

Theses Summaries

1999

1) What Happens During the Prenatal Genetic Counseling Session: Exploratory Study of Genetic Counseling

Yelena Kemel, Advisor-Debra Roter, DrPH, MPH, 1999. Oral presentation NSGC 1999.

Abstract

To gain better understanding of communication between genetic counselors and patients, we conducted an exploratory study of the communication process in genetic counseling encounters in the context of prenatal diagnosis. This study examined the content, structure and patient satisfaction of prenatal diagnostic consultations of patients referred to genetic counseling centers following an abnormal triple screen result. Data were collected at two prenatal diagnostic centers in the Baltimore/Washington area. Ten genetic counseling sessions conducted by five genetic counselors from these two prenatal diagnostic centers were audiotaped, transcribed, and analyzed for content using the Roter Interaction Analysis System and qualitative content analysis. Patient satisfaction was measured by an exit questionnaire. The consultations were dominated by biomedical information exchanges. The amount and nature of information provided by the counselors was very similar between different sessions and consistent with published report on the content of prenatal genetic counseling sessions. Some of the areas where the most variability was observed included decision-making discussions, psychosocial discussions, and discussions of the outcomes of testing. The relevance of these findings for clinical practice and training of genetic counselors, as well as directions for future research are discussed.

2000

2) Perceptions of Reproductive Risk and Carrier Testing among Adolescent Sisters and Parents of individuals with Chronic Granulomatous Disease

Cynthia James, Advisor-Neil Holtzman, MD, 1999. James CA, Holtzman NA, Hadley DW. Perceptions of reproductive risk and carrier testing among adolescent sisters of males with chronic granulomatous disease. *Am J Med Genet.* 2003 May 15;119C(1):60-9.
http://www.ncbi.nlm.nih.gov/pubmed/12704639?ordinalpos=2&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum ; Oral presentation. NSGC 2000.

Abstract

Although substantial research has investigated the impact of having a sibling with a chronic disease, little is known about how genetic etiology influences an adolescent sibling's experience, impeding the provision of effective genetic counseling. To explore adolescent sisters' (1) perceptions of their reproductive risks, (2) resources for information and support, and (3) attitudes toward the appropriate age for, and risks and benefits of, carrier testing, we interviewed 14 parents and 9 sisters (ages 12-15) of males with Chronic Granulomatous Disease (CGD), a primary immunodeficiency disorder inherited in both an X-linked recessive and autosomal recessive manner. The semi-structured telephone interviews were audiotaped, transcribed, and subjected to template analysis (a common qualitative methodology). Girls were all aware that CGD is a heritable condition and each had made an assessment of her reproductive risk. All of the girls

considered their parents to be their best source of information and support. However, girls sometimes had trouble initiating discussions for fear of upsetting their parents. Girls commonly favored carrier testing at a later age than their parents. Even the girls who favored carrier testing for minors expressed more concerns about psychological risks associated with testing than did their parents. 4/14 parents considered childhood carrier testing necessary, believing it is a parents' responsibility to obtain medical information and emotionally beneficial for a child to grow up with it. 9/14 parents advocated carrier testing during adolescence based on concerns about their child becoming sexually active. All girls and parents believed eventual carrier testing was vital because of its relevance to reproductive decision-making and relationship-building. This study provides insight into the experiences of families with X-linked and autosomal recessive CGD, which may have relevance for other X-linked and autosomal recessive conditions. We found that adolescent sisters think about their reproductive risks and develop opinions about carrier testing and want to discuss these issues with their parents, but have trouble initiating conversations. Therefore, including adolescent sisters in family-based genetic counseling would be beneficial. Further research is needed to determine effective ways to aid adolescent siblings in thinking about their reproductive risks and options for carrier testing and to facilitate communication within families about these issues.

3) Parental Attitudes Towards a Diagnosis in Children with Unidentified Multiple Congenital Anomaly Syndromes

Eric Rosenthal, PhD, Advisor-Barbara Biesecker, M.S., 1999. Rosenthal ET, Biesecker LG, Biesecker BB. Parental attitudes toward a diagnosis in children with unidentified multiple congenital anomaly syndromes. *Am J Med Genet.* 2001 Oct 1;103(2):106-14.; http://www.ncbi.nlm.nih.gov/pubmed/11568915?ordinalpos=3&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum Oral presentation NSGC. 1999.

Abstract

Approximately 1% of newborns are affected with multiple birth defects, and in over half of these cases it is impossible to identify a specific syndrome to explain the pattern of anomalies. Although genetics clinics provide care for many children with unexplained congenital anomaly syndromes, very little is known about the ways in which the lack of a diagnosis affects parents. We undertook an exploratory investigation of this phenomenon by interviewing twenty-nine parents of sixteen children born with an unidentified multiple congenital syndrome in order to learn more about their attitudes towards diagnostic information. The parents in our sample had all been aware of their child's birth defects for between 2 and 23 years, and they had all sought multiple evaluations in an attempt to find a diagnosis that could explain their child's condition. Since our sample was selected from a pool of parents who had already demonstrated an interest in diagnostic information it was not surprising that most of them told us that they remain interested in identifying their child's syndrome. Typically, they told us that they had been especially anxious to obtain a diagnosis when the child was younger, but their interest has diminished to varying degrees with the passage of time. Using qualitative methodology, we identified six general areas where parents felt that a diagnosis would have a significant impact: Labeling, Causes, Prognosis, Treatment, Acceptance, and Social Support. Some of the most significant issues revolved around obtaining special education services, knowing what to expect in the child's future, watching out for new problems, fears about the child's life-expectancy, concern about recurrence risks, and a desire

to make sure that the child was receiving appropriate treatment. We discuss these results in relation to clinical practice and suggest areas for further research.

2000

4) Experienced and Anticipated Stigma and Coping Responses in Children with Neurofibromatosis Type I and their Unaffected Siblings

Lisa Pike-Buchanan, Advisor-Barbara Bisesecker, M.S., 2000. Oral presentation NSGC 1999.

Abstract

No research studies have yet been completed that investigate the stigma experience and coping responses in children with neurofibromatosis type 1 (NF1) or their unaffected siblings. Information gained from this investigation will allow a counselor to anticipate concerns about, and potential effects of, stigma and to explore effective ways of coping with them for a particular child or unaffected sibling. It is particularly important to begin facilitating the coping process during childhood, given some of the enduring psychosocial detriments living with NF1 may present. Using a qualitative approach, we interviewed eleven children with NF1 and eleven unaffected siblings of children with NF1 between the ages of 7 and 17 years. Themes were identified within the six dimensions of stigma, including origin, aesthetic qualities, concealability, course, peril and disruptiveness. The two most significant dimensions contributing to the stigma experience of these children were concealability and disruptiveness. The themes that emerged from the concealability dimension were covering the physical stigmata, passive concealment of the diagnosis, and active concealment of the diagnosis through secrecy. Significant themes that materialized from the disruptiveness dimension were teasing, ability to form and maintain friendships, learning disabilities, diminished involvement in sports activities, and pain. Visibility of stigmata is a more meaningful predictor of the stigma experience than severity. Stigma, whether experienced or anticipated, was found to some degree in all of the affected participants. With one exception, the unaffected siblings did not experience or anticipate courtesy stigma. Themes were also recognized within the three categories of coping responses to a stigmatizing event, including those with attempt to: 1) prevent the event by changing the situation around it; 2) change the meaning of the event; and 2) control stress after the event has happened. The coping mechanism utilized most often by this study population involved an attempt to prevent the stigmatizing event from happening by changing the situation around it, which coincides with the concealability dimension of stigma. A strong need for social support from other children with NF1 was identified in the affected children. Implications for genetic counseling and areas for future research are discussed.

5) Genetic Testing for Breast Cancer Susceptibility: A Qualitative Assessment of Black Women's Attitudes

Grace-Ann Olayinka-Fasaye, Advisor-Barbara Bisesecker, M.S., 2000. Oral presentation NSGC 2000.

Abstract

Black women are less likely to undergo genetic testing for breast cancer susceptibility and are poorly represented in genetic counseling testing research. To understand this, qualitative methods were used to assess health beliefs and practices of twelve Black women with a family history of breast cancer. A semi-structured interview guide was used to elicit insights into how

health beliefs influence attitudes toward genetic testing for breast cancer risk. The women received regular breast cancer screening and believed breast cancer could be effectively treated. All of the women spontaneously referred to spirituality when describing their health beliefs. Most women were uncertain about the causes of breast cancer but felt that they could reduce their chances of getting breast cancer by taking care of themselves. None of the women would consider prophylactic mastectomy because of their belief that other effective methods exist for prevention. Most women acknowledged that breast cancer can be hereditary but did not feel that it was running in their family. Nine of the twelve women were hypothetically interested in genetic testing. Perceived advantages of testing included getting a complete understanding of their risk, reassurance and relief from uncertainty. Women believed a positive test result would encourage them to monitor their breasts more carefully, and motivate them to urge their relatives to monitor their breasts aggressively and to get genetic testing. Perceived disadvantages of testing included the inconvenience of having to make several trips for testing, dislike of needles, stress associated with a positive result and believing that the test offers no guarantees that breast cancer will not occur, no matter what the test result. Women would prefer to have their physician approach them about genetic testing and would like testing to be performed with other routine blood work or when they receive their mammograms. This research suggests reasons that Black women may see potential personal value of genetic testing. Measures need to be taken to ensure that Black women are aware of the availability of testing. Genetic counseling with Black women should be individualized to facilitate an exploration of the impact that their perceptions have on their decision to have or decline testing.

6) Analysis of Referrals for Prenatal Genetic Counseling: Recommendations for Delivery of Genetic Services and Implications for Genetic Counseling

Carrie Mastromarino, Advisor-Barbara Bernhardt, M.S., 2000. Bernhardt B, Mastromarino C, Roter D, Geller G, How do Obstetric Providers Discuss Referrals for Prenatal Genetic Counseling? *J Genet Couns* 2005 Apr;14(2):109-17.

[http://www.ncbi.nlm.nih.gov/pubmed/15959642?ordinalpos=206&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum] Oral presentation NSGC 2000.

