PERSPECTIVES IN GENETIC COUNSELING

newsletter of the National Society of Genetic Counselors, Inc.

Vol 12, No. 4 Winter 1990/91

1991 MEETINGS MANAGER

FEBRUARY: AIDS CONFERENCE RESCHEDULED

The 6th Annual National Pediatric AIDS Conference has been postponed until February 9 - 12 to allow time for implementation of the Ryan White Comprehensive AIDS Resource Emergency Act of 1990 on Federal, State and Local levels. For information, contact Patricia A. Tompkins, RN, MS, Conference Chair, 202-328-0199.

MARCH: WOMEN IN PSYCHOLOGY TO MEET IN CONNECTICUT

"Women in the 90s: Unified in Diversity," the 16th National Conference of the Association for Women in Psychology, will be held March 7-10 at the Sheraton Hartford (CT) Hotel. This conference is open to professionals in a variety of psychology-related disciplines. It will focus on sharing perspectives and concerns related to issues of cultural differences, discrimination, women's health, aging, racism and employment. For more information, contact Joan C. Chrisler, Box 5578, Connecticut College, New London, CT 06320 or NSGC liaison to AWP, June Peters, 213-595-2000.

APRIL: BIOTECHNOLOGY AND THE DIAGNOSIS OF GENETIC DISEASE

Georgetown University School of Medicine will host a two-day conference in Washington, DC commencing April 18. This meeting is being endorsed by the NSGC. The conference will focus on policymaking implications of new biotechnologies at the federal level and will provide a forum for exchange of information related to scientific, clinical, economic and societal issues associated with the applications of biotechnology as they relate to the diagnosis of genetic disease. For more information, contact Susann Wilkinson, MPH, Project Director, Georgetown University School of Medicine, 202-687-5391.

JULY: MARCH OF DIMES CONFERENCE ANNOUNCED

The 23rd March of Dimes Clinical Genetics Conference, "Developmental and Genetic Disorders of the Central Nervous System," has been scheduled for July 7-10 in Vancouver, British Columbia, Canada. The conference will provide a comprehensive overview of recent advances in molecular, cellular, developmental, cytogenetic, biochemical and clinical genetics, as they apply to normal and abnormal central nervous system development and function. Information is available by calling March of Dimes Birth Defects Foundation, 914-428-7100.

OCTOBER: INTERNATIONAL CONGRESS OF HUMAN GENETICS

The 8th International Congress of Human Genetics will be held in Washington, DC, October 6 - 11. The NSGC will not schedule a self-contained Annual Education Conference. However, the NSGC's Annual Business Meeting, a dinner, will be held for NSGC members and guests on Monday, October 7 at the Ramada Renaissance. The Ramada has also been designated as the "official" hotel for genetic counselors. Program, registration and fee information will be mailed in February by the ICHG office.

LEARN YOUR LABS

by Andrew Faucett, M.S., Savannah Perinatology, Memorial Medical Center, Savannah, GA

Part One of a Two Part Series

Many genetic counselors are now working in private physicians' offices and smaller hospitals. As a result, they play an integral role in selecting genetic service laboratories. Most other counselors are directly affiliated with a major genetic center or private lab, and it is routine for them to use their own institution. However, as the rapid growth of DNA testing and the rise in the number of cytogenetic studies continues, some "full service" centers need to identify reference and "overflow" laboratories. In this two-part series, we will explore issues that have been raised when these choices need to be made. Part One will focus on cytogenetics and alpha-fetoprotein testing. Part Two, in the next issue, will focus on DNA and biochemical testing.

• continued on page 4 •

on the inside...

The NSGC gratefully acknowledges Integrated Genetics' support of this issue of *Perspectives*

FROM THE PERSPECTIVE OF...

Presidential Viewpoint Stresses Genetic Counselors as Educators; Researchers

Because today's technologies make genetic services relevant to greater numbers of people, and because delivery of genetic services is truly becoming a public health issue, the subject of the 1990 NSCG Annual Education Conference in Cincinnati, "Interface between Public Health and Clinical Genetics," was particularly appropriate. While there are many ways genetic counselors can have an impact on the genetic health care delivery system, I find education and research especially important.

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Send case reports, resources, materials and books for review to appropriate editors; address changes, subscription inquiries and advertisements to Executive Director; all manuscripts and correspondence to Editor-in-Chief.
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EDUCATION IS KEY TO FUTURE

Clearly, today's technical advances widen the gap between the lay and professional consumer and the genetics community. Education is essential at all levels to bridge this gap. As I see it, there is no one better suited to provide this education than genetic counselors.

Genetic education needs to begin in the junior and senior high classroom. Teachers need to learn how to incorporate genetic information into their science classes to create an informed public for the future.

Since health professionals are the first to see families with genetic disorders, ideally, *all* should have genetics incorporated into their basic training. Genetic counselors can help develop curricula for specific training and provide continuing education opportunities for health professionals in practice.

Of course, none of this can be accomplished without more genetic counselors. To meet that goal, new training programs must be developed. It is our responsibility to maintain a good applicant pool for these programs by promoting the profession to the colleges and universities in our regions.

RESEARCH NEEDS TO BEGIN IN OUR OWN BACKYARDS

It is imperative that genetic counselors become proactive in the vast potential for genetic counseling research. This will become increasingly important as genetic services become more widespread. I believe that genetic counselors are the most appropriate people to conduct this research, in the scope and in the direction it takes.

The future, indeed, holds much promise. It also holds much work.

Joan A. Scott, President

MEET YOUR BOARD

The list below includes the officers, committee chairs and regional representatives currently serving you. Column 3 indicates the dates their terms end.
Your Board has been elected or appointed to serve you. Please call on them to address issues related to their roles.

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The Art of Genetic Counseling

Case Report No. 23

When Reality Differs from Expectations

by Donna Blumenthal, Schneider Children's Hospital, Long Island, NY

Schneider Children's Hospital has a clinic for disorders of gender development, a joint venture of the Divisions of Genetics, Pediatric Endocrinology and Child/Adolescent Psychiatry. The staff consists of a medical geneticist, a genetic counselor, a pediatric endocrinologist, two psychiatrists, a psychiatric social worker and a pediatric urologist. Some of the staff attended a six month seminar on management of patients with gender disorders.

