PERSPECTIVES
in genetic counseling

Volume 15:4 Winter 1993/94

New Inheritance Patterns; New Counseling Dilemma

Jack Tarleton, PhD, Greenwood Genetics Center, Greenwood SC

Classical genetics describes a biological world where there are a few simple rules of inheritance. Mendelian laws provide genetics with a paradigm for understanding human genetic diseases as a consequence of the assortment of chromosomes and/or the segregation of mutant alleles — a paradigm which sufficed quite well until recently.

During the last few years, it has become apparent that all is not well with the paradigm — there appear to be a number of non-traditional modes of inheritance and exceptions to the rules of Mendelian genetics. Examples include somatic cell and germline mosaicism, transposable elements, mitochondrial inheritance, heritable unstable elements and uniparental disomy. The understanding of these non-Mendelian mechanisms continues to add information which has to be mastered by genetic professionals to provide the best explanations available for their patients. The two cases presented in this issue, with differing information given to the involved couples, highlight the evolving understanding of uniparental disomy (UPD) and illustrate a number of counseling issues.

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Your Board In Action

Nancy Callanan, MS, Univ North Carolina at Chapel Hill

The Fall Board of Directors meeting traditionally introduces and orients new board members. Regional representatives, committee chairs, editors and officers report their activities and discuss their goals and objectives for the coming year.

This year, two visitors briefly joined the meeting. Vivian Wang, recipient of the 1992-93 Special Projects Fund, presented her *Handbook of Cross-Cultural Genetic Counseling.* José Cordero, director of the Birth Defects Branch, Center for Disease Control (CDC), spoke about CDC programs that focus on prevention of birth defects and developmental disabilities. (See page 6.)

At the previous board meeting, NSGC's strategic plan was developed. As reported in *Perspectives* (15:2) several issues were identified, and priorities and timelines were assigned to board members and committees. Betsy Gettig, as Past President I, has responsibility for overseeing the strategic planning process. Board members reported on projects aimed at meeting these goals:

- Regional representatives are discussing strategies for improving communication among members in each region.

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The leading voice, authority and advocate for the genetic counseling profession.

ON THE INSIDE

NSGC gratefully acknowledges Integrated Genetics for a grant in support of this newsletter.

Message from Your President

Since we formed a national society almost 15 years ago, the Boards, committees and regions of NSGC have worked to formulate and promulgate a strong professional identity for master's level genetic counselors. One has only to reread the first issues of *Perspectives in Genetic Counseling* to become pridefully aware of the growth in our 'perspective.' Our position within the clinical genetics community was much more tenuous than it is today; our perception of our role within that community was different than now.

Although professional issues will continue to evolve within the society and the field, now is the time for NSGC to take a more proactive stance regarding the delivery systems providing clinical genetic services and health care in our country and our world. The new Genetic Services Committee, chaired by Becky Anderson, MS, JD, was a carefully thought-out step in this direction. Subsuming two earlier *ad hoc* committees on Quality Assurance and Human Resources, the committee will begin to tackle issues not previously considered.

International Opportunities

One of my goals as your president is to 'get the word out' about the diverse expertise among our membership to colleagues overseas attempting to set up genetic service or counselor training programs. I would like to see members of NSGC's Genetic Services Committee and others move into collaborative relationships with Ministries of Health, universities and medical centers designing or expanding such programs. Why are so few countries outside of North America training or hiring allied health professionals with expertise in medical genetics, psychosocial aspects and public health when this model has proven to be so successful here?

Proactive Evaluation of Genetic Counseling Services

I am also convinced that we need proactive participation in the process of evaluating clinical genetic services as they are being provided in our country. Very little is known regarding the ability of clients to access our services or client satisfaction once they do so. Evaluation of our programs and development of means to maximize their benefits will be important aspects of clinical genetics in the 1990s. Let's make sure that we're sitting at the table where policies are set!

