOMPLISHMENTS OF THE PROJECT

Overview

With funding from the US Department of Energy, NCHPEG has developed a CD-ROM titled, "Genetics and Major Psychiatric Disorders: A Program for Genetic Counselors." The program was released in September 2002 and was provided free-of-charge to members of NCHPEG, the National Society of Genetic Counselors, the Canadian Association of Genetic Counselors, the International Society of Nurses in Genetics, and all accredited genetic counseling training programs in the US and abroad. In addition, copies of the CD-ROM were made available at the meetings of the American Society of Human Genetics and the American College of Medical Genetics, to the Web Based Genetics Institute in Cincinnati, and to participants in the Genetics Interdisciplinary Faculty Program (Duke University) and the NINR Summer Nurses Institute. Dozens of CD-ROMs have been mailed to non-genetics health care professionals who requested copies. Total distribution as of March 2003 is approximately 4,500.

Project Objectives

The specific objective of this project was to provide genetic counselors with an educational program that will improve their understanding of the following topics:

- description, incidence, and natural history of schizophrenia (SZ), schizoaffective disorders (SZA), and bipolar disorder (BPD);
- current approaches to and limitations of diagnosis and treatment, and the implications for genetic counseling;
- current explanations for the causes of the disorders;
- the status of research into genetic contributions to SZ, SZA, BPD, including a review of research methodologies;
- current approaches to genetic counseling for SZ, SZA, and BPD, the implications for genetic counseling of potential gene discovery and genetic testing, and considerations for the development of standardized counseling protocols;
- the potential involvement of primary-care practitioners and mental health professionals in providing referrals for psychiatric genetic services, and the role of genetic counselors in educating those practitioners;
- ethical, legal, and social issues that arise from continued research into the genetic bases of SZ, SZA, and BPD; and,
- the ways in which these adult psychiatric disorders demonstrate the growing importance of common, complex diseases in genetic medicine.

The educational program we developed meets the above-mentioned objectives, with the following exceptions:

- Rather than focus on schizophrenia, schizoaffective disorder, and bipolar disorder, the advisory and writing committees suggested that we review unipolar major depression, schizophrenia, and bipolar disorder. This suggestion was based largely on the significant prevalence of unipolar major depression in relation to the relative rarity of schizoaffective disorder. Some information about schizoaffective disorder is provided in the overviews of schizophrenia and bipolar disorder.

- Because the status of genetic research in psychiatric disorders is changing rapidly, we chose to provide an overview of findings for each disorder and focus more heavily on a review of research methods so that genetic counselors will be better able to evaluate new reports.
- We did not focus on the development of standardized counseling protocols. While much of the content in the genetic counseling section would be applicable to those developing counseling protocols, we felt that there was not enough data about the needs of psychiatric consultands to allow us to address standardized protocols specifically.

We followed the developmental process outlined in the grant, which entails using the expertise of an advisory committee to guide product development, a review of the advisory committee's recommendations by expert evaluators, a writing committee to develop the majority of the content, a field-test of an alpha version (including target audience and expert evaluators) for formative evaluation, a reconvening of the advisory committee to suggest changes based on the field test, and a reconvening of the writing committee to implement those changes. As proposed in the grant, we used the framework for genetics and complex disease developed by Barton Childs and Joseph McInerney to guide development of the section on complex genetics.

We chose to add several steps to the process outlined in the grant document. First, we surveyed listserv members of the National Society of Genetic Counselors to gain more insight about their needs, and used the results to help guide development. (These data are reported in the RIMS report of May 2002.) We also chose to convene two groups of experts, one at the 2001 meeting of the American Society of Human Genetics and one at the 2001 meeting of the National Society of Genetic Counselors, to discuss risk assessment in complex disorders. The discussions at those meetings helped guide the development of the risk assessment materials in the common disease and genetic counseling modules.

At the end of the initial grant period we requested a no-cost extension that allowed us to put the CD-ROM materials on our website. We chose this route in part because non-genetics health care professionals have expressed interest in the product and our limited supply of CD-ROMs will soon expire. Having this material on our website will allow us to offer the materials for many years to come, and to update materials to reflect major changes in the field.

Based on the total grant award and the number of CDs produced we are able to quantify the cost per CD-ROM, which is approximately $135 per copy. These calculations do not take into account that all materials are available on the internet for the use of an untold numbers of individuals.

UNEXPENDED FUNDS

We are returning a small portion of our total funds because we did not incur all of the indirect costs that we expected. Details are found on form SF 269A.

FEEDBACK

Because we have not yet completed an evaluation of the product, we do not have quantitative feedback. We have received many positive comments about the CD-ROM. To date the only negative comments we received were from individuals who were unable to load the CD-ROM onto their machines.