A.B., a 19-year-old white male, was referred to our clinic after presenting to his physician with a one-year history of malaise and decreased energy and libido. Physical examination revealed hypogonadism in an otherwise normal male. Facial and body hair distribution and body habitus were all normal male. The past medical history was unremarkable, as were developmental milestones. Laboratory tests revealed low testosterone levels. The karyotype was 46,XX. The referring physician described the patient as a bright, athletic college sophomore who had not yet been told of his chromosome status. However, his parents had been given the results.

Prior to the patient's visit, the team met to discuss the option of testosterone therapy, which requires biweekly or monthly IM injections. The patient attends an out-of-state college. We decided that the campus infirmary or a local, off-campus physician could be presented as options to the patient. We also discussed the best way and time to reveal his chromosome status and the issue of infertility to him without threatening his gender identity. We decided that A.B. should first meet with the psychiatrist, who would assess his psychological status and offer to accompany him to the physical examination by our endocrinologist.

Following that protocol, the psychiatrist reported that, contrary to what we had been told, A.B. knew of his chromosome status and had been told that he may have Klinefelter syndrome. A.B. had researched the syndrome himself and said he did not think this was his diagnosis. He also surmised that he must have "a piece of Y floating around somewhere" to be the way he is. Following our standard explanation of sexual differentiation, including the occurrence of XX males and XY females, A.B. reported that he felt secure as a male and noted that he had never been questioned about his gender based on his appearance. His main concerns were whether or not to receive testosterone and whether he would be fertile. The psychiatrist felt that the patient was handling this unusual chromosomal diagnosis extremely well, and that his masculinity did not appear to be threatened. The patient declined Y probe analysis because he felt further confirmation of his gender was unnecessary.

The geneticist and I met with the parents while the patient was being examined. We asked them to describe their feelings upon first receiving the chromosome results and to compare those feelings with their current ones. The patient's mother revealed that she was upset initially but has accepted the diagnosis, largely due to her son's attitude. The father claimed to have accepted it from the onset. The mother then asked for our opinion about the option of freezing her husband's sperm for purposes of artificial insemination when the patient is ready to start a family. It was their son's idea, and they were in favor of it. We supported this as a viable option. When they had raised this issue with the referring physician, she had responded that it was bizarre and possibly incestuous.

A.B. and the endocrinologist then joined us to conclude the session. The patient requested a referral to an adult urologist. He also chose to undergo testosterone therapy, to be administered at the school infirmary. He voluntarily expressed his idea to freeze his father's sperm and received everyone's endorsement. Since that visit, he has had at least three testosterone injections. He claims his symptoms have subsided.

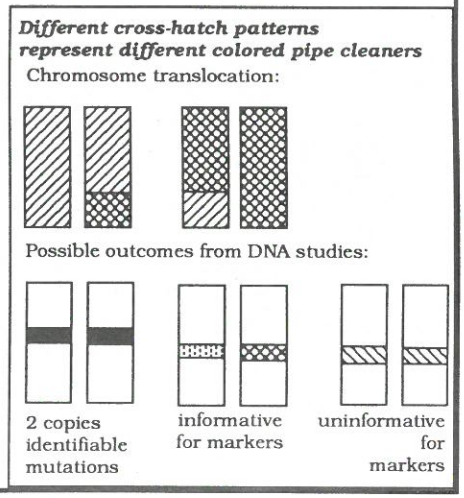
This patient presented quite differently than we had anticipated. Our words and actions concurred with and supported his own concept of himself as a normal male, in control of his life and his medical options.

Tips of the Trade

Pipe Cleaner Chromosomes as a Teaching Tool

Colored pipe cleaners, available in craft and notion stores, are an inexpensive and invaluable aid for demonstrating abstract genetic concepts. Different colored pipe cleaners, cut and "rearranged" to demonstrate crossovers and inversion, can be used to represent different chromosomes. Because they bend, unequal crossovers and inversion loops can be easily demonstrated. The "chromosomes" can be sent home, thereby functioning as a take-home, three-dimensional learning tool. Small pieces of different colored pipe cleaners can be twisted around the "chromosome" to represent alleles, genes, RFLPs or mutations. This is especially useful when a variety of difficult concepts needs to be discussed.

Patricia Himes, M.S., Oregon Health Sciences Univ Child Development and Rehabilitation Center, Portland, OR



Part I: Do You Know and Trust Your Lab?

continued from p. 1

Getting Started

When I moved to Savannah, I was unsure of the quality of the laboratory services that would be a major component of patient care. I knew the quality of the counseling, but questioned whether or not I could trust the results from the genetics reference laboratory. Over several months, I developed a survey to evaluate which laboratories would service my patients and my own needs best.

Facing Some Realities

The majority of genetics-based testing does *not* occur in a pathology lab. As a result, it does not fall under the established scrutiny of frequent laboratory inspections and proficiency tests. Only this year have the American Society of Human Genetics and the College of American Pathologists (CAP) begun discussions regarding the qualifications of cytogenetics laboratories.

Some CORN regions and states have developed their own cytogenetics proficiency tests. However, when developing general baseline standards, the laboratory inspections may not be as stringent as genetic counselors would prefer. Additionally, the level and quality of testing have to be evaluated against health care costs and the amount of information required.

Keys to Assessing Labs

Three general areas were important to me when I began the process of evaluating laboratories.

- Quality control (QC) and quality assurance (QA),
- The procedure or protocol used to provide the most useful and relevant information, and
- Service.

Questions about QC/QA

Standard questions in this category should include:

- What precautions are taken to reduce sample mix up, bad lots of media, technician errors or incubator problems?
- For MSAFPs, are all high *and* low values repeated for confirmation? Also, how accurate is the assay used on the low end and is it a special assay for low values?
- What are the routine QC checks performed in the daily assays... monthly, etc?
- What certifications do the laboratories maintain?

Policies, Procedures and Protocols Need Careful Examination

Standard questions in these areas should include:

- What is the standard and special case levels of banding?
- What number of cells are counted and what number of karyotypes are produced?
- Are some follow-up tests (ACHE, fetal hemoglobin, special stains) automatic or are they done only when requested and additional charges approved?
- Are confirmation studies requested on abnormals and at what price?
- What cell counts are routine for various unusual scenarios and what protocols occur with a mosaic or non-mosaic marker or a structural rearrangement?
- For MSAFP, how are the MOMs calculated and data developed? What is the percent of abnormalities? What parameters are used and what type of dating changes are required for recalculation.