I still remember the arguments of a colleague resistant to the idea of a professional society for genetic counselors in 1977. (She still hasn't joined!) Her concern was that defining the role of a genetic counselor would squelch creativity and limit the available boundaries. How mistaken this view has turned out to be! As I communicated in my presidential address, our diversity has turned out to be one of our strengths. Let's capitalize on these as we move into new arenas. ■

Karen Greendale, MA
President

A Commendable Effort

Thank you to Judith Benkendorf and Rose Grobstein, chairs of the *ad hoc* Bill of Rights committee. Other members were Shane Palmer, Carol Strom, Helen Travers, Wendy Uhlmann, Ann P. Walker and Joan Weiss.

Spotlight on the Board

Home Alone…

We join Bea Leopold in NSGC's Executive Office, a 9 x 13 room neatly lined with an eclectic gathering of hi-tech equipment and scattered with casual disarray. There's a computer with a huge screen, two printers, a fax, a postage meter and, of course, her telephone, all just about within arm's reach. There are shelves of books, supplies and journals, two file cabinets and, among some special photos and her degrees (her masters is in communications, not genetic counseling), is her cherished recognition plaque honoring the "professionalism, direction and vision that she brings to the Society," presented by NSGC in San Francisco.

A 4 x 6 bulletin board is scattered with NSGC's vision and mission statements, To Do lists, To Bill reminders, the Connecting Links master list and an array of cartoons and quotes that express various feelings she shares with the likes of Calvin and Hobbes, Peanuts and the Outland characters. Taped to the wooden sides of the Board are four worn and curled sticky notes, two each from her now married children, copying (and out-doing) the other with sibling playfulness, "Thanks, Mom. I Love You" and "Mom, Thanx, I Love You Very Much."

Dressed in brightly colored print leotards, a T-shirt, rolled-down socks and sneaks, she looks more like she's ready for an aerobics class than a day at the office. This vision contradicts my preconceived notion of how NSGC's Executive Director, the voice at the other end of the line, would present.

...Reflections from the Other End of the Line

What's it like to work at home?
Questions about working at home are the most frequent ones I am asked, usually followed by, 'It must be great.' I began working at home in 1980 as the first Executive Director of the local Huntington's disease chapter, long before home offices became a trend. It was then and is now a matter of mutual convenience.

Clearly, there are benefits and deficits. While I gain precious autonomy, save NSGC overhead and avoid petty office politics, working at home can become quite lonely at times.

Describe a typical day working as NSGC's Executive Director.
I am contracted to work 3½ days per week, interpreted as an *average* of 28 hours per week. The easiest way to describe a typical day would be to look at my phone log, which averages 12-36 calls each day. Let's choose last Monday…Calls ranged from a member looking for another who had recently relocated; a reporter checking facts about the genetics of alcoholism in the Native American population; six requests for career packets; a call from a new support group wanting exposure to our members; two Board members requesting Connecting Links information; a member seeking salary information; an exhibitor requesting labels; a member requesting the registration list for the last Annual Education Conference; a medical center genetic department seeking patient literature on 13-14 translocation; and two calls about the availability of liability insurance. I triage to the membership and other resources a lot!

Had that been a first or third Monday of the month, I would have also been working on the JobConnection, a task that takes at least all morning. Had it been within the six week period prior to *Perspectives* going to press, when Vickie and I spend about 60-80 hours each editing and copyfitting your newsletter, that, too, would have been interspersed with calls.

Another way of looking at my day in a more global sense would be to read my quarterly reports, which carry the following standing subheads:

- Site Visits
- Board and Committee Membership Interactions
- Publication Fulfillment
- Referrals and General PR
- Annual Education Conference (when timely, which is always, except for the six week break immediately following the conference)
- New, Ongoing and Scheduled Project Updates
- Administration
- Statistics (of just about every imaginable item that passes through this office) and,
- Professional Development (as it relates to NSGC).

My assistant, Audrey, processes all mailings; career requests, now totalling about 800 each year; database updates and most of the prep work for the membership directory and conference book.

