PERSPECTIVES IN GENETIC COUNSELING

NATIONAL SOCIETY OF GENETIC COUNSELORS, INC.

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CASE REPORTS IN GENETIC COUNSELING

Case No. 5

The following case report exemplifies an on-going problem for many genetic counselors. We learn to be honest with our patients and to help them deal with the reality of their particular situation, no matter how difficult that situation may seem. Is it the responsibility of the genetic counselor to be sure the patient is aware of all the ramifications of the situation, particularly if there are implications for other family members? At what point do we draw the line between respecting the patient's wishes and providing complete information?

D.P. was referred to our genetics department early in her pregnancy because she was 36 years old. During our routine intake process, she mentioned to the secretary that she has two maternal cousins who have hemophilia (sons of her mother's sister). Her maternal grandfather also had hemophilia. She also mentioned that it would be best not to speak with anyone else who might answer her telephone at home about the family history of hemophilia.

When I called the patient to schedule an appointment for genetic counseling I asked her if her husband knew about the family history. She said that they had talked about it a little bit but there had not been much time for discussion of hemophilia. She added quickly that it did not matter anyway. We scheduled an appointment for the couple to come in for counseling. That appointment was later rescheduled by D.P. for a day when her husband would be out of town. She said that he did not really want to come in for any further information.

When we met I found out that the only other person in D.P.'s house who might answer the telephone was her husband. It became apparent that D.P. had not explained her family history to her husband. When I inquired about that, she stated that she and her husband had known each other for 18 years and that this was just not that important relative to everything else they had been through together. I suggested that she discuss the information thoroughly with her husband so that he could help her make an informed decision.

We discussed the importance of finding out what type of hemophilia affected her cousins and her maternal grandfather so that we could offer her prenatal diagnosis. She said she would have medical records sent and that she would call her maternal aunt. D.P. added that she probably did not want any special prenatal diagnosis other than the chromosome analysis and the alphafetoprotein (AFP) studies that are done routinely for everyone who has an amniocentesis for advanced maternal age. I reassured her that the decision was hers, but it would be helpful to get medical records on her cousins to be thorough.

She called me back the next week and told me that her cousins had Classic Factor VIII deficiency. We again reviewed the option of specific prenatal diagnosis for hemophilia. She said she was not sure about the procedure, so I suggested she discuss it further with her husband and get back to me as soon as possible.

Several weeks later, D.P. called to tell me she had almost miscarried. D.P. said when she came so close to losing the baby it convinced her that she wanted to have the baby no matter what the tests revealed. She said she would just like to have the routine amniocentesis. She also included that this was a joint decision made by her and her husband.

The day of D.P.'s amniocentesis was the first time I met her husband. Earlier on the telephone, she had asked that we not talk about the hemophilia because it was not an issue anymore. Again, I asked whether or not her husband was fully aware of the situation and the risks. D.P. said yes. The amniocentesis was performed without complications. Two weeks later, the results revealed 46,XY and a normal AFP level.

I called D.P. to let her know the results. I asked if both she and her husband could get on the telephone so that I could give them the results. The couple was quite happy about the normal chromosomes and AFP. I then mentioned that we could test the baby's factor VIII levels at birth. That would allow us to know as soon as possible if any special precautions should be taken. As soon as I said that, her husband asked, "What is factor VIII?" D.P. said quickly she would go over that with him later. She then thanked me and hung up the telephone.

The conversation that had just transpired made me feel very uncomfortable. It became clear that if D.P. had explained her family history to her husband, either it was incomplete or he did not understand. I became even more uncomfortable when I discovered that this couple was married after D.P. became pregnant. Had D.P. lied to me about how long they had known each other? Had she also lied to me about the fact that she had explained the whole situation to him? Was it my responsibility to inform this husband and father-to-be that his son had a 25 percent risk of having hemophilia? To whom is the genetic counselor responsible?

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BOOK REVIEW

Psychological Aspects of Genetic Counseling, by Alan Emery and Ian Pullen, London, Academic Press, 1984, 236 pages, $32.00 (cloth), $14.00 (paper).

Genetic counseling has arisen as a chimera of research in human genetics, preventive health care, and crisis intervention counseling. That segmented approach changed in 1979 with the publication of Seymour Kessler's book about the psychological dimensions of genetic counseling; Kessler's work ushered in an era of "person-oriented counseling." The trend toward a more integrated, personalized approach to genetic counseling is now

extended with the appearance of Emery and Pullen's *Psychological Aspects of Genetic Counseling.* This collection is intended for medical geneticists, genetic associates, and other providers of health care and social services.