Addressing Service Needs

- Are abnormal results reported directly to the genetic counselor?
- What is the availability of a PhD cytogeneticist or clinical geneticist in that loop?
- What is the turnaround time?... very short *or* very long times *both* raised concerns.
- What is the set-up frequency of MSAFP, AF-AFP, ACHE assays?
- Are active cultures maintained on all cytogenetic studies after reporting and for how long?
- Are there toll free numbers?
- Are educational materials available?
- Are shipping costs included?
- Is there a mechanism for acceptance of emergency specimen?
- Are mechanisms available to subsidize indigent patients?
- Is a pricing structure based on volume available?

In Conclusion

After reviewing all the information, I obtained input from my Center's two perinatologists before making the final decision. As the professionals who are responsible for communicating and explaining the results to both the patient and the referring obstetrician, it is imperative that we have confidence in the technical expertise of the laboratories we choose. While genetic counselors will weigh their priorities differently when evaluating labs, all three components are essential ingredients in the equation.

To obtain a copy of the questionnaire, contact: Andrew Faucett, M.S., Savannah Perinatology, 4750 Waters Ave, Suite 202, Savannah, GA 31404; 912-351-5971.

NSGC ...What's in it for me...

"One of the benefits of belonging to the National Society of Genetic Counselors is the opportunity to be listed in our annual membership directory. As an independent genetic counselor, I work in as many as four locations each week. Since a traditional table-top card file is too bulky to carry, my NSGC directory serves as my most complete and current resource of our membership. It enables me to contact my colleagues for information and support, refer patients or extended family members to a genetic counselor in another part of the country, and reach those colleagues working outside traditional medical center locations. I would encourage all individuals working in the field to join NSGC...and be listed in the directory. You never know who may be trying to reach you and why!"

Beth Balkite, M.S.
Chair, Membership Committee

Courts to Consider Petition in Frozen Embryo Case

To the NSGC Membership:

I am writing to update you on developments in *Davis v. Davis*, the frozen embryo case in which we just won a significant victory in the Tennessee Court of Appeals. The Tennessee Supreme Court last week accepted a petition by Ms. Davis (now known as Mary Stowe) to reconsider the Court of Appeals' decision awarding the parties "joint control" over the embryos.

The Supreme Court's willingness to address the *Davis* dispute raises great opportunities, and some risks as well. On the positive side, we now have a chance for a ruling explicitly stating that embryos will not be accorded legal rights under the law. On the other hand, the Tennessee Supreme Court could reinstate the trial court determination that "life begins at conception," a ruling which we worked so hard (and so successfully) to defeat.

Given these opportunities and risks, the need for your continued participation is greater than ever. We are currently formulating an *amicus curiae* brief to the Tennessee Supreme Court, and assume your organization will continue its support as *amici*. We will forward a final draft for your approval by mid February.

Thank you for your support.

Janet Benshoof, Director
and David L. Goldberg,
Cooperating Attorney
American Civil Liberties Union Fdt
Reproductive Freedom Project

Note: NSGC's Social Action Committee has been following this and other legislation related to the genetic counseling profession. To learn more about the activities of this committee, contact Shane Palmer, Chair, Social Issues Committee, 407-738-0448; or Trish Magyari, Legislative Liaison, 301-588-5484.

Active involvement in legislative issues is one way the NSGC helps you and helps the profession.

Medical Ethicist Responds to Issue of Non-Directiveness in Genetic Counseling Setting

To the Editor:

Even before reading the Fall 1990 issue of *Perspectives in Genetic Counseling* (Vol. 12, No. 3), I was troubled by what I call the "radical non-directive" approach in clinical ethics. In genetic counseling this "radical" approach finds expression in the profession's insistence that counselors not disclose their opinions, views or personal decision making processes to their patients. Odd as this may sound, non-directiveness so understood actually seems antithetical to the concepts of patients' rights and patient autonomy that it is supposed to affirm.

Virtually everything that one reads about professional/patient relationships in the clinical setting extols the virtues of respecting patient autonomy (and self direction) and inveighs against directiveness (or paternalism). In fact, contemporary medical ethics has convinced medical practitioners that the patients' rights/autonomy model of patient interaction is *incompatible with* any form of directiveness — that paternalism in any form is to be avoided. I have argued elsewhere that this so-called incompatibility leads us to misdescribe important questions in clinical ethics and may actually disserve patients' best interests.[1]

As Polzin and others point out, one of the basic tenets in medicine is *primum non nocere* (above all, do no harm). That same tenet can also be said to require that we help others further what they take to be in their own best interests. Self-interest and self-direction are clear examples of important and legitimate interests of patients. We might even say that as persons they are the *most* important and legitimate interests patients have and as such, should be *furthered* by clinical geneticists.

It might also be that a patient could reasonably choose that self-direction is *not* in her best interest. Surely one can acknowledge, for example, that there are instances where self-direction or autonomy serves a patient's best interests and other cases where it does not. Surely one can even acknowledge that failing to provide direction may in some instances harm a patient.

So, while it is true that "it is never possible to completely gauge how a couple will view and use the response," if a counselor answers the question, "What would you do if you were in my place?", and while it is true that "no amount of prior disclaimer can completely remove the possibility of the client giving undue weight" to the counselor's imagined choice (Polzin), it seems counterintuitive to believe that considering the counselor's own choice or decision-making process is not sometimes both instructive and important... not to mention in a patient's best interests.

Some patients' choices can be better informed by learning how a genetic counselor would think through options. In fact, too strong a reliance on non-directiveness, as traditionally conceived, can approach abandoning one's patients. Part of C.K.'s anxiety lay in her desire to have all the desiderata before her, in her case, knowing how her counselor would work through such a decision was an important piece of the puzzle. Conceived of in this way, so-called directiveness can sometimes actually enhance patient autonomy, and rational, self-directing patients have been thankful for such direction.

Not only does such an approach "demystify" the right choice, it affirms the professional/patient *relationship* in which counselors and patients find themselves. It acknowledges a more pliable (perhaps more female) model of decision-making — one where the agony of decision can be shared. To deny patients that option is directive in its worst sense.

Nora Kizer Bell, Ph.D.
Professor and Chair, Department of Philosophy
University of South Carolina School of Medicine, Columbia, SC

[1] Bell, NK. Patient's best interest. *Medical Ethics for the Practicing Physician.* February 1990.

