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Attitudes regarding genetic counseling issues of the Pakistani population at Maimonides Medical Center in Brooklyn. S.K. Barrett, D.A. Rosa, S. Begum, A. Hafeez, and G.S. Kupchik. Maimonides Medical Center, Brooklyn NY and SUNY Health Sciences Center at Brooklyn.

Maimonides Medical Center is located in an area of racial and ethno-cultural diversity. Approximately 15% of the patients in the medical genetics division originate from Pakistan and many practice a conservative form of Islam. Abortion, autopsy and prenatal diagnosis are generally considered to be forbidden by Islam. In our experience, many patients adhere to this belief system, while others seem to make reproductive decisions independent of religious influences. We have developed a survey designed to examine demographic and social parameters and how those factors may influence response to three scenarios: prenatal diagnosis, abortion, and autopsy. The Pakistani subjects and their partners (when present at the counseling session) were asked to fill out the questionnaire and respond to what they would likely do in such a situation. The questionnaire was also presented to pregnant Pakistani couples NOT referred to genetics, and controls consisting of non-Pakistani genetics patients of various ethnic and religious backgrounds living in Brooklyn. The response of this population on critical genetic counseling issues as well as possible correlations to demographic/social variables and previous experience with genetics services will be presented.

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Patient perspectives on the process of informed consent for DNA testing. C. Cytrynbaum[1], R.Babul-Hirji[1], M. Rowell[2], K. Henderson[3], K. Australie[4], H. Druker[1], L. Dupuis[1], N. Quercia[1], C. Shuman[1], S. Kennedy[1]. Division of Clinical and Metabolic Genetics[1], Department of Bioethics[2],The Hospital for Sick Children and University of Toronto, Ontario, Canada., Division of Medical Genetics, Montreal General Hospital, Quebec, Canada[3]. Yale University School of Medicine, Connecticut, U.S.A.[4]

The informed consent process (ICP) has long been recognized to be an important component of clinical practice and , more recently, of research as well. Yet limited information is available regarding the impact of ICP on patients and their families. We set out to evaluate patient perceptions of ICP which includes a newly devised DNA testing/banking consent form. In addition to addressing the issues currently recognized to be critical to informed consent, this form allowed individuals to choose between the options of closed consent (sample destroyed after requested testing completed) versus open consent (sample banked for use in ongoing research). Participants completed self-administered questionnaires pre and post ICP. Results are available from 50 completed questionnaires to date. 76% of participants chose open consent and 24% chose closed consent. 72% indicated an interest in a detailed discussion in the pre-counselling questionnaire and 76% responded that ICP was helpful in the post-counselling questionnaire, although 83% indicated that they had decided to proceed with testing prior to ICP. Of note, 68% agreed with the statement "consent forms are mostly to provide legal protection for health care professional". Anxiety levels ascertained by visual analog scale revealed no significant change in anxiety levels pre and post ICP in half of the respondents. The remaining respondents reported increased or decreased anxiety levels with approximately even distribution. Preliminary findings indicate a positive attitude towards ICP and provide justification for its implementation in clinical practice. More detailed data analysis, from a larger sample size, will be presented and this information will be utilized to develop guidelines to improve ICP to better meet patient needs.

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Genetic counseling for mitochondrial disorders. M.A. Del Vecchio, G.L. Matika, C. A. Bay. Children's Hospital of Pittsburgh, Pittsburgh, PA.

Mitochondrial disorders are being diagnosed with increased frequency, and present clinicians and genetic counselors with a multitude of issues to address. Mitochondrial disorders generally result in deficient energy production or availability. Mitochondrial disorders are typically thought of as maternally inherited, but all modes of inheritance have been described and need to be considered during the genetic evaluation. The counseling issues involved with mitochondrial disorders that are inherited in a Mendelian fashion are similar to non-mitochondrial Mendelian disorders. The maternally inherited mitochondrial disorders present the genetics team with a variety of unique issues to address. Mitochondrial disorders can affect virtually all organs systems. Most commonly the central nervous, cardiac, muscular and ophthalmologic systems are involved. When suspicious of a mitochondrial disorder it is necessary to determine the most likely mode(s) of inheritance, explain difficult genetic concepts and to provide psychological support. While these roles are typical for a genetic counselor, mitochondrial disorders do present a unique set of challenges. The genetic counselor must obtain a very targeted pedigree with special attention paid to the "soft signs" in family members, such as migraines, seizures, mental retardation, gastrointestinal complaints, chronic fatigue and weakness. Explanations of difficult concepts such as heteroplasmy, oxidative phosphorylation, nuclear DNA, mitochondrial DNA, and nuclear-mitochondrial gene interactions are unique Providing psychological support can be a daunting task, even to an experienced team. When one makes a diagnosis of a maternally inherited mitochondrial disease in a proband, one usually also makes the same molecular diagnosis in the mother, and potentially other maternal-line relatives. In many cases prognosis varies considerably, thus counseling must by necessity be vague as to the prognosis. Frequently there is no reliable prognostic indicator that can be used to predict future health concerns Like with other genetic disorders the diagnosis is often accompanied by feelings of grief, despair and guilt. While those feelings are not specific to maternal line inheritance, the extremely high recurrence risk in maternal line disorders is specific to this type of inheritance. Reliable prenatal testing is not available for many mitochondrial disorders, and reproductive options and treatment are limited. In summary, mitochondrial disorders, especially those inherited maternally present many unique and difficult issues for the genetic counselor.

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Constructing rapport in televideo genetic counseling. L.A. Flore[1], S.T. Risinger[2], D.W. Britt[2], I.E. Zador[1], A.D. Gilbert[1], M.I. Evans[1] and A. Johnson.[1] [1]Ob/Gyn, WSU, Detroit, MI, and [2]Sociology, WSU, Detroit, MI.

The typical strategies that genetic counselors use to establish rapport with patients in face-to-face situations are undermined by the used of televideo. Leaning forward to show concern and support, establishing direct eye contact to convey interest, respect, and receptiveness, and being able to reach out and touch someone when they display signs of anxiousness or fear: these things just do not "work" in a televideo counseling session. In a series of pretest using PC computers networked to simulate an Internet-like connection, video cameras, and headsets, we have developed a set of strategies for establishing a physical and social context conducive to the development of rapport in televideo counseling sessions. The physical context of the session may be optimized by altering camera placement to approximate direct eye contact and to permit viewing of both facial features and hand gestures; and by orienting the patient to the unique features of the televideo equipment (such as cursor movements, the short audio time delay, positioning of visuals aids, and so on). The social context of the televideo consultation may be made more conducive to the development of rapport by establishing patient control over the situation (having them adjust the camera for better viewing, encouraging the patient to use non-verbal cues to interrupt the counselor, and having the counselors practice ways of repairing communication break-downs that occur from "talking over" one another). These general strategies may be supplemented by having the counselors practice the kinds of rapport-building techniques that seem to work in both traditional face-to-face and televideo counseling sessions: using non-verbal cues(smiling, nodding), verbally assessing patients' understanding and perception of risks, verbally expressing empathy, etc. We are utilizing this protocol in an on-going prospective trial regarding the use of televideo genetic counseling in an advanced maternal age population.