In the opening chapter Emery sets the stage for the material that follows by discussing principles of genetic counseling. He documents current changes in the attitudes of the public and health-care professionals. Those changes are reflected in trends that include increased awareness and use of genetic counseling and prenatal diagnosis, and greater emphasis on the specific needs of the individual and family who seek counseling. It is disappointing to see no mention of nonphysician genetic counselors as a component of the genetic counseling team. That would reinforce the authors' point about changes within the profession.

Sixteen additional chapters by recognized British and American experts cover topics of interest to most genetic counselors including basic counseling techniques, coping and decision making, marital pathology, Down syndrome, mental and physical handicaps, and prenatal diagnosis. The text exceeds other available collections on medical genetics by including essays on the psychological aspects of early infant loss, infertility, sterility, and artificial insemination.

Some chapters are less successful than others in their originality and usefulness. The chapter on counseling techniques, for example, appears written primarily for the general practitioner who will try his or her hand at genetic counseling shortly after reading the chapter. While the essay contains useful illustrations of open-ended questions and techniques to encourage patients to talk about sensitive matters such as sexual difficulties, the overall discussion is too brief and superficial for practicing genetic counselors.

Of exceptional merit is the chapter by Nancy Wexler on counseling families with Huntington disease. She makes a persuasive plea for counselors to take a preventive role in dealing with psychological issues. That is necessary because "families with a genetically ill person, or individuals at risk, live in a state of varying chronic and acute stress" (p. 140). By putting Huntington disease in the context of stress management and adaptation, Wexler offers opportunities for intervention. Specific proposals include teaching clients at risk to recognize the wide range of normal behavior and to appreciate their own baseline functioning and variation. Clients can also be taught to avoid symptom-seeking and misinterpretation and to redirect their energies toward health maintenance, psychological adaptation, and personal and social competence. As she advocates the counselor's encouraging the client with a sense of realistic optimism, so too does Wexler foster optimism in counselors. One's dark feelings of helplessness about the unrelenting progress of Huntington disease fade with Wexler's illumination of the many possibilities for meaningful action. Wexler's reminder of the words of Robert Louis Stevenson, "life is not a matter of holding good cards, but of playing a poor hand well," may be one of the most succinct descriptions ever proposed as the central task of genetic counseling.

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A number of other pleasant surprises emerge in this collection. One is an appendix containing recurrence risks for a number of common conditions with Mendelian, chromosomal, or multifactorial etiologies. Another is a complete subject index, making this a useful reference resource. Finally, the quality of writing is uniformly clear and concise and without technical jargon. The only editorial omissions of note were the failure to use gender-inclusive language in all chapters and omission of a contribution by a genetic counselor. The authors' stated purpose, "to provide practical guidance to important psychological problems involved in genetic counseling and the skills required to tackle them" was, with few exceptions, accomplished very well.

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RESOURCES

Amniocentesis ... A Parent's Choice (10 minutes; $295) and *Amniocentesis/Chorionic Villus Sampling . . . A Choice to Make* (12 minutes; $495), 1985. Available for preview or purchase from Communication Resources, 2021 West Main Street, Houston, TX 77098, phone: (713) 529-3312.

Communication Resources, in conjunction with a team of experts in prenatal diagnosis, has developed two videotapes for use as an adjunct to a prenatal genetic counseling session. The news release that accompanies the videotapes states that "with this program you can: (1) save 15-30 minutes per counseling session; (2) consistently present pertinent information; (3) improve patient understanding; and (4) increase elections for procedures."

Amniocentesis . . . A Parent's Choice presents concise, accurate information about the importance of fetal chromosomal makeup to pregnancy outcome, the effect of maternal age on chromosomal makeup, and the significance of screening for neural tube defects. The procedure, risks, and reliability of amniocentesis are accurately described and illustrated with numerous graphics.

Amniocentesis/Chorionic Villus Sampling . . . A Choice to Make expands the information presented in *Amniocentesis . . . A Parent's Choice* to include data and graphics describing chorionic villus sampling. The material presented is generally accurate; the illustration of the procedure for chorionic villus sampling, however, suggests that 5-10 cc of fluid are withdrawn from the gestational sac during the procedure. Patients viewing the film should be informed that the fluid depicted in the syringe is culture media into which the small sampling of chorionic villi is aspirated and has not come from the gestational sac.

There is a basic assumption that more informed individuals are more likely to make decisions based on facts rather than myths or hearsay, and that certainly seems to hold true in the field of prenatal genetics. And, although both tapes are factually correct, I recommend that each counselor preview the videotapes critically; be sensitive to the formating of the films. The use of the computer in the films is distracting and the delivery style of the presenter may be interpreted by some patients as "talking down to the viewer." Both tapes have the potential to fulfill the first three goals described earlier. The fourth goal, "increase elections for procedures," may be beyond the capability of these programs.