Point CounterPoint: A Genetic Counselor

Dear Readers:
On these pages, Seth Marcus has challenged us to defend our rationale for practicing in a certain manner. He asks us to evaluate the way "everyone does it" and provide reasoned (and reasonable!) arguments to support the way we provide genetic counseling to our patients. This section has provided dialogue, and though the opinions expressed may not represent those opinions of the general membership, they allow us to reflect and to grow.
Seth is developing additional topics for future issues, and he invites your participation. Members can contact him if they wish to defend a particular side of an issue or if they have ideas for discussion. Some topics under current consideration are:

- *The merits of obtaining a pedigree for "routine" amnio*
- *The benefits of certification*
- *The availability of CVS at the patient's discretion (i.e. ≤35yo)*

Do you have any ideas of your own? Why not call Seth and pass them along?

Vickie Venne, M.S.

Look to Colleagues in Other Health Professions to Help Meet the Needs of Underserved Populations

by Bonnie S. LeRoy, M.S., Director, Genetic Counseling Program, University of Minnesota

I propose that we look to our existing colleagues in the health care system to assist in providing genetic counseling and education.

In 1989, a group of people met at the Asilomar Conference Center to discuss issues pertaining to the education of genetic counselors. As a program director, I participated in that meeting. Two of the issues we grappled with were the shortage of genetic counselors and the needs of underserved populations. One proposal to meet these needs was the training and use of "single gene counselors" who did not have graduate training. There are potential long term problems with this solution in our still evolving profession.

"There are potential long term problems with (the use of single gene counselors) in our profession."

An additional group of individuals involved in the delivery of genetic care may continue to blur the lines of responsibility. In a field that is about 20 years old, many of us still regularly respond to the question, "What does a genetic counselor do?" That question is asked by both the public and, more importantly, by other health care professionals. Until there is widespread understanding of the role of the genetic counselor, are we not diluting our role if additional professionals with similar titles perform some, but not all of the tasks associated with genetic counselors?

With the blurred lines of responsibility come blurred financial rewards. Genetic counseling services have traditionally not supported themselves. In a budget-conscious era, health care administrators usually look at finances rather than education when cutting positions and benefits such as support for travel and membership dues. This will be especially true if job titles appear similar.

Genetic counselors do not currently have a defined career ladder. In large settings, counselors with seniority may become super-

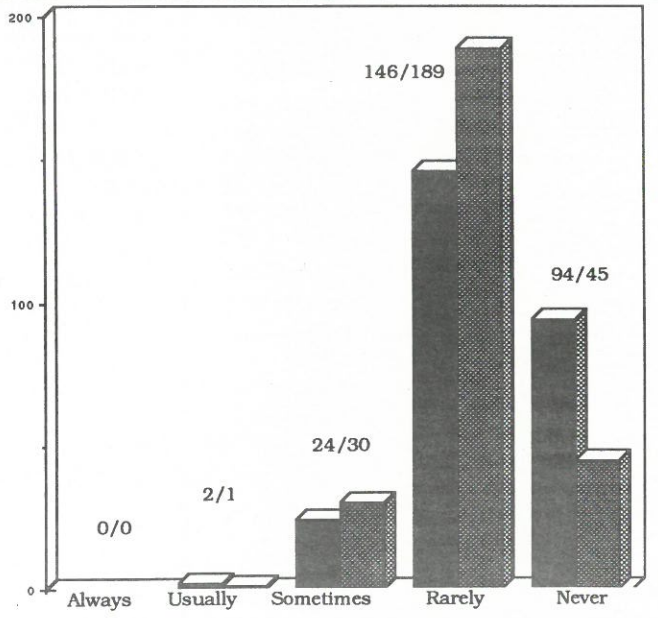
Survey Results: Membership Speaks Out on Issue of Counseling Directiveness

The graph to the right represents the results of the opinion poll conducted in concert with Point CounterPoint in *Perspectives in Genetic Counseling*, Vol. 12, No. 3, Fall 1990. The topic, *Can Non-Directiveness be Non-Helpful?*, was presented by three colleagues.

- Question One, (Column One): "When genetic counselors are asked by a patient, 'What would you do if you were in my place?' should genetic counselors state what they would do?", received 266 responses.
- Question Two, (Column Two): In a genetic counseling session, are you, as a genetic counselor, directive?" received 265 responses.

Thanks to all of you who took a few moments from your busy schedules to answer the survey. It certainly has provided for interesting dialogue that we suspect will continue well beyond the boundaries of *Perspectives*.

Seth Marcus, M.S.



By Any Other Name...

visors or department administrators. It has been argued that the ability to supervise "single gene counselors" might provide one component of a career ladder. However, "single gene counselors" are needed most in areas with minimal services, usually where genetic counselors are not currently employed.

At the open discussion of this topic during the NSGC meeting in Cincinnati, I was pleased to hear about the many creative ways genetic counselors across the country are dealing with the shortage in their areas. At our large cystic fibrosis clinic, I have found the nursing staff eager to help with the genetic needs of the patients. Our nurses play a large role in the care of CF patients and have a well established relationship with families. They are the obvious professionals to provide education for routine carrier testing. Benefits of cross-training include expanding the nurse's role with families and improving the genetic counselor's awareness about routine management of CF patients. I become a more integral part of the clinic, and the nurses become more knowledgeable about the molecular genetics of CF testing.

These experiences have led me to believe that we could learn from clinic and public health nurses and medical social workers while we provide training that would expand their role with patients and families in the area of genetics. Our colleagues in other health professions have much to teach us, and patients can benefit from a team approach to providing genetic care rather than the training of nonmedical personnel to provide only a very narrow service.

This approach will keep our professional standards high, allow more health care professionals to understand the unique abilities of the trained genetic counselor and ensure that our field will continue to grow as high quality students are attracted to the profession.

Single Gene Counselors Enhance Genetic Services

by Susan Fortune Pinheiro, M.S., Coordinator,
Sickle Cell Counselor Training Program, Children's Hospital Oakland, CA

The current shortage of genetic counselors can be attributed to the lack of cultural diversity among existing genetic counselors and the reluctance of genetic counselors to relocate to meet minority or rural needs. Single disorder counselors can provide valuable assistance by addressing these increased counseling needs.