What is the biggest challenge of your workstyle as it relates to your role as our Executive Director?
I have a hard time reconciling that even though I know every member by name, I do not necessarily remember each of you when we re-meet at the Annual Education Conference. I expect myself to recognize all of you by face, as well. It frustrates me to have to be introduced more than once or twice, because in my mind, I *do* know you. Additionally, I have fairly poor body observation skills… change a hairstyle, begin wearing glasses, gain or loss a few pounds, and you are new to me. I always appreciate it when you (re)introduce yourselves to me at the conference.

How has your role as NSGC's Executive Director evolved and where do you see it going?
Since becoming your Executive Director in 1987, my role has developed neatly into three categories: Leadership Management, Centralization and Smooth Day-to-Day Operations. I see these major headings as they integrate into NSGC's vision and mission.

I would like to transfer more operational activities to Audrey and concentrate more on the leadership role, keeping NSGC competitive in the evolving field of professional membership organization development. I would also like to continue fostering the strides we have made in interactions with other genetic and related associations. Most importantly, however, I see my role as serving you, the membership, in any usual or creative ways you might need.

— Bea Leopold, MA
Executive Director

■ *Editor's Note: Because she works alone, Bea decided to honor my request to feature your Executive Office in the ongoing series of articles highlighting your leadership by painting a picture of her office for you and interviewing herself.*

— Vickie Venne

MEMBERSHIP ALERT

Members can look forward to receiving the following important Society mailings:

- 1994 Dues Invoices, mailed December 1 with a January 31 return deadline.
- Winter Membership Mailing to include: 1993 Annual Report, Call for Nominations, Call for Abstracts and the new LDS Speaker Fund information, among other important Society business.
- Professional Status Survey, slated for Spring '94. If you have an issue you would like to address in the upcoming survey, contact Ann Happ Boldt, 317-274-1061.

Perspectives in Genetic Counseling is published quarterly by the National Society of Genetic Counselors, Inc.

Send articles, reviews, correspondence to the Editor-in-Chief; address changes, subscription inquiries, advertisements and classifieds to the Executive Director. Contact any member of the Editorial Board with ideas or suggestions.

Publication Date, Next Issue: March 15
Deadline for Submissions: February 10

[Important: The* Perspectives *Office and the area code for the Executive Office have changed. The corrections are noted above.]

COMMITTEES AT WORK: ACTION IS KEY…

13th Annual Education Conference

"TWENTY FIVE YEARS OF GENETIC COUNSELING: EXPANDING ROLES, EXTENDING HORIZONS"

■ *NSGC's 13th Annual Education Conference commemorates 25 years of genetic counseling. Genetic counselors represent a wide range of expertise which the conference will explore in a celebration of this profession's successes. The conference will address historical perspectives, current methodologies and future trends. Discussions will feature leading spokespersons in the field of genetic counseling and medical genetics with a focus on the expanding roles for genetic counselors and extending horizons of genetics in health care. Issues to be addressed will challenge the genetic counseling profession at large, as well as the individuals comprising this growing profession.*

DATES: October 15 - 18, 1994

LOCATION: Le Meridien, Montreal, Quebec, Canada
Le Meridien, located in the heart of Montreal, connects to the Complexe DesJardins, which includes 120 shoppes, boutiques, restaurants and movie theaters. It is the closest hotel in Montreal to the Montreal Convention Centre, Place des Arts and is conveniently located by a metro stop. Le Meridien is within walking distance to Old Montreal, Chinatown and other major attractions.

Rooms are an affordable $115/room, single or double; $140 triple, in Canadian funds, currently favorable to the American dollar. A limited number of suites are available. Arrangements have been made for NSGC registrants to retain their rooms throughout the duration of the ASHG meeting.

WORKSHOP TEASER: If you have successfully used results of the NSGC Professional Issues Survey or any other materials in your job negotiations, we welcome your story or participation in an upcoming workshop. Please contact Diana Chambers at 901-528-6595.

