platform presentations in genetic counseling/ perinatal genetics

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I don't want to hear you, but I'd like to see you. C.A. Downs. University of Michigan, Department of Ophthalmology and Vision Sciences.

Purpose: To compare attitudes towards clinical genetics between respondents who are Deaf and those who are Deaf-Blind. Previous studies involving Deaf patients have been limited by the barriers inherent in obtaining responses in a clinical setting as well as by a language barrier. This is the first work that elicited non-censored responses from Deaf and Deaf-Blind adults. **Methods:** Participants in a retreat for Deaf-Blind adults were interviewed concerning their views relating to genetic counseling and treatment of hearing and vision loss. Respondents consisted of Deaf adults (those who identify as part of the Deaf culture in the U.S. and Canada and whose primary language is American Sign Language) and Deaf-Blind adults (those who have both hearing and vision loss). Initial interviews were conducted in-person in the respondent's preferred language and mode of communication. Communication was direct (no interpreter needed) and included American Sign Language (ASL), tactile ASL (the Deaf-Blind respondent read ASL by placing his/her hands on the interviewer's hands while the interviewer signed), and spoken English. Follow-up interviews were obtained within a six-month period. **Results:** The Deaf adults had negative views towards medical genetics, drawing parallels between treatments to "cure deafness" with eugenics and genocide. However, one respondent would be interested in genetic counseling if the medical personnel had positive attitudes towards the Deaf and would meet the needs of Deaf clients. The Deaf-Blind adults fell into two categories. Those whose communication included both signing and oral methods were interested in preserving both their hearing and vision. Those who identified as culturally Deaf had similar responses as the Deaf adults in terms of resenting the idea of "curing deafness," while at the same time interested in any avenues that would preserve or restore vision. **Conclusions:** To provide appropriate services to Deaf and Deaf-Blind adults, it is imperative to establish trust and effective communication. Identifying a contact in the Deaf community who can serve as a cultural mediator is an important tool. In addition, expanding the time frame for clinical appointments and utilizing the preferred mode(s) of communication conveys respect and allows for successful communication. The attitudes of Deaf-Blind adults towards medical interventions differed between those who communicate orally and those who identify as culturally Deaf.

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Optimal cancer risk assessment program professional roles; analysis of 30 American centers. E.R. Knell and C.A. Presant. Los Angeles Oncologic Institute (LAOI) and California Cancer Medical Center, Los Angeles, CA.

Genetic cancer risk assessment programs, until recently found only at major university and teaching hospitals, have moved into clinical use in community-based centers. While genetic counseling is recommended, little emphasis has been placed on what constitutes comprehensive risk assessment and counseling. As risk assessment has moved into community usage, little attention has been given to the types of professional or medical staff required for an effective, quality program. To better understand the scope and practices of existing programs, we surveyed members of the National Society of Genetic Counselor Cancer SIG. Here we report on some of our detailed findings from 30 programs who responded.

Most programs (23/30) relied on the genetic counselor as gatekeeper for entrance into the program, and an additional 4 had no restrictions. The majority of programs (21/30) did not involve a physician anytime during the first visit, yet most (23/30) presented risk figures at this time, thus relying on the expertise of the genetic counselor in education, as well as family history collection, evaluation and interpretation. The counselor spent a minimum of 5 hours per patient in pedigree analysis, counseling, informed consent and test interpretation. Many programs (11/30) relied on the counselor as the main or even sole contact, and often genetic testing decisions were at the discretion of the counselor. The more respected programs utilized the unique training and skills of the genetic counselor for risk assessment and counseling issues, as well as for communicating detailed knowledge of genetic risk and gene testing, while relying on the physician for expertise on medical management issues, tailored to the risk for the individual, considering their mutation status and the particular cancer syndrome.

We conclude, from this and additional data to be presented, that a balanced program requires the expertise and coordination both of a genetic counselor and an oncologist, as well as the availability of other professionals. The implementation of genetic testing without a genetic counselor poses risks for the patients: inadequate collection and genetic interpretation of the family history, only empiric risk analysis, as well as misunderstanding of genetic testing. Cancer risk programs are improved by physician consultation, particularly with description and consequences of possible interventions and promoting and monitoring compliance of individuals with recommendations.