As research continues to uncover the genetic links of common hereditary conditions, the shortage of genetic counselors is likely to continue. Many jobs remain vacant for years while only about 100 genetic counselors graduate from certified programs each year. Training programs are unable to expand because of the limited placements for clinical training opportunities.

The lack of cultural diversity among genetic counselors also poses problems. The vast majority of genetic counselors are Caucasian. Few are bilingual, making it difficult for them to provide adequate services to diverse ethnic groups. Training health educators and other professionals to counsel families about one particular disease would help reach families who currently go without services. Single disorder counselors familiar with a client's language and/or cultural myths about disease can be an integral component of the health care team. They can facilitate trust in a white dominated medical system, where minorities traditionally underutilize services.[1]

Many genetic counseling positions that remain vacant are those serving minorities or rural areas. The fact that genetic counselors have the freedom to choose among numerous positions in varied locations does not negate the genetic needs of those underserved populations. If counselors are unwilling to accept those positions, single gene counselor alternatives might be reasonable.

The California State Department of Health Services has developed a Sickle Cell Educator/Counselor Training Program to address the shortage of master's level counselors. A panel of genetics, medical, public health, education and public policy experts developed a rigorous program to train sickle cell educators and counselors. Two levels of certification were developed to reflect the service needs for the patients and to acknowledge varying knowledge and job responsibilities. The panel balanced the need for adequate training against limited resources. As a result, sickle cell counselors now provide the bulk of the education and counseling efforts for the 10,000 babies born annually in California with a hemoglobin trait.

A recent Children's Hospital Oakland study compared knowledge about sickle cell trait of Sickle Cell Educator/Counselor Training Program graduates to other health professionals. The study concluded that graduates from the program had a greater knowledge about sickle cell trait than genetic counselors and physicians, despite considerably less formal education. Although the study was not designed to assess counseling skills, the study also revealed that single disorder counselors have the knowledge to counsel effectively and make appropriate referrals to genetic counselors.

To assure quality services, basic standards should be adopted, including minimum education prerequisites, training requirements and built in follow-up supervision provided by a genetic counselor and a physician. The ultimate goal is national examination and certification.

Single disorder counselors offer one practical solution to the demand for counselors. As they contribute substantially to a genetics team, the role of the master's level genetic counselor will also change. Genetic counselors will need to accept greater responsibility for supervision, training and single disorder program development, which can lead to additional advancement opportunities and greater job satisfaction for genetic counselors.

1 Harwood A (ed) *Ethnicity and Medical Care.* (1981) Harvard University Press, Cambridge, Mass.

2 Fine B, Gettig B et al, *Strategies in Genetic Counseling: Reproductive Genetics & New Technologies,* March of Dimes Birth Defects Fdt Original Article Series, V 26:3, 1990

BULLETIN BOARD

CALL FOR NOMINATIONS

The leadership of the NSGC is largely determined by you, our membership. The 1991 nominating committee, chaired by Past President I Barbara Bowles Biesecker, also includes: Andrea Gainey, Alison Warner, Diana Punales-Morejon and Rhonda Schonberg. Nominations are being accepted for the following positions: President-Elect, Treasurer and Representatives to Regions I, III and V. Send your nominations to: Andrea Gainey, 6140 N. Calle de la Culebra, Tucson, AZ 85718.

PIC TO COMPARE AND CONTRAST SALARIES; JOB DESCRIPTIONS

The Professional Issues Committee is planning to conduct a salary inquiry of professionals in related health care fields. The Committee will also assemble a notebook of job descriptions for genetic counselors, documenting specific responsibilities held by counselors in a variety of positions. This will become a resource for genetic counselors.

If you are interested in working on either project *or* if you have your job description to contribute, contact Wendy R. Uhlmann, 313-745-7066.

SPEAK OUT: LONG RANGE PLANNING COMMITTEE APPOINTED

The *ad hoc* Long Range Planning Committee is soliciting ideas about future goals, issues and directions for the NSGC over the next five to ten years. Members of the committee are Debra Collins, Ginny Corson, Mimi Rietsch-Donnelly, Andrea Fishbach, Denise Greene, Ann Happ and Trish Magyari. Send your thoughts or comments by February15 to: Ginny Corson, CMSC 1001, John Hopkins Hospital, 601 N. Broadway, Baltimore, MD 21205.

HEALTHY PEOPLE 2000

Genetic counselors involved in writing or reviewing grants might benefit from reading *Healthy People 2000: National Health Promotion and Disease Prevention Objectives.* This three-year national effort outlines the health issues which our Federal government deems the most important. Copies of this comprehensive directive are available for $31. by ordering Stock #017-001-00474-0 from Superintendent of Documents, Government Printing Office, Washington, DC 20402-9325.

NEW IMMIGRATION RAISES CROSS CULTURAL ISSUES

With the growing number of Russian immigrants who have relocated to the U.S., there is a growing need for counseling materials, aids and understanding of cross cultural issues, particularly related to Tay Sachs screening and counseling. Genetic counselors who have developed materials or have successfully overcome cultural barriers in Russian populations are invited to contact Karen Heller, Childrens Medical Center, 1935 Motor St, Dallas, TX 75235; 214-920-2357.

FEEDBACK FOR ETHICS CODE REQUESTED

The *ad hoc* Committee on Ethical Codes and Principles is currently accepting membership feedback on the draft of the proposed NSGC Code of Ethics, which was mailed to all members in September. Please send comments to Judith Benkendorf, by mail c/o Dept. OB/GYN, Genetics, Georgetown University Medical Center, 3800 Reservoir Rd NW, Washington, DC 20007, by FAX 202-687-7752, or by phone 202-687-8810, no later than January 31.

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BITS 'N PIECES FROM YOUR EXECUTIVE OFFICE

- Dues invoices for FY1991 were mailed in early December. Please help us serve you better. Respond by the January 31 deadline.
- Special Projects Fund applications will be accepted through May 15.
- Revised Career Packets for your Career-oriented presentations are now available. Please specify College or High School level.
- NSGC's 1989 Annual Report of Activities, distributed to all registrants at the 10th Annual Education Conference in Cincinnati, is now available upon request.