Abstract

Physicians and other health care providers serve as gatekeepers for the majority of genetic counseling clients. Yet few studies have been conducted to assess if or why obstetric providers refer their patients to prenatal genetic counseling. Further, it is unknown what patients are told about genetic counseling by those who refer them. Previous research suggests genetic services are underutilized, and ambiguity exists among non-genetics providers about what constitutes genetic counseling. This study utilized secondary data to examine aspects of the referral made to prenatal genetic counseling. Audiotapes were available from the first prenatal visits of a total of 104 patients with forty-five providers (24 obstetricians and 21 certified nurse-midwives). Seventy-two visits were with actual patients and 32 visits were with a simulated patient. Thought all patients had indications for being offered genetic services, the simulated patient was considered at higher risk than were most actual patients. Content analysis was performed on transcripts of these visits to document the circumstances under which patients do and do not get referred as well as to provide information about what actually occurs during a discussion about genetic counseling. Finally, interviews with 23 of these obstetric providers were analyzed to help interpret what was observed in the prenatal visits. Findings indicated that most actual patients were

not referred to genetic counseling, while the simulated patient usually was referred to genetic counseling. Second, when genetic counseling was discussed, minimal description of the service was provided. Variance in how genetic counseling was discussed may be explained, in part, by different characteristics of the actual and simulated patients, such as degree of genetic risk and interest in prenatal diagnostic testing. In addition, data from interviews with providers revealed that providers refer less often than they say they refer, but that their perceptions of genetic counseling generally match how they discuss the service with patients. These data prompt specific recommendations for the obstetric community about referring patients for genetic counseling and what to tell patients about this service. IN addition, Implications of these findings for the field of genetic counseling are addressed.

7) Development of a Patient Activation Intervention and Assessment of the Feasibility of its Use in a Study of Prenatal Genetic Counseling Clients

Melissa Barber, Advisor-Barbara Bernhardt, M.S., 2000. Oral presentation NSGC. 2000.

Abstract

Genetic counseling represents the opportunity for clients to have their informational and psychosocial needs addressed by the counselor. Studies have indicated that genetic counseling sessions may not be addressing all of a client's psychosocial needs. The aims of the study were to develop and revise a patient activation intervention tool and to assess the feasibility of conducting future studies to evaluate it. A health education model was used to develop an intervention aimed at increasing client participation in genetic counseling by addressing factors known to effect participation. Qualitative data concerning the design and use of the intervention and the feasibility of assessing its effectiveness with genetic counseling clients were collected from genetic counseling clinic staff (n=14) and genetic counseling clients (n=16). The findings from this study led to a revised version of the patient activation intervention tool that takes into account most of the concerns and suggestions made by the research subjects. The intervention is a two-sided document that includes a brief introductory paragraph about prenatal genetic counseling, a chart for clients to indicate their questions and concerns, and a statement emphasizing the importance of their preparation in their genetic counseling session. We found that while genetic counselors were somewhat hesitant about exposing their clients to the intervention because it might lead to increased anxiety, the clients felt that having something like it prior to their counseling sessions might have helped them be better prepared for their session.

8) Discussions of Personal Meaning in Pre-Amniocentesis Genetic Counseling

Lori Hamby, Advisor-Debra Roter, DrPH., MPH, 2000. Oral presentation NSGC 2000.

Abstract

The information that women and couples learn during prenatal genetic counseling has relevance beyond the information itself. People process such information in terms of the personal meaning that it has for them. In order to learn more about the way discussions of personal meaning unfold during prenatal genetic counseling sessions and the factors that effect their inclusion, I conducted an exploratory qualitative study of the effect of the beliefs of prenatal genetic counselors on their counseling practices and the fulfillment of client needs. The study involved interviews with seven prenatal genetic counselors from four prenatal genetics centers about their general approaches within pre-amniocentesis genetic counseling sessions with these prenatal genetic counselors and their clients, and eleven interviews with a sub-set of the participating

clients. The combined analysis of these three sets of data provide preliminary evidence that there is discrepancy between genetic counselors' plans and practices as well as clients' desires and needs. The audio-taped sessions show variation in terms of the frequency and depth of such discussions in actual practice. In addition, the clients in this study expressed mixed feelings concerning their preferences for discussing personal meaning with their genetic counselors. The implications for the practice of genetic counseling, the training of genetic counselors and needs for future research are discussed.

2001

9) Why do Fertile Couples Choose Preimplantation Genetic Diagnosis: An Exploratory Study

Jennifer Leib, Advisor-Barbara Biesecker, M.S., 2001. Poster presentation NSGC 2001.

Abstract

In preimplantation genetic diagnosis (PGD), a couple uses in-vitro fertilization (IVF) to create embryos, and then one cell is removed and genetically tested. Unaffected embryos are then transferred to the woman's uterus. The possibility of misdiagnosing the embryo, the medical risks for the women, the low pregnancy rates and the financial cost of participating in PGD would all seem to deter a fertile couple who would not ordinarily participate in IVF from choosing PGD. Fertile couples may even be further deterred in that they are making the decision to engage in PGD in the presence of less risky, less expensive and more successful options, namely prenatal diagnosis, donor gametes and adoption. This study investigated the decision-making process of fertile couples who have had PGD, including the factors, beliefs and experiences that lead a couple to choose PGD over other options. An exploratory approach of qualitative research methods was used to analyze the data collected through semi-structured telephone interviews. The decision-making process to engage in PGD was found to be minimal. When specifically probed about other options, they described choosing PGD over prenatal diagnosis because of their discomfort with abortion. The majority of the sample reached this decision without any prior experience with prenatal diagnosis and a pregnancy termination. They chose PGD over the options of donor gametes and adoption based on their strong desire to be genetically related to their children. The impact of the genetic condition on the family and on the affected individual's life also served as a strong motivator to pursue PGD. An additional factor in their decision to participate in PGD was found to be the accessibility of PGD. Thus, when their attitudes and beliefs eliminated other reproductive options, the impact of the condition further motivated these fertile couples to gain access to PGD. This study generated several implications for genetic counselors, including the need to increase access to this new technology for their clients, the development of educational and supportive materials on PGD and the need to incorporate the impact of the genetic condition on the family in their counseling agenda.

10) The Decision to Continue: The Experiences and Needs of Parents who Receive a Prenatal Diagnosis of Holoprosencephaly

Krista Redlinger-Grosse, Advisor-Barbara Biesecker, M.S., 2001. Redlinger-Grosse K, Bernhardt BA, Berg K, Muenke M, Biesecker BB. The decision to continue: the experiences and needs of parents who receive a prenatal diagnosis of holoprosencephaly. *Am J Med Genet.* 2002 Nov 1;112(4):369-78.

<http://www.ncbi.nlm.nih.gov/pubmed/12376939?ordinalpos=2&itool=EntrezSystem2.PEntrez.Pub>

[med.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum](#)]. Oral Presentation NSGC 2001.

Abstract

Holoprosencephaly (HPE) is a condition characterized by a defect in the midline of the embryonic forebrain. When found prenatally, the diagnosis of HPE offers parents a poor but often uncertain prognosis for their baby. Previous studies have examined the psychological and social impact of abnormal prenatal diagnostic on parents. Because the majority of parents receiving abnormal prenatal results terminate their pregnancies, studies have yet to examine parents' experiences and needs throughout their decision to continue a pregnancy. This study explored this issue by interviewing twenty-four parents who received a prenatal diagnosis of HPE and continued their pregnancy. Parents were asked about the process of their decision making to continue the pregnancy and their needs throughout that process. The interviews were analyzed qualitatively for common themes that emerged from the parents' experiences. It was found that most parents did not make an active decision about continuing the pregnancy. Rather, parents described a more subtle, passive decision that evolved over time and consisted of several factors. These factors included the parents' religious background, personal beliefs, past experiences, and the uncertainty involved in the diagnosis of HPE. Parents also discussed many coping strategies that factored into their decision. In addition, they described their informational, emotional, and supportive needs throughout the process of decision making. Specifically, these needs stemmed from experiences with health care professionals that were both not helpful and helpful. These factors all contributed to the evolution of parents' decision to continue the pregnancy and their eventual adaptation and acceptance of their decision. The clinical and research implications of these results for the field of genetic counseling are discussed.

11) Stigma, Culture and the Genetics of Schizophrenia: The Family Member's Perspective

Robin Cohen, Advisor-Gail Geller, Sc.D., 2001. Oral presentations at the American Public Health Association's Annual Conference and at the 2001 NSGC AEC.

Abstract

The purpose of this study was to identify and describe family members' perceptions of schizophrenia in a climate in which there is increasing evidence for a genetic contribution to the etiology of this condition. This study addresses three primary research questions: (1) What do family members understand about and expect from genetic research related to schizophrenia? (2) How do family members think the discovery of a gene related to schizophrenia will influence the nature and degree of stigma associated with this illness? (3) Are responses to genetic research influenced by cultural factors? The power of molecular genetic tools is accelerating the process of understanding schizophrenia at a biological level. Despite the fascinating contribution these findings may be making to science, it is critical to understand their impact on families with schizophrenia since they will be the direct recipients of scientific knowledge and clinical interventions that may evolve from this work. It is essential that scientists and families work together to ensure that emerging genetic information is understood, disseminated, and utilized in a manner that best meets families' needs. This can be facilitated by including family members in empirical research about their expectations and perceptions of the role of genetics in schizophrenia.

2002

12) Spirituality and its Inclusion in U.S. Genetic Counseling Practice

Jessica Sparrin, Advisor-Gail Geller, Sc.D., 2002. Oral presentation NSGC 2002.

Abstract

For the vast majority of the general population, spirituality plays a prominent role in decision-making, coping and adaptation. Genetic counseling often brings individuals or couples into a situation where their religious and spiritual beliefs come to the forefront. Whether the reason for referral is genetic testing, prenatal diagnosis or the continuing care of an affected individual, by the very nature of the topics discussed, clients will often examine their personal morals and values. It is during this exploration that genetic counselors may encounter individuals' spiritual beliefs or religious convictions. However, very little is known about how genetic counselors respond both professionally and personally to this topic when it arises, or the degree to which counselors initiate such discussions. In order to fill this gap, a descriptive survey was sent to 1075 genetic counselors practicing in the U.S. 45% (N=477) responded. Although 18% of respondents say they "frequently" inquire about their clients' religious beliefs, 57% (N=234) of those who inquire do so to discuss the increased genetic risks associated with Ashkenazi Jewish descent. Four percent of respondents say they "frequently" discuss spirituality and 2.8% say they "frequently" inquire about the spiritual beliefs of their clients. Most commonly, such discussions focus on the clients "sources of support" and methods of "coping," (68.3% and 60.2%, respectively). Belief about "spiritual care" as the role of a genetic counselor ($p < 0.01$) and comfort level with the topic ($p < 0.01$) are the best predictors of a counselor's willingness to inquire about the spiritual beliefs of their clients. Respondents also provide their recommendations for future training needs including an overview of belief systems and techniques for initiating discussion about spirituality and religion.

13) Genetics and Alcoholism: Beliefs and Behaviors of At-Risk Relatives

Jennifer Gamm, Advisor-Barbara Biesecker, M.S., 2002. Gamm J, Nussbaum R, Biesecker B: Genetics and Alcoholism Among At-Risk Relatives I: Perceptions of Cause, Risk, and Control *Am J Med Genet* 128A(2):144-150, 2004.

http://www.ncbi.nlm.nih.gov/pubmed/15214005?ordinalpos=2&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum . Gamm J,

Nussbaum R, Biesecker B: Genetics and Alcoholism Among At-Risk Relatives II: Interest and Concerns about Hypothetical Genetic Testing for Alcoholism Risk. *Am J Med Genet* 128A(2):151-155, 2004.

http://www.ncbi.nlm.nih.gov/pubmed/15214006?ordinalpos=1&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum Oral presentation NSGC 2002.