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ANNOUNCEMENTS

Workshop in Human Teratology

The second workshop in human teratology will be held 10 July 1986 in Boston, in conjunction with the 16th meeting of the Teratology Society. The focus of this workshop will be methods of human risk evaluation. For further information and materials, please contact Marjorie H. Tasin, MS, Children's Hospital Medical Center of Akron, Genetics Center, 281 Locust Street, Room 530, Akron, OH 44308, phone: (216) 379-8792.

Annual NSGC Conference

The Sixth Annual Education Conference of the National Society of Genetic Counselors will be held in Philadelphia on 30 October-1 November 1986, immediately prior to the meeting of the American Society of Human Genetics. The meeting is titled "Strategies in Genetic Counseling: The Challenge of the Future." Solicitation forms for abstracts will be mailed soon. For further information, please contact: Susie Ball, MS, Central Washington Genetics Program, Yakima Valley Memorial Hospital, 2811 Tieton Drive, Yakima, WA 98902, phone: (509) 575-8160.

POSITIONS AVAILABLE

Genetic Counselor: A position is available with the established maternal serum alphafetoprotein screening program based in the Division of Maternal-Fetal Medicine, Department of Obstetrics and Gynecology, University of North Carolina School of Medicine. Responsibilities will include interpretation and reporting of test results, counseling of high-risk patients, public and professional education, and data collection and analysis. A master's degree (ABMG certified/eligible) and experience in genetic counseling required. You may contact: Lauren Lingley, University of North Carolina, School of Medicine, Department of Obstetrics and Gynecology, 214 MacNider Bldg. 202H, Chapel Hill, NC 27514. Minorities and females are encouraged to apply. An affirmative action/equal opportunity employer.

Genetic Associates: Two positions are available 1 June 1986 at the clinical faculty (assistant in pediatrics) level within the Division of Genetics, Department of Pediatrics at the University of Florida. A master's degree in genetic counseling and certification eligibility or certification by the American Board of Medical Genetics are required. This is a diversified position that involves genetic counseling for prenatal diagnosis and a wide range of disorders in a pediatric population, education of health professionals, and participation in satellite and specialty clinics and clinical research. Interested applicants should submit a curriculum vita and three letters of recommendation by 1 April 1986 to: J.L. Frias, MD, Professor and Chief, Division of Genetics, Box J-296, JHMHC, University of Florida, Gainesville, FL 32610. For more information about the position, please call Shearon Roberts or Sonja Rasmussen at (904) 392-4104. The University of Florida is an equal employment opportunity/affirmative action employer.

Genetic Counselor: The Division of Genetics at Vanderbilt University will have a position available for a genetic counselor in April 1986. The responsibilities include counseling patients and families seen in the genetics, neurogenetics, and prenatal diagnosis clinics, as well as those from consultations and satellite clinics. Applicants who are board eligible or certified in genetic counseling are preferred. Salary is commensurate with experience. Applicants should contact: John Phillips, MD or Melinda Cohen, MS, Vanderbilt University School of Medicine, T-2404 Medical Center North, Nashville, TN 37232, phone: (615) 322-7601.

Genetic Counselor: A full-time position is available immediately at the North Los Angeles County Regional Center, a private, nonprofit corporation serving developmentally disabled persons and parents at risk. Primary responsibilities include amniocentesis counseling, general genetic counseling, outreach education, participation in genetics clinic, as well as program initiation and development. Applicants must be board eligible by the American Board of Medical Genetics. Full use of an auto is required. To apply, send letter of interest and resume to: NLACRC, 14550 Lanark Street, Panorama City, CA 91042, attention: Gary Frohlich, Prevention Unit. The North Los Angeles County Regional Center is an Equal Opportunity Employer, M/F.

Genetic Associates: The Southern California Kaiser Health Care Program has openings for genetic associates in the Fontana, Bellflower, and Harbor City areas. Duties will include working in MS-AFP screening, prenatal diagnosis, and clinical genetic counseling. Candidates should be board certified or board eligible in genetic counseling. Starting salary is $25,000, based on experience and qualifications. Excellent benefits. Send inquiries to: Diane L. Broome, MD, Anaheim Medical Arts Building, 1188 N. Euclid Avenue, Anaheim, CA 92801, phone: (714) 778-8624 or (800) 422-4739 (in California only).

Genetic Counselor: The medical genetics unit of Westchester County Medical Center has an immediate opening for a full-time genetic counselor. A master's degree in human genetics/genetic counseling and board eligibility or certification by the American Board of Medical Genetics are prerequisites. Westchester County Medical Center is affiliated with New York Medical College; the program offers comprehensive medical genetics services to the seven-county lower Hudson Valley region, comprised of approximately two million people. Excellent fringe benefits, salary commensurate with experience. Send resume to and contact: Linda Higgs, MS, Coordinator, Genetics Counseling Services, Medical Genetics Unit, Westchester County Medical Center, Valhalla, NY 10595, phone: (914) 347-7627.

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