• REFERENCE •

The Family Genetic Sourcebook
Author: Benjamin A. Pierce
Publisher: John Wiley & Sons, Inc. 1990
Price: $14.95 pbk, 340 pp
Reviewed by: Karen Brookhyser, M.S., UC Davis, Sacramento, CA

The Family Genetic Sourcebook is an excellent resource for both the layperson interested in human heredity and for health care professional dealing with topics in genetics. The book does not intimidate the layperson who is attempting to understand how genes influence the individual and family, yet it provides enough detail to give professionals a background for discussing current topics. This balance is difficult to achieve in the rapidly-evolving discipline of genetics, but the *Sourcebook* strives to this goal.

The book has three main parts. The first addresses genetic principles such as definitions, inheritance patterns, family pedigree construction and genetic counseling. The second part is an alphabetical "Catalog of Genetic Traits," which describes disorders and discusses the heredity of more than 100 traits from Achondroplasia to Wilson's disease. The third consists of three Appendices, including Genetic Service Centers in the U.S. by state. (Caution: some are not current.) The second lists "State Genetic Services Coordinators," and the third provides addresses of eight national family support organizations. A glossary and index are also included.

This book is easy to read, with many examples of common traits such as hair color, blood type and ear wax. Definitions, vocabulary and ideas are repeated; cross-references are used. Some entertaining and useful analogies are included.

The *Sourcebook* may be too sophisticated for some of our patients, but it could help them clarify their questions before a genetics appointment.

The scope of genetic topics is wide. For the most part, rare or unusual disorders are avoided. The author presents complex ideas such as meiosis, heritability, the genome, genetic heterogeneity and empiric risks in a concise, understanding manner. These descriptions should be a useful outline for answers to questions that we often hear.

Chapter six discusses genetic counseling in a positive tone, adhering to the broad "communication process" definition adopted by the American Society of Human Genetics. Disclaimers throughout the book emphasize the variable nature of genetic disorders and caution that the book should not be used as a substitute for professional genetic counseling.

Dr. Pierce is Associate Professor of Biology at Baylor University and the book has a distinctly scientific rather than psychosocial emphasis. Therefore, this book would not be an appropriate reference for families or individuals seeking psychological support. The *Sourcebook* does not discuss arguments surrounding controversial issues in genetics.

I do have criticisms of the *Sourcebook*. Screening and diagnostic tests such as maternal serum alpha-fetoprotein could have been discussed more clearly in the section on neural tube defects. In some cases, terminology is disturbing or inaccurate. Readers maybe offended to see Lesch Nyhan Syndrome described as a "bizarre" genetic disorder. A table describing chromosome variations has a poor description of Turner syndrome, although it is more accurately described later in the book. The same table lists a "higher rate of criminality" as a symptom of males who have an XYY karyotype.

Amniocentesis performed before the 16th week of gestation is not mentioned, and the description of a couple contemplating amniocentesis is not presented as a good example of nondirective genetic counseling. Throughout the book, risks are interpreted as "small" or "low" instead of stating the numerical risk. Although annoying, I do not believe that these problems negate the general usefulness of the *Sourcebook*.

In summary, the *Family Genetic Sourcebook* is a good introduction to human genetics. It addresses many topics in our field that are encountered in the popular press. It can also be a useful resource for nurses, social workers, doctors and other professionals seeking simple and fairly accurate descriptions of the concepts in genetics.

• ORGANIZATION •

The American Self-Help Clearinghouse is a national organization encompassing resources for a range of self-help groups worldwide. Outreach ranges from addictions and compulsions; bereavement and loss; disabilities; physical and mental health; parenting and family; and numerous others, including many genetic-related groups. Names and numbers are maintained on a computerized database and are regularly updated.

This 174 page, comprehensive national guide includes updated contacts and descriptions for over 600 national and model self help groups plus over 100 national toll-free helplines.

A copy of *The Self-Help Sourcebook*, last updated in September 1990, is available by contacting Self-Help Clearinghouse, Attn: Sourcebook, St. Clares-Riverside Medical Center, Denville, NJ 07834. Please send $10 for each copy.

• More Resources on Next Page •

• BOOK BAG •

Syndromes of the Head and Neck
by Robert J. Gorlin, M. Michael Cohen, L. Stefan Levin, 3rd ed., 1990
reviewed by Elsa Reich, M.S., New York University School of Medicine

Reviewing this classic reference text is like seeing an old friend become wiser and more beautiful with age. This is the third edition of a text that has been a standby of clinical geneticists, genetic counselors, plastic surgeons and other specialists for almost 20 years. The expansion of this book to its present size and new format mirrors the explosion in genetic knowledge over the last 20 years. Those who are unfamiliar with the text are in for a treat. This is an encyclopedic work, describing in depth some 700 syndromes, all of which have recognizable dysmorphic facial features. As Drs. Gorlin and Cohen state in their preface, the book describes numerous conditions affecting multiple organ systems, but to maintain the identity of the book, the authors have retained its original title. Both of these authors, along with collaborator Dr. L Stefan Levin, are experts in the identification of craniofacial disorders. However, due to the number of syndromes that have been newly identified since last edition in 1976, they invited 18 other specialists to contribute. This collaboration has resulted in a resource excelling in breadth as well as depth.

The format of the book has been changed. The conditions are no longer arranged alphabetically, but are placed in categories that recognize similar etiologic, developed or anatomical abnormalities. There are categories such as chromosomal syndromes, teratogenic syndromes, syndromes with craniosynostosis and first and second bronchial arch syndromes. An arbitrary categorization has sufficed when a condition could easily be listed under more than one heading. The descriptions of the conditions are quite detailed and always include one or more pictures of affected individuals. Those pictures alone make the book worthwhile. In addition, the bibliographies are extensive and have been expanded since the last edition.

Comparison of the sections on hemifacial microsomia/Goldenhaar syndrome from the 1976 edition and this recent one provides a good example of how the content has grown. Not only is there more extensive descriptive text in the new edition, but there are also several additional photographs, including radiographs and close-ups of several anomalies. The bibliography has grown from 44 to more than 100.

The quality of a reference text is in part dependent on the quality of the index, and it appears that this is one quite complete. There is a real attempt to provide a list of syndromes in which specific abnormalities are found, so that if one looks in the index for the symptoms of the unknown condition, one has a good chance of identifying the syndrome.

Syndromes of the Head and Neck is a must for any clinical genetics unit and will probably be the first text to reference when diagnosis of a dysmorphic individual is uncertain. In addition, there is valuable information to be gleaned from the text and the bibliography to enhance understanding of a known condition. For those medical geneticists and genetic counselors whose first love is dysmorphology, this new edition of *Syndromes* will surely be your bible.