Abstract

There is evidence that genetic factors can influence a person's risk for developing alcoholism. Yet, the extent to which people recognize the existence of these factors and integrate this information into their own belief systems is unknown. In other disorders with a hereditary component, there is evidence that a belief in a genetic etiology may lower an individual's perceived personal control or lead to a fatalistic outlook that may adversely affect behavior. The purpose of this study was to explore the beliefs concerning etiology of alcoholism, specifically beliefs about the role of genetics, among individuals whose families have been affected by alcoholism. Additionally,

we (I) explored the relationships between these beliefs and individuals' feelings of personal risk and perceived control related to alcoholism, and we (did you decide to revert to we?) investigated the level of interest in possible future predispositional genetic testing. In-depth interviews were conducted with 27 individuals who had at least one first-degree relative affected with alcoholism. The interviews revealed that participants attribute multiple factors to the cause of alcoholism in their families, and that these factors are combinations of biological, genetic and environmental influences, as well as personal characteristics. The perception of being at-risk most often stemmed from a belief in a genetic or biological etiology for alcoholism, while participants' strong sense of personal control was associated with beliefs in environmental or personal characteristics that differed between them and their alcoholic family member. Among the interviewees, interest in genetic testing for susceptibility to alcoholism was moderate, and anticipated reactions to testing did not support the concept that fatalism is at work in this population. The various beliefs that individuals hold concerning the etiology of alcoholism, their feelings about risk, and their sense of perceived control are all important issues for patients behaviorally and psychosocially. Professionals should be aware of these beliefs and the possible relationships among them.

14) Language Interpreters in Genetic Counseling

Jane Peredo, Advisor-Barbara Bernhardt, M.S., 2002. Oral presentation NSGC 2002.

Abstract

Increasingly, in the United States, persons with limited or no English proficiency are seeking medical care, including genetic counseling services. Thus, a language barrier is likely to exist when this population seeks genetic counseling services. Given its client-centered approach with an emphasis on patient autonomy about decision-making regarding genetics and the impact of that information on one's life, communication is integral to genetic counseling. This study addresses the extent to which language interpreters are needed and utilized in genetic counseling, and genetic counselors' satisfaction with interpreters. Genetic counselors (N=711) working in states where persons with limited English proficiency are presumed to reside were asked to complete a questionnaire about the need and utilization of interpreters in this setting. In addition, factors that influence decision-making about requesting a trained interpreter were explored. This study found that persons with limited or no English proficiency comprise, on average, 21.3% and 12.7%, respectively, of clients seeking genetic counseling services. When encountering a language barrier, genetic counselors use *ad hoc* interpreters twice as often as trained interpreters. However, use of trained interpreters in genetic counseling is higher than previously reported in the medical setting. Although trained interpreters are available to approximately 60% of genetic counselors, their ability to speak another language, the convenience of recruiting an *ad hoc* interpreter, the administrative burden of locating a trained interpreter, cost, having one's request bumped by another clinician and satisfaction with *ad hoc* interpreters were identified as barriers to the utilization of trained interpreters.

15) Newborn Screening for Duchenne Muscular Dystrophy

Nicole Johnson, Advisor-Neil Holtzman, MD, 2002. Oral presentation NSGC 2002.

Abstract

Duchenne muscular dystrophy (DMD) is an X-linked genetic disorder that often results in death within the third decade of life. DMD is a relatively common disorder affecting approximately 1:3500 live born males. There is currently no cure for DMD and medical intervention primarily

focuses on supportive care. Technology was developed in the mid-1970's that would allow the identification of newborns who would eventually develop the symptoms associated with DMD. Between the years of 1985 and 1997, 58 males were identified with muscular dystrophy through a Western Pennsylvania pilot newborn screening project. The experience of the parents of the children identified, and how the presymptomatic information affected their families, was explored with parents from this pilot screening program. Seventeen parents, from twelve families, completed a semi-structured interview. These seventeen parents, along with two who did not participate in the interview, completed a questionnaire that assessed fifteen areas of family functioning such as overprotection, integration into the family and limits on family opportunity. The interviews were analyzed for common themes that emerged from the parents' experiences. Parents perceived the presymptomatic diagnosis as beneficial for: helping to emotionally prepare themselves and their family prior to their son's own awareness of his disorder, time to prepare their house for a handicapped child, and for planning reproductive decisions. Seven out of the twelve families stated that they changes their original reproductive plans based on knowing their son's diagnosis. Negative aspects of screening centered around the diagnostic process in which parents first learned of their son's potential diagnosis of muscular dystrophy. Some parents experienced feelings of detachment or interference with bonding as a result of learning their child's diagnosis during the neonatal period. Scores on the family functioning questionnaire indicate that families of ambulatory children diagnosed through neonatal screening tend to have a level of stress more similar to families of non-ambulatory children than families of ambulatory children that received a clinical diagnosis. While all families believed that learning their son's diagnosis prior to a later clinical diagnosis was beneficial, some of the families believed that the neonatal period was too soon for an early diagnosis. In the absence of pre-symptomatic clinical trials for treatment of muscular dystrophy, heightened awareness about the early signs and symptoms of muscular dystrophy for pediatricians and the general public may be a better alternative to addressing the problem of diagnostic delay than neonatal screening for muscular dystrophy. Prior to implementation of potential future pilot neonatal screening programs, particularly for pre-symptomatic and untreatable conditions, protocols should be developed that create the best possible approach for pediatricians to disclose results to parents. Genetic counselors familiar with the condition should be involved with the initial disclosure and available to families on both an immediate and long-term basis after they receive their child's screening results. In addition, pilot neonatal screening programs should remain optional and include an evaluative component to monitor and assess parents' experiences so that programs can be modified and/or discontinued in response to their experiences.

2003

16) Genetic Counselor's Descriptions of Down Syndrome: Promoting Informed Choice for Prenatal Testing

Elizabeth Lardy, Advisor-Barbara Biesecker, M.S., 2003. Oral presentation NSGC 2003.

Abstract

Prenatal diagnosis is offered to pregnant women 35 and older because of their increased risk of having a child with Down syndrome or another chromosomal anomaly. Because there are no standard guidelines as to how Down syndrome should be described in a prenatal genetic counseling session, there may be tremendous variation in counselor's individual practice.

Members of the disability and genetic counseling communities have expressed concern that couples are not given a balanced and accurate portrayal of the nature of Down syndrome before prenatal diagnosis, and thus clients are not able to make informed choices about prenatal testing. The purpose of this study was to gain a better understanding of the ways in which Down syndrome is described in prenatal genetic counseling sessions and what factors might influence the description. A qualitative research design was employed to better understand how Down syndrome is described, to assess the type of personal experience prenatal genetic counselors have with people with developmental disabilities, and to describe counselors' attitudes toward Down syndrome. One hundred and sixteen prenatal genetic counselors participated in 15-minute phone interviews. The results suggested that the majority of counselors use a medical description of Down syndrome, most often mentioning mental retardation, facial features, and heart defects. Few counselors mentioned the capabilities of people with Down syndrome, and even fewer mentioned the positive aspects of Down syndrome. Most counselors appreciate both positive and negative aspects of Down syndrome, and believe that people with Down syndrome can have friendships, hold jobs, and go to school, but few point out these abilities in their prenatal descriptions of Down syndrome. This study facilitates understanding of what is said about Down syndrome in prenatal genetic counseling sessions and what factors might influence how Down syndrome is described. It brings into question the information presented to clients for making an informed choice about prenatal testing and has several implications for genetic counseling practice and further research.

17) Concerns About Genetic Discrimination: Origins and Effects

Kira Apse, Advisor-Barbara Bernhardt, M.S., 2003. Apse K, Biesecker B, Giardiello F, Fuller B, Bernhardt B: Perceptions of Genetic Discrimination Among At-Risk Relatives of Colorectal Cancer Patients *Genet Med* 2004 Nov-Dec;6(6):510-6.

http://www.ncbi.nlm.nih.gov/pubmed/15545747?ordinalpos=1&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum Oral presentation NSGC 2003.

Abstract

In the midst of extraordinary advances in genetic research, some consumers are afraid to utilize genetic services because of concerns about genetic discrimination. Several public opinion polls and research studies suggest that concerns about the misuse of genetic information by employers and insurers may be widespread. This study explores the nature and extent of consumers' concerns about genetic discrimination as well as the source of such concerns and the resultant impact on utilization of genetic services. A questionnaire was sent to unaffected individuals with a family history of colorectal cancer who had enrolled in the Hereditary Colorectal Cancer Registry of the Johns Hopkins Hospital (N=777). Of the 470 respondents, approximately half rated their level of concern about genetic discrimination as high. Few respondents, 12%, had participated in genetic counseling and 8% had undergone genetic testing. The majority, 68%, had heard of genetic discrimination before receiving the questionnaire of which 79% learned about it from at least one media source (television, newspapers, magazines and radio). Awareness of legislation regarding genetic discrimination was found to be minimal. Specifically, 88% of respondents were not aware of any federal legislation protecting against genetic discrimination, and 96% did not know if their home state had pertinent legislation. Respondents with a higher level of concern about genetic discrimination were significantly less likely to consider meeting with a healthcare professional to discuss genetic testing or to undergo testing. In addition, if they were to pursue genetic testing, those with a higher level of concern would be significantly more likely to pay

out of pocket, use an alias or ask for test results to be excluded from their medical record. Findings from this study demonstrate the negative effect of concerns about genetic discrimination on interest in and decisions about utilization of genetic services. Stronger legislative protections against genetic discrimination and increased public education through the scientific community and media sources are needed.

18) Asking Fathers: A Study of Psychosocial Adaptation Among Fathers Boys with Hemophilia

Erin Herrick, Advisor-Lawrence Wissow, Ph.D., 2003. Herrick EK, Nussbaum R, Holtzman NA, Wissow L. Asking fathers: a study of psychosocial adaptation. *Haemophilia*. 2004 Sep;10(5):582-9[PUBMED, PMID: 15357787] ; Oral presentation NSGC 2003, International Hemophilia Foundation Conference 2004.