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A Practical Theory-Based Method to Improve Lay Decision-Making for Genetic Testing. J.R. Sorenson, C. Lakon, T. Spinney, T. Jennings-Grant. University of North Carolina, Chapel Hill, N.C.

Research on lay decision-making for genetic testing has identified many concerns. These include (a) discussions of testing risks/benefits often reflects a professional more than a lay perspective; b) risks/benefits typically provided to patients constitute only part of a broader list of lay reasons for/against testing; and (c) patient's personal considerations are as important as medical considerations in a patient's testing decision. Drawing on decision-making theories in psychology, we developed an instrument that asked women at-risk to be a carrier of a Hemophilia A mutation to list (a) the personal consequences for themselves and significant others of accepting/declining carrier testing; (b) the reasons for accepting/rejecting testing in terms of their and significant others values and beliefs; and (c) their four most important reasons for/against testing. Women are provided with examples of the above. Innovative aspects of this approach include: (a) women construct their own list of personal reasons for/against testing instead of getting a provider list of risks and benefits; (b) the instrument can be completed at home allowing women time to consider the test; and (c) the instrument can be used for provider-lay discussion of testing in the clinic. Pilot work and trial experience (N=76) suggest: (a) women of varying educational backgrounds can complete the instrument; (b) women identify reasons for/against testing beyond examples provided; (c) many of the most important reasons for/against testing are reasons women generate, not examples provided; (d) the most important reasons cited include personal values/beliefs, not just risks/benefits; and (e) women using the instrument accept testing at the same rate as women who do not. The method is applicable to many genetic tests in various clinical settings.

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Lack of a cardiac bulge in human growth disorganized embryos: evidence for cardiac malformation leading to pregnancy failure
C.Craven[1-3x], W.Bugielski[2x], C.Castro[2x], T. Macpherson[2,3x] Magee-Womens Research Institute[1], Magee-Womens Hospital[2], and the University of Pittsburgh[3], Pittsburgh, PA

Introduction: Cardiac malformations occur in some humans who have chromosome abnormalities or malformation syndromes. Genetic control of heart development has been investigated in animal models, and early pregnancy failure is observed in mice when certain genes are altered. We hypothesize that some Growth Disorganized (GD) human embryos have had a failure of cardiac development. We suggest that cardiovascular abnormalities contributed to the pregnancy failure and spontaneous abortion (SAB). Methods: To test this, we reviewed the Embryo Collection of the Perinatal Pathology Service of Magee-Womens Hospital, comparing 35 externally normal embryos, Carnegie Stages 11-16, to 41 externally abnormal embryos, GD 2 and 3, for external evidence of normal heart formation: the presence of a heart bulge. We also evaluated the gross appearance of placental villi, if these were stored with the embryo. A difference in frequency of observations between the two groups was sought by $\chi 2$ analysis. Results: Each 35 of the normal embryos had a heart bulge, and all 10 of the embryos with stored villi had grossly normal villi. The GD embryos were significantly different. Only 11 of 41 GD embryos (27%) had a heart bulge ($\chi 2 = 39.3$, $p<0.001$). Of the 17 GD embryos stored with villous tissue, 8 had abnormal villi (47%, $\chi 2 = 4.55$, $p<0.05$). The 8 GD embryos without a heart bulge were significantly more likely to have abnormal villi (7 of 8, 88%) when compared to the 9 GD embryos with a heart bulge (2 of 9, 22%, $\chi 2 = 4.86$ $p<0.05$). Conclusions: GD embryos show evidence of failed cardiovascular development. Significantly fewer GD embryos have a heart bulge, and significantly more have abnormal placental villi compared to the normal embryos. Histologic studies of cardiovascular development in these embryos may be useful in understanding normal human heart formation and abnormal heart formation in some cases of pregnancy failure.