Following is a selection of comments about the CD-ROM and on-line materials:

> "I was delighted to see the program you have developed. Thank you for your work on this project."
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> "I took some time this weekend to review the CD on Psych Disorders that you distributed to NSGC's membership, and I just want you to know how incredibly impressed I was. The quality of the production and content is superb. The professional presentation will, I would hope, serve as the first in a long series of others! Thank you for this fine (and first!) education about psychiatric genetic counseling to our community. I trust you are as proud of your work as it will be beneficial to its audiences."
>
> "I received a copy of the CDRom. You did a marvelous job! Congratulations!!!! The balance between facts, application of principles and case examples was fantastic."
>
> "I received a copy of your CD and have just had a chance to review it. WOW! It is terrific, user friendly and contains new information. Many thanks."
>
> "What a great site! Thanks for such a great job!"

In the next few months we hope to complete a quantitative analysis of the product. Information gained from the analysis should assist us as we develop future educational products for health care professionals.

ABSTRACT

"Genetics and Major Psychiatric Disorders: A Program for Genetic Counselors" provides an introduction to psychiatric genetics, with a focus on the genetics of common complex disease, for genetics professionals. The program is available as a CD-ROM and an on-line educational resource. The on-line version requires a direct internet connection.

Each educational module begins with an interactive case study that raises significant issues addressed in each module. In addition, case studies provided throughout the educational materials support teaching of major concepts. Incorporated throughout the content are expert video clips, video clips from individuals affected by psychiatric illness, and optional "learn more" materials that offer greater depth about a particular topic.

The structure of the CD-ROM permits self-navigation, but we have suggested a sequence that allows materials to build upon each other. At any point in the materials, users may pause and look up terms in the glossary or review the DSM-IV criteria for selected psychiatric disorders. A detailed site map is available for those who choose to self-navigate through the content.

The program begins with a module on complex disease. Major teaching concepts are introduced in a case study that focuses on Alzheimer's disease. The module reviews the tenets of complex disorders, discusses the similarities and differences between complex

and Mendelian disorders, reviews the importance of age-at-onset, discusses the threshold concept, and challenges the traditional method of risk assessment in common disorders.

The second module is an overview of psychiatry. The case study for this module shows a question from an individual at risk for bipolar disorder, and the response given by a mental health professional, on a health-related website. The professional's response, which includes several inaccurate statements, is critiqued. The module goes on to provide an introduction to psychiatry, which is necessary because genetics professionals are, in general, not well-educated about the field. Major topics include psychiatry as a field of medicine, the etiology of psychiatric disorders, review of psychiatric terminology, methods of psychiatric diagnosis, treatments for psychiatric disorders, and the public health implications of psychiatric disorders.

Research in psychiatric genetics is the focus of the third module. The case study considers a fictional email from a participant in a research study on psychiatric genetics; the module goes on to answer the questions posed by the participant. As a framework, this module uses a flowchart of the sequence of events that generally occur in psychiatric genetics research. Users can learn more about topics such as establishing the genetic basis of the disorder, defining the sample, data analysis, identification of a region likely to harbor a susceptibility gene, identification of candidate genes, mutation detection, and confirmation of a susceptibility gene. Each heading includes examples from recent psychiatric research.

The materials continue with introductions to unipolar major depression, bipolar disorder, and schizophrenia. Case studies for each section present fictional, but realistic, accounts of course of illness and impact on the lives of the characters. For each disorder we provide a clinical description and information about course of illness, prevalence, comorbidity, etiology, genetic studies, environmental factors, and treatments. Each section includes a video clip of an affected individual discussing his or her experiences with the disorder in question.

The penultimate module explores issues in psychiatric genetic counseling. The case study introduces a fictional consultand who is seeking genetic counseling because of her family history of schizophrenia; the case study continues throughout this section. The module on genetic counseling instruct users how to gather information (including reviewing medical records and taking a psychiatric family history), risk assessment (including the use of empiric risks, individualizing risk assessment, evaluating multiplex families, and providing risks to clients), counseling concerns of consultands (including consultands who are affected or are close relatives of an affected individual, consultands who are adopting, and consultands who are seeking genetic services for another indication with a family or personal psychiatric history that arises during the medical/family history), and managing uncertainty.

The final module begins with a fictional case study that emphasizes both the potential and the undeniable limitations of future knowledge about the etiology of psychiatric disorders. In this case study, results from a clinical evaluation and genetic testing allow

the patient to be categorized into narrow diagnostic categories resulting in an improved understanding of natural history and medication response. In addition, an at-risk relative is able to learn more about her risk through genetic testing. The future module includes predictions from experts in genetics, psychiatry, and research. In this module we discuss potential improvements in psychiatry, psychiatric genetics, and psychiatric epidemiology; ethical issues related to these potential improvements; and expectations that are unrealistic given the complexity of psychiatric disorders.

Supporting materials for the product are available on the NCHPEG website. These materials include counseling aids (including empiric risk data, age-at-onset curves, teratogen information, and ELSI content), information on genetic disorders with psychiatric phenotypes, research updates, and links to outside resources.