Abstract

In the scope of research involving families with children who have chronic illnesses or special needs, paternal responses are generally lacking, and often combined with maternal responses to reflect overall parental functioning. While such data are useful in providing anticipatory guidance for couples, they tell us little about the unique needs of fathers. This study aimed to explore the psychosocial adaptation of fathers who have boys with hemophilia. A questionnaire was sent to fathers recruited from hemophilia support organizations in Maryland, Virginia, Pennsylvania and the D.C. area (N = 320). Quantitative measures included the Perceived Sense of Parenting Competence (PSOC), and the Locke-Wallace Marital Adjustment Test (MAT). Qualitative items addressed the challenges of rearing a son with hemophilia, barriers to father's involvement, and how their son's diagnosis has changed their life. Among the 83 eligible respondents, MAT and PSOC scores were significantly correlated ($p < .01$), indicating a relationship between adaptation in marriage and in parenting. Neither socio-demographic variables, nor medical characteristics of the boys were significant. Rather, psychosocial variables such as feeling "left out" of decision making regarding their son's care by their wife or by medical staff, worry about infection and limited activity, and not being able to give infusions significantly and negatively affected perceptions of parenting satisfaction, efficacy, and marital adjustment. Qualitative responses provided further support for quantitative findings. A more proactive attempt to include and engage fathers is warranted by all members of the healthcare team. Suggested educational interventions include more frequent discussions of inheritance and recurrence risks, and creating opportunities for hands-on practice giving infusions. Psychosocial interventions include: Asking fathers about and addressing their questions and concerns; providing anticipatory guidance on the importance of shared decision-making and paternal involvement in care, which may benefit marital adjustment, parenting sense of efficacy and satisfaction, as well as the child's adaptive outcome; and offering referrals to support groups and counseling services. As culture and medicine have advanced together, it is time to challenge some of the assumptions that have reinforced fathers' absence from the clinical setting and contributed to their minority status in social science research.

19) Japanese People in the U.S.: Their Needs and Attitudes toward Prenatal Genetic Counseling

Chieko Tamura, Advisor-Debra Roter, DrPH, MPH, 2003. Presented as a poster NSGC 2003.

Abstract

Genetic counseling, as practiced in the United States for over 30 years, has been based mainly on Western cultural values. Yet, the need for genetic counseling services in different socio-cultural settings has been increasing. This study was designed to understand the needs and attitudes toward prenatal genetic counseling of people with a Japanese cultural background. Three focus groups were held with Japanese women between 20 and 45 years of age, who had come to the US after turning 20 years old. Participants were asked about their needs and expectations toward prenatal general counseling, their social and familial support systems associated with their decision-making process, their thoughts and cultural views toward pregnancy and prenatal diagnosis, and their attitudes toward psychological counseling. Focus group data has been analyzed to identify themes related to needs and attitudes. The result shows that these women are interested in prenatal genetic counseling, and want to obtain as much information related to prenatal diagnosis as possible. Additionally, participants indicated that psychological counseling would be helpful for them. However, some suggested that they would like to know that genetic counseling sessions consist of not only information provision but also psychological counseling, prior to actual sessions, so that they could prepare to discuss medical issues as well as psychosocial concerns. Most of these women believe that the decision related to prenatal diagnosis should be made primarily by the couple. These women identify strongly with their Japanese cultural background, but do not want genetic counselors to treat them differently because of their background. The findings of this study will have implications for the delivery of prenatal diagnosis and genetic counseling services that take into account the needs of people who are living in the US and have a Japanese cultural background. The findings of this study will also be used to make suggestions for the future direction of prenatal genetic counseling practice in Japan.

2004

20) Prognostic Uncertainty and Personal Control: Influences on Parental Coping

Shawn Sinesky (Lipinski), Advisor-Barbara Biesecker, M.S., 2004. Lipinski SE, Lipinski MJ, Biesecker LJ, Biesecker BB. *Am J Med Genet C Semin Med Genet.* 2006 Nov 15;142C(4):232-40.

http://www.ncbi.nlm.nih.gov/pubmed/17068805?ordinalpos=2&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum Oral Presentation NSGC 2004 and as a poster presentation NSGC 2004.

Abstract

Research suggests that appraisals of a stressful event such as a health threat, including perceived uncertainty and personal control, are important predictors of the use of coping strategies. This study explored whether emotion-focused coping is employed more often than problem-focused coping when parents of a child with a chromosome abnormality perceive higher levels of uncertainty and lower levels of personal control over their child's condition. A survey was mailed to 875 parents of the Chromosome Deletion Outreach support group. Measures included the Parental Perception of Uncertainty Scale and the Ways of Coping Checklist-Revised. Perceived control, perceived benefit of the diagnosis and perceived severity of the condition were assessed using Likert-scales, along with collection of sociodemographic data. Open-ended items addressed the experiences of receiving a rare chromosome diagnosis for their child. Among the 363 respondents (41.5% response rate), the majority were female (95%), averaging 37 years of age. Parents waited

an average 1.7 years for a diagnosis. Parents' perceived level of uncertainty about their child's condition was greatest in the domains of Unpredictability and Ambiguity. Multivariate analysis revealed lower Perceived Personal Control, Ambiguity, and Lack of Clarity predicted the use of emotion-focused coping ($p < 0.01$) while use of problem-focused coping strategies was predicted by higher perceived personal control, parents' age, child's age, and perceived benefit of diagnosis ($p < 0.05$). A parent's perceived personal control and his/her perceptions of uncertainty do predict the use of coping strategies in adjusting to their child's condition. Genetic counseling interventions are proposed to enhance parental use of effective coping strategies given circumstances that entail a great deal of uncertainty about a child's future.

21) Exploration of Transitional Life Events in Individuals with Friedreich's Ataxia: Implications for Genetic Counseling

Valerie Brook Waggoner, Advisor-Barbara Biesecker, M.S., 2004. Presented as a poster NSGC 2004.

Abstract

Human development across the lifespan is a process of change, adaptation and growth. Throughout this process, key transitional events mark important points in time when one's life course is significantly altered. Awareness of transitional events, brought about or altered by chronic illness and disability, may assist health care providers in recognizing the changing needs of their patients over time. In this qualitative study, perceptions of adults with Friedreich's ataxia regarding transitional events are described. One-time, semi-structured interviews were used to elicit information regarding transitional events and data were analyzed to identify common themes. Adults between eighteen and sixty-five years of age were interviewed ($N=42$). Overall, Friedreich's ataxia increased the complexity and magnitude of transitional life events. Additionally, events commonly represented significant loss to the individual and presented challenges to his/her self-esteem and identity. Identified transitions were either a direct outcome of Friedreich's ataxia, or a developmental task altered by having the condition. Specifically, an awareness of symptoms, fear of falling and changes in mobility status were the most salient themes from the experience of living with Friedreich's ataxia. Developmental events influenced by the condition were one's primary relationships and life's work. Participants reported using both emotion and problem-based coping strategies during these times of change. These results, as well as data obtained from participants about experiences with health care providers, help to alert professionals of potentially challenging times in patients' lives, which are subtly or profoundly influenced by chronic illness or disability. Additionally, areas for further research are explored, and implications for developmental counseling approaches, which emphasize key transitional events, are suggested for genetic counseling.

22) Exploring the Value of Health Information to Adult Adoptees

Karen Tam, Advisor-Sara Hull, Ph.D, 2004. Oral presentation NSGC 2004.

Abstract

Adoption creates the legal relationship of parent and child between individuals who are not each other's biological parent or child. Unlike most individuals, many adoptees do not have access to personal and/or familial health information because of laws and practices that inhibit the disclosure of this information. Lack of access to information, coupled with the increasing consciousness of family history information in health care, brings to light unique challenges that may be associated with being an adoptee-patient. Because this issue has been insufficiently

addressed in the research literature, the primary goal of this study was to explore adult adoptees' perceptions regarding the value of personal and familial health information. Data were collected from 42 adopted individuals, ages 21 to 67 years, using semi-structured qualitative telephone interviews. Interviews were tape-recorded and transcribed, and transcripts were imported into QRS-NUD.IST, to facilitate coding and analysis. Data collected suggested several major themes related to: adoptees' motivations for seeking information (including health-related information) about their biological families; their thoughts on the perceived role and importance of familial health information; their experiences not having such information; and their recommendations for improving patient-provider interactions. The desire for health information - a common trigger and motivation leading adoptees' to search for their biological families - appeared to be related to developmental stages in the adult lifespan. Familial health information was seen as useful for a number of reasons, including: to assist in diagnosis, to help predict future health, to understand current symptoms, and as a basis for considering preventative measures. Adoptees described a range of negative feelings, including frustration and anger, when they did not have familial health information. Participants also thought that doctors often avoided the topic of adoption, and that their psychosocial concerns related to being adopted and/or having limited access to familial health information remained unaddressed in health care interactions. Participants raised suggestions to improve the health care of adoptees. Adoptees in this study demonstrated a practical understanding of the significance and limitations of having familial health information. Further, the experience of having limited access to familial health information plays a role in the complex relationships between searching for birth relatives and notions of kinship, identity, and health. Clinical implications, for primary care physicians and genetics specialists, are discussed in relation to caring for adoptees in the absence of familial health information, and facilitating adoptees' acquisition of it.

23) Adoption of the 2001 ACOG Guidelines for Cystic Fibrosis Testing by Obstetricians

Delphine Blain, MBA, Advisor-Ben Wilfond,MD, 2004. Oral presentation NSGC 2004.

Abstract

In 2001, 12 years after the gene for cystic fibrosis (CF) was discovered, the American College of Obstetrics and Gynecology (ACOG) published guidelines for CF carrier screening for the pregnant and non-pregnant patients of obstetricians. This foray of genetics into mainstream medicine came as research still showed knowledge deficits in genetics and CF among obstetricians, and an inconsistent pattern of adherence to guidelines. Still, data seemed to indicate an increase in acceptance rates for CF carrier screening in the subsequent year. Little was known, however, regarding the practical details of CF carrier screening administration. The overall objective of this study was to assess the incorporation by obstetricians of the ACOG recommendations for CF carrier screening into routine medical care. A questionnaire was sent to a sample of obstetricians from six different states selected from the American Medical Association (AMA) Physician Masterfile (N = 1,373). The questionnaire assessed the extent to which screening is being offered and agreed to, how pretest education is conducted, how results are communicated to patients, obstetricians' knowledge of CF, and obstetricians' attitudes regarding the guidelines. This study also determined relationships between these outcomes and other variables, such as demographic characteristics of physicians and attitudes about CF carrier screening. There were 723 respondents, which corresponded to a 61% response rate. There was variation in adherence to specific components of the guidelines. Nearly all obstetricians routinely offered CF carrier screening to patients with a family history of CF (97%) and most offered it to Caucasian pregnant

patients (77%); about half (52%) used printed educational materials in conjunction with direct talk, and, when test results come back positive, the majority reported offering to test the patient's partner (92%) and referring the patient to a genetic specialist (76%). Areas of lower adherence included offering testing preconceptionally to non-Caucasian women (39%), using videos for test education (<1%), having the patient sign an informed consent document (35% document at least some of the time that patients agree to testing), and using letters to communicate test results (10% when test results are positive). Test uptake was found to be significantly lower than uptake of CF carrier screening in pregnant populations in previous studies. Approximately half of the respondents estimated that less than 25% of their patients agreed to testing, while earlier studies quoted rates upward of 57%. Malpractice concerns did not appear as a key driver of test offering or consent documentation at this stage, even though it was clearly associated with the level of agreement to these guidelines. Finally, knowledge about CF was good overall, with a few noted areas for improvement. There is evidence that some components of the ACOG guidelines for CF carrier screening have been integrated into regular obstetrics care, but utilization of the test is relatively low. It is unclear the extent to which the guidelines will be followed in the future, what might increase adherence, and how incorporation of the guidelines into obstetrical care will affect the practice of geneticists and genetic counselors.