• DISORDER RELATED RESOURCE •

Living with Difference: Families with Dwarf Children
author: Joan Ablon Ph.D.
publisher: Praegar Publishers, One Madison Ave, NY, NY, 10010
reviewed by: Mimi Rietsch-Donnelly, second year student, Sarah Lawrence College

In her book, *Living with Difference*, Joan Ablon leads her readers into the lives of parents of dwarf children. Ablon uses the interviews of a subset of six couples who were followed between 1977 and 1983 as her main source of information. By using this technique, the author provides the reader with a first hand account of various topics of family life including, but not limited to, the initial response to the birth of their dwarf child and aspects of family life associated with the presence of a dwarf child as family member. The success of the book lies in Ablon's use of parental interviews, although the text provides only a narrow view of parental thoughts, feeling and attitudes because those interviewed are from a selected subgroup.

The parents interviewed appear well-adjusted to their situation, often becoming their child's advocate and often stating that coping with their child made them more sensitive and understanding toward other people.

Expressed negativism about their dwarf children and their children's effect on marital relationships, family structure and family planning were conspicuously missing, an omission which could lead a reader to believe that these feelings do not exist.

In part, the parents interviewed may appear better adjusted than one would expect of a general population of families with dwarf children because of a bias in Ablon's selection of couples. All the couples interviewed belong to a parent's group of Little People of America (LPA), an organization established to meet the supportive and social needs of dwarfs and their families. Belonging to such an organization may make parents more inclined to adopt a positive attitude than those who are not involved in the support group. A more objective treatment of the topic would have included interviews of parents who did not participate in a support organization such as LPA.

While Ablon employs the powerful technique of parental interviewing, the text has an inherent bias toward parental optimism of which any potential reader should be cognizant before reading this book.

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FRESNO, CA: Immediate opening for Administrative Director of Medical Genetics. Minimum 3 years health care experience. Masters degree, business experience preferred.
Responsibilities: Plan, direct, coordinate all aspects of medical genetics and prenatal counseling.
Contact: Joan Ishimoto, Dept. Human Resources, Valley Childrens Hospital, 3151 N. Millbrook, Fresno, CA 93703; 209-225-3000 x 1143. EOE/AA.

LOS ANGELES, CA: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Opportunity for independence and development in growing, challenging program. Prenatal counseling for amnio; coordinate pediatric clinic and inpatient consults; large satellite clinic base. Currently developing CVS service.
Contact: Rosetta W. Hassan, MD, King/Drew Medical Center, 12021 S. Wilmington Ave, Dept. OB/GYN, Los Angeles, CA 90059; 213-603-4638. EOE/AA.

LOS ANGELES, CA: Immediate opening for BC/BE Genetic Counselor with Masters in GC or Nursing.
Responsibilities: Join large diverse program: counseling and case management as well as involvement in administrative, research and educational aspects of prenatal and pediatric genetics.
Contact: Bill Herbert, MS or Ann Garber, DrPH, Cedars-Sinai Medical Center, Medical Genetics and Birth Defects Center, 444 S. San Vicente Blvd, Ste 1002, Los Angeles, CA 90048; 213-855-2214. EOE/AA.

OAKLAND, CA: June 1991 opening for BC/BE Genetic Counselor. (Full time thru Dec; Permanent Part time from Dec)
Responsibilities: Join multidisciplinary team in hemoglobinopathy/coagulation disorders unit; occasional hemoglobin trait and prenatal referrals; community and professional education; research possibilities.
Contact: Susan F. Pinheiro, MS, Childrens Hospital Oakland, 747 52nd St, Hematology Dept, Oakland, CA 94609; 415-428-3167. EOE/AA.

SACRAMENTO, CA: Immediate opening for BC/BE Genetic Counselor. Part time with good potential for Full time.
Responsibilities: Join active, comprehensive team in primarily prenatal position: CVS, amnio, PUBS, diagnostic U/sound.
Contact: Douglas Hershey, MD, Sutter Perinatal Center, 5275 F St, Sacramento, CA 95819; 916-733-1750. EOE/AA.

SEPULVEDA, CA: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Join interdisciplinary team with consultations from Childrens, UCLA & USC; coordinate and participate in diagnostic prenatal services, genetic counseling and consultation; professional and community education and outreach; liaison to genetic clinics.
Contact: Jaime Mejlsenkier, MD, Chief of Health Services, North Los Angeles County Regional Center, 8353 Sepulveda Blvd, Sepulveda, CA 91343; 818-891-0920 x315. EOE/AA.

DENVER, CO: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Prenatal counseling and follow up for fetal structural abnormalities, AMA, MSAFP screening and teratogens; report directly to chief OB.
Contact: Roger Lenke, MD, University Hospital, 4200 E 9th Ave, Dept OB/GYN, Campus Box B198, Denver, CO 80262; 303-270-8982. EOE/AA.

PEORIA, IL: Immediate opening for BC/BE Genetic Counselor with Masters degree in medical genetics or related field.
Responsibilities: Coordinate on-site and satellite pediatrics clinics for Regional Genetics Program, including CVS, amnio; inpatient consults at Level II tertiary care hospital; professional and consumer education.
Contact: William H. Albers, MD, Dept. Pediatrics, University Illinois College of Medicine at Peoria, Box 1649, Peoria, IL 61656; 309-655-2570. EOE/AA.

LEXINGTON, KY: Immediate opening for BC/BE Genetic Counselor. Potential for Faculty Position.
Responsibilities: Join active team in expanding program: preconceptual and prenatal counseling for MSAFP, amnio, teratology, malformation counseling; outreach to professional community; some teaching.
Contact: Anjana Pettigrew, MD, University of Lexington Medical School, Dept. Pathology, Lexington, KY 40536-0093; 606-257-4089. EOE/AA.

SCARBOROUGH (PORTLAND), ME: Immediate opening for BC/BE Genetic Counselor. Experience preferred.
Responsibilities: Comprehensive regional prenatal and general services: large MSAFP (triple marker screen) and pregnancy loss evaluation/treatment programs in conjunction with Maine Medical Center; ongoing research and educational projects; OB/Pediatric resident education.
Contact: Richard Doherty, MD or Ed Kloza, MS, Foundation for Blood Research, P.O. Box 190, Scarborough, ME 04074; 207-883-4131. EOE/AA.