24) Causal Attributions and Perceived Personal Control in Caregivers of Children with Autistic Spectrum Disorders

Danielle Dong, Advisor-Benjamin Wilfond, MD, 2004. Oral presentation at NSGC 2005.

Abstract

The purpose of this study is to determine the association between causal attributions of primary caregivers of children with autistic spectrum disorders (ASD) and their perceived personal control (PPC). Primary caregivers were recruited through notices to autism-related support groups and advertisements in the Washington Post. Surveys could be completed on the internet or by mailing in a printed version. The survey assessed causal attributions with open-ended questions and the Causal Dimension Scale II, a previously validated measure. The survey also asked about the child's diagnosis, the age of the child at diagnosis, and perceived severity of the child's ASD, and assessed caregivers' levels of PPC over a number of aspects of their child's condition. There were 196 respondents. The majority were biological mothers (81%) or biological fathers (12%). 86% were caregivers for boys and 12% for girls. The children's diagnoses included Autism (40%), Asperger's syndrome (12%), Pervasive Developmental Disorder-Not Otherwise Specified (25%) and no response (21%). Respondents expressed a range of causal beliefs including genetics, environmental exposures, and congenital/birth injury. Additionally, caregivers' causal beliefs varied in their level of certainty. On a five-point scale (1= no control and 5= most control), caregivers control over their emotions was 3.0 (+/-0.9), control over their child's treatment and medical care was 3.3 (+/- 0.9), control over their child's symptoms was 2.7 (+/- 0.8), and control over personal relationships in their life was 2.9 (+/- 0.9). Further analysis will investigate the association between causal beliefs and perceived personal control.

25) The Range, Origins, and Strengths of Women's Attitudes Toward Amniocentesis

Julie Sapp, Advisor-Barbara Biesecker, M.S., 2005. Presented as a poster at NSGC 2005 and as a seminar at Kings College London.

Abstract

It is generally accepted in the U.S. that amniocentesis is an optional procedure made available to women at increased risk for having a baby with a chromosomal trisomy. Prenatal genetic counseling strives to assist women and their partners in making informed choices. An informed choice is one that is based on an understanding of quality information and is consistent with the decision-maker's attitudes. The Theory of Reasoned Action (TRA) provides a theoretical framework around which to conceptualize informed choice. According to this theory, a person's behavior is predicted by her behavioral intentions, which are shaped by her attitudes towards the behavior and the subjective norms she perceives to surround the behavior. The purpose of this study is to explore these dimensions of the TRA as they pertain to pregnant women's decision-making about amniocentesis: to gain insight into pregnant women's attitudes toward undergoing amniocentesis, to explore the sources or origins of these attitudes, and to examine important referents that participants have consulted regarding amniocentesis. These objectives were addressed in semi-structured telephone interviews with English-speaking pregnant women 35 years of age or older. Participants between 8 and 16 weeks gestation were recruited from a private prenatal diagnostic center and from ads in a local newspaper. The data generated from these interviews was subjected to coding for thematic analysis. Findings suggest that the majority of participants had ambivalent attitudes toward amniocentesis and considered their partners, friends, and physicians to be important referents when deciding whether or not to have amniocentesis. The implications of this exploratory study include its use as preliminary data for a larger, quantitative study of the components of the TRA as they relate to prenatal testing decision-making.

2005

26) Coping with a Presymptomatic Genetic Test Result for Huntington's Disease

Sarah Baxter Smith, Advisor-Rajiv Rimal Ph.D., 2005. Oral presentation at NSGC 2005.

Abstract

Research suggests that appraisals of a stressful event, such as the likelihood of change and perceived ability to cope, are important predictors of the use of particular coping strategies and overall adaptation. This study hypothesized that the predominant use of emotion-focused coping over problem-focused coping would be negatively associated with grief scores in individuals having presymptomatic genetic testing for Huntington's Disease (HD). Surveys were sent to 198 individuals having participated in presymptomatic genetic testing at Indiana University (N=98) and Johns Hopkins University (N=100). Eligibility criteria included individuals who tested negative or positive, and who were without symptoms of HD. Measures included coping self-efficacy, coping strategies, grief experience, exposure to HD, and experiences of undergoing predictive genetic testing. Among the 83 respondents (41.9% response rate), 63 surveys (45 negative/18 positive result) were eligible for analysis. On average, participants had known their test result for 6 years, were 45 years old, and 63.5% were female. Analysis revealed coping self-efficacy to be negatively associated with the use of avoidance coping ($p < .001$), and avoidance coping to be positively associated with grief scores ($p < .001$). Respondents showed greater use of problem-focused coping strategies compared to emotion-focused strategies ($p < .001$). Problem-focused coping strategies were negatively associated with grief ($p < .05$), while emotion-focused coping strategies were positively associated with grief ($p < .005$). An individual's coping self-efficacy predicts the use

of particular coping strategies which in turn can predict the grief experienced. Genetic Counseling interventions are proposed to maximize coping self-efficacy and effective coping strategies.

27) Use of Health Services Among Adults with Intersex Conditions

Tricia Gasser, Advisor-Colleen McBride, Ph.D., 2005. Presented as a poster at NSGC 2005.

Abstract

Previous research on individuals with intersex conditions suggests that constrained and secretive disclosure patterns have resulted in feelings of stigmatization and distrust of health care providers. Furthermore, there is theoretical and empirical data to suggest that having high levels of perceived stigma and low levels of trust have a negative effect on health seeking behavior. This study examined the associations between previous disclosure patterns, perceived stigma, trust in health care providers, and current healthcare utilization among adults with intersex conditions. Adults with intersex conditions were recruited from three main sources: support groups, health care providers, and alternative newsweekly advertisements. A total of 74 participants completed a questionnaire that included previously validated measures of perceived stigma, general physician trust, health status, and current health seeking behaviors. The questionnaire also included closed and open-ended items to assess previous disclosure patterns related to their condition and attitudes surrounding that experience. Demographic data was gathered, including participant age, insurance status, and time since receiving a diagnosis. The majority of participants self-identified as female, had private health insurance, and carried a diagnosis of androgen insensitivity syndrome. Participants reported having learned of their diagnosis at a median age of 21 years with a median time since diagnosis of 13 years. Bivariate analyses revealed a significant positive association between evaluation of the disclosure experience and general trust in health care providers. ANOVA showed significant differences in levels of perceived stigma between those who indicated receiving incomplete or inaccurate information from a health care provider about their condition and those who did not. Multivariate analysis revealed general trust in health care providers, having a usual place for seeking care, and overall health rating in the past 12 months to be significant predictors of total health care visits in the past 12 months. Having lower general trust predicted greater number of visits ($p < 0.05$). The results of this study suggest that how individuals learn of their condition may influence later feelings of perceived stigma, trust in health care providers, and use of health services. It is important for health care providers and genetic counselors to understand how disclosure experiences influence these outcomes, and work to incorporate more open and honest communication into disclosure standards. In addition, having lower trust in health care providers may lead to increased use of health services and greater health care costs. Interventions should be developed to increase trust among those with potentially stigmatizing conditions.

28) An Exploration of Psychiatrists' Management of Genetic Concerns: Implications for the Future

Marie Cudoc, Advisor-Barbara Biesecker, M.S., 2005. Presented as a poster at NSGC 2005.

Abstract

Due to recent developments in psychiatric genetics, the need for genetic counseling for patients with psychiatric disorders will likely increase. Current practices of psychiatrists referring patients for genetic counseling are unknown and there are few guidelines to help psychiatrists determine who should be referred. Thus, the goals of this study were to determine how genetic

information and counseling are incorporated into psychiatric sessions and what factors are the best predictors of psychiatrists' behavior related to the genetic concerns of their patients. These objectives were explored using a questionnaire sent to a representative sample of general adult and child psychiatrists from ten states who are members of the American Psychiatric Association (N=1216). Likert scales and semantic differential scales based upon the Theory of Planned Behavior, clinical vignettes, and open-ended questions were used to assess awareness of genetic issues, attitudes toward referrals, subjective norms, perceived personal control, behavioral intentions, current practices, perceptions of psychiatric genetics, and provider characteristics. There were 480 respondents (40% response rate). Most respondents were male (78%), white (88%), and had been practicing psychiatry for over 20 years (72%). 62% of respondents reported that they frequently or very frequently encounter patients with conditions for which there is a genetic component, but 76% of respondents reported that they referred none of their patients for genetic counseling over the past year. Most respondents (97%) reported that they discuss genetics with at least some of their patients. Yet, the majority (63%) of respondents reported that they do not know the name or contact information for a genetic specialist. Child psychiatrists indicated higher intentions to refer to genetic specialist than general adult psychiatrists ($p=0.001$). Multivariate analyses revealed that attitudes, subjective norms, and perceived personal control were all significant predictors of psychiatrists' intentions to refer and together, they explained 43% of the variance in intentions. Of these three constructs, subjective norms was the best predictor of intentions, accounting for 34% of the variance in psychiatrists' behavioral intentions. Although psychiatrists' indicate a high frequency of encountering patients with genetic concerns, the majority do not regularly refer patients with genetic concerns to genetic specialists. Our results suggest that strategies aimed at changing subjective norms, such as the development of clear written clinical guidelines for psychiatrists about when to make referrals to genetic specialists, may effect the most change in subsequent referral behaviors. Such interventions aim to increase collaboration between psychiatrists and genetic specialties so that both professions are better prepared for helping patients as genetic advances in psychiatry emerge in the future.

2006

29) Exploring Paternal Participation in Decision-making about Diagnostic Prenatal Testing Suzanna Schott, Advisor-Barbara Biesecker, M.S., 2006.

Abstract

Making a decision to undergo diagnostic prenatal testing such as amniocentesis can be a challenge because the procedure is invasive and involves a risk to the fetus. Both parents may be highly invested in the outcome of the pregnancy, therefore male-partners are likely to have a stake in the process of prenatal testing decision-making (PTD). It would benefit genetic counselors to have a better understanding of when, how, and why male-partners participate in decision-making. The objective of the current study was to illuminate some of the factors that shape male-partner involvement in the PTD process. While there are several variables that could influence participation, this study specifically sought to identify the attitudes, beliefs, and perceptions held by male-partners that might influence their participation behavior. The conceptual framework of the Theory of Reasoned Action (TRA) was utilized to structure a questionnaire to explore these potential antecedents of paternal participation in the PTD process. Participation was defined as sharing one's thoughts and opinions about whether the woman should undergo invasive, diagnostic

prenatal testing. Male partners were recruited via pregnant female patients who had been referred for genetic counseling at one of three perinatal clinics in the Washington D.C.-Baltimore region. Male partners were also recruited via general obstetric practices, via fliers and online advertisements. Generalizability of results was limited due to small sample size. Most respondents indicated positive attitudes toward participation and strong intentions to be involved in the PTD process. The men in this sample demonstrated moderate levels of ambivalence toward participation, but there were no significant relationship between ambivalence and behavioral intentions. Respondents indicated identification with several active roles in the PTD process, including information gathering, weighing pros and cons, persuasion, and support. Answers to open-ended questions indicate a number of motivations for participation. Suggestions for future research in this area are made, and potential clinical implications of the themes arising from the data are discussed. Recruitment challenges are discussed at length and alternative strategies suggested for the involvement of fathers in research about the PTD process.

30) The Role of Family History and Perceived Heredity in Adolescents' Adaptation to their Mother's Breast Cancer

Alissa Bovee, Advisor-Barbara Biesecker, M.S., 2006.

Abstract

The diagnosis and treatment of most cancers is a life-altering event for the patient, the spouse, and their children. While only a small percentage of breast cancer is thought to be hereditary, researchers have begun to consider how factors including family history, genetic information, and perceptions of heredity influence the way individuals cope with and adapt to cancer. Although existing research describes the emotional reactions, coping, and adaptation observed among adolescents when a parent has cancer, very little is known about the role of having a family history or perception of heredity in their coping and adaptation process. Lazarus and Folkman (1984) propose a model of stress and coping that suggests an individual's appraisals of a stressful event are important predictors of overall coping and adaptation. Theoretical and empirical data suggest that family history and genetic information influence appraisals of causal attribution, perceived control, and perceived burden. The goal of this study is to examine how a family history of cancer and perceptions of heredity influence personal control, impressions of burden, coping, and adaptation in adolescents whose mothers have had breast cancer. Participants were recruited from the Mid-Atlantic Cancer Genetics Network (MACGN), the FORCE support group for women at increased risk for cancer due to heredity, and the Y-Me support group for women who have had breast cancer. Families in which a mother has been diagnosed with breast cancer while their child was an adolescent were invited to participate. Mothers were asked to provide family history, genetic, demographic, and diagnostic information. Adolescents were asked to complete a self-administered questionnaire that included measures to assess their causal attributions, perceived control, perceived burden, coping strategies, cancer worries, benefit finding, and distress. Among the 35 adolescent respondents, the majority were female (71%) and the average age was 18. They were on average 15 when their mother was diagnosed with cancer, and it had been on average 2.8 years since. The average age of the 28 mothers was 48 years, they were largely Caucasian (89%), married (89%) and 61% had genetic testing. Almost all of the adolescents (30/35) had heard about another member of the family having had cancer, and about half reported having interacted with another relative who was affected with cancer. Adolescents causal beliefs about their mother's cancer were predominantly hereditary ($p=.000$), adolescents felt significantly more control over their emotions and relationships than over the symptoms and

treatments of cancer ($p=.000$), and used significantly more problem focused coping than disengagement coping ($p=.001$). Their level of distress was similar to a national sample of adolescents. Multivariate analysis revealed that the use of disengagement coping was predicted by lower perceived control ($p=.002$), and younger age ($p=.002$); that distress was predicted by increased use of disengagement coping ($p=.000$), and increased perceived burden ($p=.000$); that cancer worry was predicted by lower levels of perceived heredity ($p=.020$); and that benefit finding was predicted by increased levels of perceived burden ($p=.018$). There were no significant differences between adolescents with and without a personal experience with cancer with regard to the appraisals, coping, or indicators of adaptation. An adolescent's perceived control and burden predict the type of coping strategies they use, and together, appraisals and coping predict the way they adapt to their mother's breast cancer. Health care provider and group interventions are proposed to enhance control, coping, and benefit finding in this adolescent population.

31) Genetic Counselors Engaging as Advocates

Meredith Weaver, Advisor-Joan Scott, M.S., 2006.

Abstract

Advocacy is considered part of a genetic counselor's scope of practice by the American Board of Genetic Counseling. However, there is little research concerning the nature of advocacy in the genetic counseling profession. Existing advocacy literature in allied health care professions lacks a consistent definition of advocacy, as well as a comprehensive description of the range of activities that are considered advocacy. This study attempts to advance a definition of advocacy within the context of the genetic counseling profession by examining the range of advocacy-related activities described by a sample of genetic counselors. Telephone interviews were conducted with 27 key informants; 17 were initial informants identified prior to the study and 10 were identified by snowball sampling. Initial informants were genetic counselors who have been in practice for many years in a variety of clinical practice settings and thus could comment from an historical perspective on the range of advocacy activities in which the profession has been, and is now, engaged. The study design was qualitative and used a semi-structured, open-ended interview format. The questions explored four domains: (1) advocacy activities in the genetic counseling profession, (2) motivations for engaging in advocacy, (3) challenges to engaging in advocacy, and (4) how advocacy activities, motivations, and challenges have changed over time for either the participant or the profession. Interviews were tape recorded, transcribed verbatim, and coded for analysis. The majority of participants defined advocacy as a broad construct with elements of acting on someone's behalf or giving support. Three common advocacy activity themes emerged: (1) offering services, (2) education, and (3) engaging social and political institutions. Participants expressed an overwhelming belief that advocacy was part of the genetic counselor's job. The most commonly expressed challenge to doing advocacy was the lack of resources, support, and time in the health care setting. The most common change over time was that participants' range of advocacy activities had broadened, which some credited to a maturation of their career and professional goals. Motivations for engaging in advocacy were both internal and external, with more participants reporting internal motivations, including high self-expectations to advocate. Finally, the most common person to influence participants to advocate was another professional, such as another genetic counselor, a geneticist, or an administrator. This study documented the notable finding that advocacy is part of the everyday work of a genetic counselor because participants considered advocacy to be part of their professional role. This study also illustrated the difficulty participants had in giving concise definitions of advocacy; a working definition of

advocacy is put forth based on participants' words. This sample of genetics professionals considered many different activities to be advocacy and many advocacy activities part of daily genetic counselor practice. Participants' motivations for engaging in advocacy were overwhelmingly internal, which suggests an association between the type of person who follows a career in genetic counseling and the type of person who engages in advocacy. The descriptive findings from this study lay the ground work for future research about advocacy in the genetic counseling profession.

32) Does the general public interpret genetic information about heart disease risk differently than non-genetic information?

Kate Reed, MPH Advisor-Ben Wilfond, MD, 2006.

Abstract

Information about individuals' genetic susceptibility to common disease will be available clinically before treatments specific to certain genotypes have been developed. This study considers whether three sources of risk information about heart disease influence motivation for health behavior change and its precursors differently. Participants received one of three versions of a questionnaire. Each version contained one vignette about John, who receives risk information about heart disease based on assessment of his diet, family history, or genetic test results. In each vignette, John's risk was 20% above the general population risk. Participants (N=158) were asked what John should think, feel and do about his increased risk. Participants who received the vignette that based John's risk on family history were significantly more likely to believe he should have high motivation to see the doctor more frequently than were those in the genetic testing group. The source of risk information did not influence the level of motivation for the other types of risk-reducing behavior: avoiding dietary fat and exercising more often. Risk based on family history was also associated with greater likelihood of believing that John should have high self-esteem as it relates to his ability to stay healthy. These results suggest that risk based on family history may be meaningful to individuals making decisions about health behavior. This may reflect a greater familiarity with family based risk information. Further, risk assessment based on family history does not directly use an individual's genetic make-up or food choices, which may allow individuals to consider risk information more objectively. The current data call into question the assumption that genetic susceptibility information about common disease will lead to significantly greater motivation for health behavior change relative to other, currently available sources of risk information.

2007

33) Invasive Prenatal Testing Decisions in Pregnancy After Infertility

Colleen Brown, Advisor-Barbara Biesecker, M.S., 2007.

Abstract

Women who are pregnant after infertility often experience heightened anxiety regarding the outcome of the pregnancy. When choosing whether or not to have invasive prenatal testing they are faced with a complex decision, set in the unique context of a pregnancy that they often perceive as exceptionally precious. The objective was to explore the invasive prenatal testing decision-making experiences of women who are pregnant after infertility. A cross-sectional design

was used to investigate relationships between characteristics of the infertility experience, subjective norms, and decisional conflict associated with deciding whether or not to have invasive prenatal testing. The main outcome measure was decisional conflict. Study design was informed by Janis and Mann's conflict theory of decision making. One hundred forty one pregnant women with a history of infertility were recruited from pregnancy and infertility websites. A minority of women (28%) chose to have invasive prenatal testing. The mean level of decisional conflict was 20.4 (SD=21.7, range = 0=100); 74% of participants had lower decisional conflict. Half (49%) of women said that infertility made the prenatal testing decision easier or did not make it any harder, while 6% of participants said that infertility made the decision more difficult. The majority of medical and psychological infertility characteristics were not significantly associated with decisional conflict. In multivariate linear regression analysis, higher decisional conflict was associated with having a history of miscarriage, lower perceived severity of infertility, and subjective norms. There was an interaction effect between prenatal testing decision and subjective norms; test decisions that were incongruent with husband's preferences were associated with higher decisional conflict. Infertility may decrease decisional conflict associated with the prenatal testing decision by helping women to clarify their values and priorities. Perceived disapproval of the testing choice by other people is an important source of decisional conflict in this population. Husbands'/partners' opinions seem to be most important, although other infertile women were also influential.

34) The Experiences of Genetics Patients with Visible Differences who Facilitate Teaching in Genetics Clinics

Jamie Dokson, Advisor-Barbara Biesecker, M.S., 2007.

Abstract

It is common for genetics patients to facilitate the teaching of others during the course of their genetics exam. Patient response to serving as a teaching case has been studied only to a limited extent. This study describes the qualitative experience of patients with visible abnormalities in the genetics clinic when taking on a teaching role. The study aimed to improve understanding of the phenomenon of patients in educator roles, relate experiences to patient expectations, and fill a gap in the published literature. Thirty participants with visible physical abnormalities were interviewed by telephone about their experiences in the genetics clinic(s). The interviews followed a semi-structured interview guide, and were audiotaped, transcribed, and analyzed qualitatively. Data analysis elicited several common themes: Participants almost unanimously felt favorable towards current teaching practices in genetics clinics and welcomed the opportunity to educate others about their condition. No meaningful differences were found in patient reports of their experience in the genetics clinic as compared to other medical settings. Provider actions and demeanor had a considerable impact on patients' experiences. Common factors contributing to patients' views of the clinics and the genetics professionals were compiled and used to inform the development of new recommendations to further enhance educational experiences in genetics clinics.