DETROIT, MI: Immediate opening for BC/BE Genetic Counselor. Part time to start, with possible growth to Full time.
Responsibilities: Coordinate and provide genetic counseling services at Wayne State University-affiliated hospital; MD, CF, craniofacial subspecialty clinics; participate in regional pediatric genetics clinics; opportunity for professional and community education.
Contact: Brawait Bawle, MD or Gillian Ingall, MS, Childrens Hospital of Michigan, 3901 Beaubien, Division Genetics & Metabolism, Detroit, MI 48201; 313-745-4513. EOE/AA.

DETROIT, MI: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Join active team in rapidly expanding large, diverse reproductive genetics center; wide range of ethnic/economic backgrounds: CVS, amnio, MSAFP, diagnostic U/sound; teratogen counseling; novel fetal therapy. Opportunity for research; publications.
Contact: Anne Greb, MS or Mark Evans, MD, Hutzel Hospital, 4707 St. Antoine Blvd, Dept. OB/GYN, Div. Reproductive Genetics, Detroit, MI 48201; 313-745-7067. EOE/AA.

ROYAL OAK, MI: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Flexible assignments according to counselor's interests in comprehensive prenatal counseling program for amnio, CVS, abnormal MSAFP, U/sound, teratogens and infections. Involvement in pediatric genetics and professional education.
Contact: Robert P. Lorenz, MD, William Beaumont Hospital Medical Bldg, 3535 W. 13 Mile Road, Ste 329, Royal Oak, MI 48073; 313-551-0395. EOE/AA.

CHAPEL HILL, NC: Immediate opening for BC/BE Genetic Counselor with Faculty Position.
Responsibilities: Join well-established prenatal diagnosis program: CVS, PUBS, hi-volume MS-hCG/AFP lab; database management; community education.
Contact: Beth Boyea, M.S., University North Carolina, Dept OB/GYN, CB#7570, Chapel Hill, NC 27599-7570; 919-966-2229. EOE/AA.

GREENVILLE, NC: Immediate opening for BC/BE Genetic Counselor with Faculty Position.
Responsibilities: Wide range of genetic counseling opportunities: pediatric, prenatal and specialty clinics; participate in satellite clinics.
Contact: Theodore Kushnick, M.D., East Carolina University School of Medicine, Brody Building 3E140, Greenville, NC 27828-4354; 919-551-2525. EOE/AA.

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National Society of
Genetic Counselors, Inc.
Executive Office
233 Canterbury Drive
Wallingford, PA 19086

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ELIZABETH, NJ: Immediate openings for BC/BE Genetic Counselors. Full time/Part time schedules available.
Responsibilities: Prenatal and pediatric counseling in urban medical center: amnio, MSAFP, teratogen counseling, family history and counseling. CF clinic and monthly pediatric genetics clinic. Opportunities available for professional and community education.
Contact: Judi A. Meseck, Employment Manager, Elizabeth General Medical Center, 925 E. Jersey St, Elizabeth, NJ 07201; 201-558-8153. EOE/AA.

BROOKLYN, NY: Immediate opening for Half time BC/BE Genetic Counselor.
Responsibilities: Newly forming genetic diagnostic unit: pre/postnatal counseling; liaison to genetic diagnostic lab; some administration; research and education opportunities available.
Contact: Rebecca Shiffman, MD, Methodist Hospital, 506 Sixth St, Dept OB/ Maternal Health, Brooklyn, NY 11215-9008; 718-780-3272. EOE/AA.

NEW YORK, NY: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Join large academic setting with comprehensive prenatal services, teratogen counseling, dysmorphology, pediatrics, MSAFP screening, extensive molecular/ biochemical lab suport. Special projects, research, other educational opportuniiesy available
Contact: Randi Zinberg, MS or Cindy Benson, MS, Dept. Pediatrics, Div. Medical & Molecular Genetics, Mt. Sinai School of Medicine, 5th Ave & 100th St, New York, NY 10029-6574; 212-241-6947. EOE/AA.

STATEN ISLAND, NY: Immediate opening for BC/BE Genetic Counselor. Full or part time, negotiable.
Responsibilities: Join active multidisciplinary genetics department with cytogenetics, biochemical & DNA labs; prenatal, pediatric and developmental disabilities counseling, research programs; professional & lay education.
Contact: Susan Sklower Brooks, MD, NY State Institute for Basic Research, 1050 Forest Hill Road, Staten Island, NY 10314; 718-494-5240. EOE/AA.

CLEVELAND, OH: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Serve as liaison between neighborhood centers, physicians and health care organizations in developing and presenting educational materials related to genetics; identify needs for services.
Contact: Patricia Saddle, Human Resources, Case Western Reserve University, 10900 Euclid, Cleveland, OH 44106; 216-368-4500. EOE/AA.

DALLAS, TX: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Full range of pediatric genetic patients. Primary responsibility for coordinating metabolic service; opportunity for developing and expanding research program at major medical center; interact with large, active genetics team.
Contact: Lewis Waber, MD, PhD, University Texas Southwestern Medical Center, 5323 Harry Hines Blvd, Div. Pediatric Genetics & Metabolism, Dallas, TX 75235; 214-688-8996/6796. EOE/AA.

CHARLOTTESVILLE, VA: Immediate opening fpr BC/BE Genetic Counselor.
Responsibilities: Join team of 4 Genetic Counselors & 4 MDs in predominant prenatal position: CVS, amnio, PUBS, AFP and teratology.
Contact: Rachel Baughman, MS, UVA Medical Center, Box 387, Dept. OB, Div. Genetics, Charlottesville, VA 22908; 804-924-2500. EOE/AA.

NORFOLK (AT VIRGINIA BEACH SATELLITE), VA: Immediate opening for BC/BE Genetic Counselor.
Responsibilities: Assume significant responsibilities for patient care and management: reproductive loss and preconceptual counseling; general, prenatal and teratogen counseling; opportunity for involvement in specialty and satellite clinics; professional and community outreach and education; potential for independent and collaborative research and publications exists.
Contact: Anna Newlin, MS, Eastern Virginia Medical School, Genetics Program, 700 W. Olney Road, Norfolk, VA 23507; 804-446-5723. EOE/AA.