35) Genetic Counselors' Experience of Moral Value Conflicts with Clients

Rachel J. Silver, Advisor-Gail Geller, Sc.D., 2007. Geller G, Micco E, Silver RJ, Kolodner K, Bernhardt BA. *Am J Med Genet C Semin Med Genet.* 2009 Feb 15;151C(1):31-40. [http://www.ncbi.nlm.nih.gov/pubmed/19170100?ordinalpos=2&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_DefaultReportPanel.Pubmed_RVDocSum]

Abstract

Genetic counselors may be faced with situations in which they disagree on a moral level with the views or decisions of their clients. These moral value conflicts between counselors and their clients have the potential to have an emotional impact on counselors. Little is known about the range of sources of these conflicts, the ways counselors conceptualize the conflicts, the approaches counselors take to address them, or the impact they have on the counselor's life and career. This study aimed to gain insight into the nature, sources, and consequences of moral value conflicts among genetic counselors. We performed semi-structured in-depth interviews with 27 genetic counselors. Interview questions were nested within interviews for a larger study entitled "Manifestations and Consequences of Moral Distress Among Genetics Service Providers". We asked participants about their personal experiences with moral value conflicts with clients, focusing on one case they found particularly distressing. Transcribed interviews were subjected to thematic analysis. We identified a number of situations under which moral value conflicts arise for genetic counselors, in the prenatal, cancer, and pediatric settings. Many of the conflicts involve either pregnancy terminations or circumstances surrounding cancer genetic testing that have implications for other family members. We propose a framework which can be used to describe the ways genetic counselors conceptualize the nature of their conflict. The majority of participants did not consider moral value conflicts with clients to be an overwhelming source of distress in their career. An unanticipated finding was that while most scenarios were described in distressing terms, a small number of counselors described ways in which moral value conflicts were enriching or rewarding experiences. Implications for training of genetic counselors are discussed.

36) Can Mental Illness Stigma Travel Through Blood? Exploring the General Population's Beliefs, Feelings and Behaviors Towards the Health Siblings of Individuals with Mental Illness

Matthew Thomas, Advisor-Colleen McBride, Ph.D., 2007.

Abstract

Individuals with mental illness experience considerable stigma from the general population. Recent findings suggest that healthy family members of individuals with mental illness also may experience stigma because of their biological/genetic relationship with the individual with mental illness. This study aims to explore the mechanism through which the general population stigmatizes healthy family members of individuals with mental illness and whether the nature of the familial relationship (i.e. biological sibling versus adopted) is associated with stigmatizing behaviors (i.e. desire for social distance). Participants (N=307) from the Maryland Motor Vehicle Administration completed a self-administered survey that includes two vignettes. The first vignette describes a hypothetical person with mental illness and the second describes the sibling of the affected person. In order to evaluate how the biological relationship between individuals with mental illness and their healthy relative influences social distance, we assigned half of the participants at random to view a vignette describing a biological sibling and half, an adopted sibling. The data were analyzed based on our conceptual model that illustrates the relationships between prejudiced thoughts, feelings, and discriminatory behavior towards an individual with mental illness and the resulting discriminatory behavior towards a healthy sibling. Participants' desire for social distance from an adopted sibling was not significantly different from their desire for social distance from a biological sibling, $\bar{X} = 1.86 (0.69)$ vs. $1.85 (0.67)$, respectively; $p=0.84$). The desire for social distance from a healthy sibling was almost significantly less for participants who

strongly supported a genetic causal attribution ($\bar{X} = 1.76$) when compared to participants who weakly supported a genetic causal attribution ($\bar{X} = 1.99$), $F(1,244) = 3.80$, $p=0.052$. Overall, our conceptual model received partial support as regression analyses revealed that certain prejudiced thoughts (dangerousness and prognosis) and feelings (anger, compassion, fear) towards an individual with mental illness predicted the desire for social distance from a healthy sibling. The strongest predictor of the desire for social distance from a healthy sibling was the desire for social distance from an individual with mental illness. Results suggest that individuals from the general population distinguish their desire for social distance when comparing an affected individual to his healthy sibling. Based on these findings, the concerns of some researchers that the recent focus on the genetic aspect of mental illness may have unintended discriminatory side effects for healthy biological siblings may not be warranted. Healthy biological relatives may not be at any greater risk for courtesy stigma than adopted siblings, even in an era where the biological cause of mental illness has become a primary focus in the media. Based on how the participants' views of individuals with mental illness predicted their views of healthy family members, if a more effective means of combating stigma towards the individual with mental illness arises, we would anticipate a decrease in subsequent stigma towards healthy family members.

2008

37.) Caregiver Adaptation to Autism Spectrum Disorders

Katie Voss, Advisor-Barbara Biesecker, M.S., 2008. Oral presentation at NSGC 2008.

Abstract

Pervasive developmental disorders (PDD) are a group of disorders characterized by impaired communication and social skills. Little information is available for caregivers of children with PDD regarding the cause of PDD, as well as the efficacy of the existing therapies and treatments, among other things. This uncertainty may present unique challenges as parents adapt to their child's conditions, particularly as it influences a caregiver's control beliefs, search for a cause or choice of coping strategies. A cross-sectional design was used to investigate the relationships between control beliefs, causal attributions, coping strategies and adaptation among caregivers of children with PDD. The main outcome measure was adaptation. Study design was informed by Lazarus and Folkman's Transactional Model of Stress and Coping. Three hundred and twenty four caregivers were recruited through support groups and websites designed for caregivers of children with PDD. Among the 324 respondents, the majority was female (95%), married (80%), had completed college (70%), were white (92%) and had an income greater than \$70,000 (51%). On average, the children had been diagnosed with a PDD five years ago. Caregivers felt that they had a moderate amount of control over all aspects of their child's life, but reported having the most control over their child's medical care and treatment. Multivariate analysis revealed that higher long-term course control and control by others, problem-focused coping, and controllable causal attributions were associated with greater levels of adaptation. In conclusion, though being a caregiver to a child with PDD has sometimes been characterized as a situation with limited opportunities for control, these caregivers reported many ways that they find control over many aspects of their child's symptoms and care. Control beliefs, particularly over long-term course, may serve as a potential target for clinical interventions aimed at enhancing adaptation. Furthermore, problem-focused coping was a significant predictor of adaptation for the

caregivers in this study, which may indicate the importance of these coping strategies, particularly in the years immediately following a diagnosis.

38.) Family History of Heart Disease and Barriers to Screening: How Do They Affect Intentions to Get Cholesterol Screening.

Courtney Nichols, Advisor-Joan Scott, M.S., 2008.

Abstract

Coronary heart disease is a common, complex disease for which individuals may lower their risk by adopting preventive behavior change. Family history has been suggested as a potentially a valuable tool for promoting preventive behavior change by assessing risk for complex diseases like heart disease. However, while family history has been shown to be a valid and reliable tool for risk assessment for coronary heart disease, the efficacy of risk information based on family history in promoting behavior change has not been well studied. If shown to be efficacious in promoting behavior change, family history holds potential as a public health tool, being more accessible to individuals with decreased access to other risk assessment strategies. Yet, it will only be most beneficial if recommended preventive behaviors are also accessible. This study sought to examine the contribution of a salience intervention of family history of coronary heart disease and perceived barriers to screening to intention to get blood cholesterol screening. In a one-time self-administered survey, a general population sample was queried about intentions to undergo blood cholesterol screening and beliefs that may influence the likelihood of taking preventive action based on the Health Belief Model (HBM). Beliefs under investigation included perceived barriers to and benefits of cholesterol screening, susceptibility and severity of heart disease, and heart disease worry. To study the role of family history salience, participants were randomly assigned into one of two groups. One group was asked to read information about family history as a risk factor for heart disease and respond to questions about their personal family history of heart disease before completing other study measures. The second group did the family history exercise only after they completed the study measures on beliefs. Analyses did not show a significant association of an intervention for salience of family history with intention to get cholesterol screening, however, the study was underpowered to detect this effect. Simply having a positive family history of heart disease was significantly associated with higher perceived susceptibility and heart disease worry, which in turn were associated with increased intention to screen in bivariate analyses. The results also showed an association between several perceived barriers and decreased intention to screen. The most common and significant barriers were related to knowledge/awareness of heart disease and screening practices, and to structural barriers to cholesterol screening. A stepwise regression model of perceived barriers showed that the most significant perceived barriers explained a majority of the variance in cholesterol screening intentions. A regression model including all HBM variables and sociodemographics showed heart disease worry and self-efficacy to have p -values < 0.100 . However, none of the HBM variables nor having a positive family history reached statistical significance in the model. Age and time since last cholesterol check showed significant associations, with older individuals and those who recently had their cholesterol checked showing the highest intentions to screen. Conclusions from the study are hindered by the lack of the desired sample size, which limited the power to address some objectives. The results did, however, indicate that individuals who have a positive family history for heart disease seem to understand that this increases their risk for the condition, with or without an intervention to make that risk more salient. While having a positive family history showed a trend toward increased intention to screen that did not reach significance, multivariate

analyses indicated that family history may play a smaller role in intentions to screen than other factors. It is suggested that important factors in intention for cholesterol screening include overcoming perceived barriers to screening, self-efficacy for screening, past screening behaviors, and heart disease worry.

39.) The Extent and Patterns of Disclosure within the Huntington Disease Community and the Association with Adaptation to Living at Risk.

Jessica Young Adcock, Advisor-Lori Hamby Erby, Sc.M., PhD, 2008.

Abstract

Little is known about the extent and process of disclosure within the Huntington's disease (HD) population. Descriptions of varying degrees of disclosure about HD status exist; however, few studies have sought to measure disclosure quantitatively and to describe patterns of disclosure. Disclosure is an important concept to consider in this population because disclosure decisions can significantly impact other at risk family members and can affect an individual's support and social isolation. In addition, HIV research suggests that a relationship between disclosure of one's HIV status and psychological adaptation to one's status may exist. This study aims to describe extent and patterns of disclosure about HD and one's status to family and friends among individuals who have tested positive or who are at risk based on family history. Additionally, this study aims to investigate the relationship of disclosure to psychological adaptation to living at risk for HD. Participants were recruited from HD clinics, HD support groups, HD online websites, and HD online mailing listservs. Eligibility criteria included individuals who tested positive for HD and who were without symptoms or individuals who were at risk for HD based on a positive family history and who were without symptoms. A cross-sectional survey consisting of closed and open-ended items was used to 1) investigate individuals' extent and patterns of disclosure about HD, 2) assess psychological adaptation to living at risk for HD, and 3) describe individuals' attitudes surrounding disclosure about HD. Among the 315 respondents, 191 surveys (136 at risk for HD, 55 with a positive HD genetic test result) were eligible for analysis. Participants were largely female, white, not of Hispanic origin, and married. Results revealed that the majority of individuals disclosed their HD status, the inheritance of HD, the features of HD, and their feelings and concerns about their HD status to the majority of members within their social network. However, the extent of disclosure about HD and one's status varied within the population, with evidence for selective disclosure and nondisclosure. Multiple regression analysis indicated that the extent of disclosure about HD status along with number of years an individual had known their HD status predict adaptation to living at risk for HD. This study demonstrates the complex nature of HD disclosure to others and also reveals an important association between the extent of disclosure about one's status and adaptation to living at risk. These findings also provide evidence for the need for health care providers (HCPs) to assess the full extent of disclosure, given the presence of selective disclosure and association with adapting to living at risk. Finally, these results suggest ways that HCPs can assist individuals in devising disclosure plans and strategies to cope with the consequences of their disclosure decisions.

40.) Meaning making among asymptomatic adults with a positive presymptomatic genetic test result for Huntington Disease.

Pooja Ramchandran, Advisor-Rajiv Rimal, PhD, 2008. Oral presentation at NSGC 2008.

Abstract

Huntington Disease (HD) is a degenerative, ultimately fatal neuropsychiatric condition inherited in an autosomal dominant manner. Eighteen to 25% of individuals at risk for HD pursue predictive testing. A positive test result can be a psychologically-threatening event, but the genetic testing process may be an important means for some individuals to gain control and make meaning. Meaning making is considered an important component of cognitive adaptation, but it is unknown whether, and if so how, individuals who test positive for HD use meaning-making strategies in response to their test result. This study describes the process of meaning making, and perceived barriers thereof, in adults with a positive genetic test result for HD who believe themselves to be asymptomatic. We conducted and analyzed nine semi-structured interviews that were informed by Park and Folkman's model of meaning making. Member checking interviews with these participants were conducted to ensure accurate interpretation of data. Results of this study indicate that, as expected, the immediate response to the test result was universally one of shock; this was accompanied by thoughts of despondency, before meaning making could be initiated. Participants then appraised and made positive meaning of their test results in three distinct ways: (1) searching for a cause, (2) benefit finding (including reappraisals of the experience, finding side benefits, focusing on the positive, avoiding the negative, downward social comparison, imagining worse situations), and (3) maintaining and relinquishing control. Perceived barriers to meaning making (including concerns about long-term care and risk to children) are presented. Results of this study expand the existing literature on meaning making and provide evidence for a less studied form of meaning making based in action. The findings also suggest approaches to counseling and facilitating meaning making and adaptation in individuals pursuing presymptomatic genetic testing for HD.

2009

41.) Screening Behavior in Adults with Hereditary Hemorrhagic Telangiectasia: Illness Perceptions and the Health Belief Model.

Melanie D. Baxter, Advisor-Alan Guttmacher, MD, 2009.

Abstract

The study aimed to better understand the factors that influence screening behaviors of adults with hereditary hemorrhagic telangiectasia (HHT). HHT is a chronic condition, but with early diagnosis followed by adherence to recommended screening guidelines, the major complications of this disorder can be lessened or entirely avoided, and disability or even death can be prevented. Despite this, many individuals with HHT do not follow recommended screening guidelines, even when they are aware of the risk of serious complications. The Health Belief Model (HBM) framed this study of HHT screening. In addition to the HBM constructs (perceived susceptibility, perceived benefits, perceived barriers, self-efficacy, response efficacy, and cues to action), this study also considered the role of illness representations. Participants were recruited from the HHT Foundation International, Inc., the "HHT Awareness" Facebook group, and six HHT clinics, in addition to a snowball recruitment technique. Eligibility criteria included adults who self-reported having a diagnosis of HHT. A cross-sectional survey consisting of closed and open-ended items was used to investigate the relationships among the domains of illness representations, HBM constructs, and the main outcome measure of HHT-specific screening behaviors in line with recommended guidelines. A total of 320 participants were included in analysis. Rates of CAVM screening, PAVM screening, and HHT annual check-up in this study population were 82.0%, 67.1%, and 56.5%,

respectively. Ordinal logistic regression analysis showed that perceived barriers ($\beta = -0.114$, $p < 0.001$), perceived susceptibility ($\beta = 0.117$, $p < 0.05$), treatment control ($\beta = 0.078$, $p < 0.05$), and emotional representations ($\beta = 0.067$, $p < 0.05$) were significant predictors of HHT screening. Qualitative findings revealed emotional and cognitive perceived barriers to HHT screening, including a perceived overall lack of healthcare providers who are familiar with and/or knowledgeable about HHT and a perceived lack of effort by healthcare providers to learn about HHT. Findings suggest that screening rates are not ideal in the population of adults with HHT, and that many factors are influencing behavior. These results also provide evidence for the overall educational need within the healthcare community regarding HHT, and may suggest several approaches that HCPs can consider to potentially improve screening adherence.

42.) Adaptation and Quality of Life Among Adults with Neurofibromatosis Type 1

Julie Snyder Cohen, Advisor-Barbara Biesecker, M.S., 2009.

Abstract

Neurofibromatosis type 1 (NF1) is a genetic disorder that primarily affects the skin and nervous system. NF1 carries a significant psychosocial burden for affected individuals. Aspects that are especially challenging include the variability in severity of symptoms and medical complications, uncertainty in progression, and vulnerability to stigmatization. The literature suggests that because of these and other challenges posed by NF1, affected individuals may struggle to adapt to their condition and, consequently, experience reduced quality of life (QoL). The aim of this study was to gain a more thorough understanding of the role of appraisals and stigma on adaptation and QoL among adults affected with NF1.

A cross-sectional design was used to investigate the relationships of appraisals and stigma as predictors of adaptation and QoL. Adults with NF1 were recruited via NF support organizations and websites, and through NIH clinical research protocols. Data were collected through a web-based survey. Approximately 482 individuals completed the survey. Overall, participants perceived a high level of social stigma, and there was a high prevalence (55%) of depressive symptoms in the study population. Multivariate analyses revealed that more positive/less negative appraisals and lower perceived stigma were associated with greater levels of adaptation and higher QoL. This study also identified new themes about the ways NF1 can have a positive impact on individuals' lives, and shed light on the conceptual relationship between adaptation and QoL. This study demonstrates the role of psychosocial factors, particularly appraisals, stigma, and depression, in influencing adaptation and QoL among adults affected with NF1. Affected individuals often struggled with adapting to their condition and experienced lower QoL. Social stigmatization was an especially challenging issue, and this may serve as a potential target for interventions aimed at enhancing adaptation and QoL. The results also provide evidence of the need for clinicians to routinely screen for depression among patients with NF1. More broadly, the findings offer insight into the relationship between adaptation and QoL, two important outcomes of living with a genetic condition.

43.) The Role of Hope in Adaptation to Uncertainty: The Experience of Caregivers of Children with Down Syndrome

Megan Truitt, Advisor-Lori Erby, Sc.M., PhD, 2009.

Abstract

Down syndrome (DS) is a condition characterized by impaired intellectual functioning and sometimes other issues that affect an individual's medical, social, and behavioral health. Although having a known genetic diagnosis may reduce some uncertainty, there are often uncertainties surrounding the prognosis of DS which extend into various aspects of a child's life. This residual uncertainty likely makes psychosocial adaptation more difficult for caregivers; yet some may view uncertainty as allowing for the opportunity of positive outcomes. In situations such as these where caregivers continue to face prognostic uncertainty, concepts such as hope may play a role in adaptation. It is not yet fully understood how caregivers adapt to having a child with DS in light of this uncertainty or the role hope plays in the process.

The objective of this study was to investigate the relationships between perceived uncertainty, hope, and adaptation in caregivers of children with DS. Participants were recruited from local and national DS groups and from a DS clinic list. A cross-sectional survey allowed for the examination of caregivers' levels of perceived uncertainty, hope, and adaptation, as well as the relationships between those three variables. The hope that caregivers had for their child in specific categories was also measured. A total of 546 surveys were eligible for analysis. Hope and adaptation were positively correlated, while perceived uncertainty was negatively related to both hope and adaptation. Perceived uncertainty and hope were found to be predictors of caregiver adaptation, with hope being the largest contributor to the variance in adaptation. Caregivers' motivation to reach their goals for their child was higher than their ability to think of ways to meet those goals, and the ability to think of ways to reach goals was significantly related to adaptation levels. Findings from this study suggest that caregivers' levels of hope and perceived uncertainty may be important concepts in the process of psychosocial adaptation. Results indicate that prognostic uncertainty tends to be perceived as a negative experience by these caregivers, being associated with lower levels of hope and adaptation. Although future studies are needed, strategies aimed at strengthening hope and reducing perceptions of uncertainty may increase caregiver adaptation.

44.) Testing for Inflammatory Bowel Disease: Predictors of Patients' Appraisals of Medical and Genetic Testing

Gillian W. Hooker, PhD, Advisor-Debra Roter, DrPH, MPH, 2009.

Abstract

Recent genetic and genome-wide association studies have identified a number of susceptibility loci for IBD and clinical integration of this genetic information is not far behind. Studies of attitudes towards genetic testing in this population have revealed overwhelmingly positive responses, with a majority of individuals surveyed stating that they desire genetic testing. However, the reasons for this remain to be seen, in the absence of interventions to prevent or treat disease following genetic testing. For many, IBD is a life-long illness which poses uncertain threats to one's quality of life. In this study, Leventhal's Common Sense Model of Self-Regulation is used as a framework to describe how individuals cognitively and emotionally represent their illness in response to the threat of chronic disease, and how these representations predict responses to hypothetical clinical testing scenarios. This study had an experimental design in which participants were randomized to a vignette depicting either a genetic testing scenario or a standard "blood" testing scenario. Participants were asked a series of appraisal questions about the person in the vignette and appraisals were compared between the two vignettes. Also, participants' perceptions of their own illness were assessed and analyzed as predictors of their response to the two vignettes. Finally, a measure of genetic literacy was incorporated to examine whether genetic literacy is a predictor of response to the vignettes. Patients were recruited to take the survey

through the Johns Hopkins Gastroenterology clinics either during their clinic visits or by mail. A total of 200 surveys were collected and analyzed for this thesis. The two study groups were comparable across demographic, disease-related and situational variables, as well as illness perceptions and genetic literacy. Differences were not observed in perceived control over IBD following a positive test result for the person in the vignette, nor were affective responses to the vignette (positive or negative feelings) different between the two groups. Participants were able to extrapolate information and applications offered uniquely by the genetic testing vignette. Though overall outcomes were not largely different, the factors predictive of those outcomes did differ between the two testing scenarios.