INFORMATION:

CO-CHAIRS:	Shane Palmer, MS, 919-946-6481 Janice Edwards, MS, 803-779-4928
ABSTRACTS:	Rob Resta, MS and Juliann Stevens, MS
COMMUNICATIONS:	Lisa Amacker North, MS
PROGRAM:	Andy Faucett, MS
RESOURCE CENTER:	Cynthia Kane, MS
WORKSHOPS:	Karen Copeland, MS
REGISTRATION, EXHIBIT INFORMATION, AUXILIARY MEETING ROOMS, HOTEL RELATIONS *(except sleeping rooms)*:	Bea Leopold, MA

Look for Call for Abstracts in the Winter Membership Mailing and complete program and registration information in March.

...TO ACHIEVING GOALS OF STRATEGIC PLAN

BOARD ACTIVITIES from p. 1

- Finance Committee, Treasurer and Executive Director are implementing their plan for streamlining NSGC's financial operations.
- Nominating and Executive Committees are exploring strategies for leadership development.
- Professional Issues Committee is developing a plan for establishing recognition awards for NSGC members.
- Membership Committee is identifying strategies to increase recruitment into the field of genetic counseling.
- Genetic Services Committee is addressing issues of quality control and standards of care.
- Social Issues Committee is exploring options for notifying members of pertinent legislative issues.

VOTES

The *ad hoc* committees on Quality Assurance and Human Resources were dissolved. The activities of these committees will be subsumed into the Genetic Services Committee.

The proposal for an LGS Speaker's Fund was approved. (See page 14.)

The *ad hoc* Bill of Rights Committee was dissolved. Their document was given to the Membership Committee for revision and consideration for alternative publication.

Article 7.5 of the By-laws was amended to read: shall tabulate ballots received no later than six weeks...

Article 1.7 was renamed 1.8.

Article 1.7 now reads: By accepting membership, a member agrees to be guided by the Code of Ethics (adopted Jan. 1992). ■

VISIONING THE FUTURE

Genetic Services Committee: Charting our Futures

A lively committee meeting in Atlanta set the tone for this newly-minted standing committee. Its missions include:

- Carrying forward the work of the previous *ad hoc* committees on quality assurance and human resources
- Participating in national efforts to draft standards of practice and
- Collaborating with other NSGC committees regarding activities such as public relations and minority recruitment.

As health care alignments are shaped around the country, we must accurately and comprehensively define our diverse roles as genetic counselors. We must also be prepared to describe the contexts in which genetic counseling occurs, the supporting staff and services we require to perform our services appropriately and the mechanisms by which we can assure that patients receive genetic services in a timely and helpful manner. A number of working groups were formed to identify salient issues and draft guidelines intended to assist genetic clinics in evaluating and shaping their programs. The following topics will receive attention:

- Defining the role of genetic counselor as laboratory broker; assessing the service, quality and reliability of reference labs
- Assessing the process of gaining access to genetic services
- Diversifying the profession
- Understanding the clinic milieu, including: stages and timelines of the evaluation process; documentation; chart content and review; case conferences as patient service; and quality assurance
- Assuring patient confidentiality
- Assessing patient satisfaction and
- Defining our duty to recontact.

To join one of these groups, call me at 402-559-6497 and I will refer you to the appropriate subcommittee chair. ■

— Rebecca Rae Anderson, MS, JD
Genetic Services Committee Chair
University of Nebraska Medical Center, Omaha NE

HUMAN GENOME PROJECT UPDATE

Research Plan for the Next Five Years

Progress over the past three years in genetic and physical mapping has exceeded expectations. The original goals, documented in a five year plan, were met at less expense than previously planned. A summary of the new five-year plan appears in the October 1 issue of *Science.* Francis Collins, MD, co-authored the article with David Galos, PhD. The new goals will span the period between 10-1-93 and 9-30-98. Advances in genome research have to be incorporated into the present strategies to ensure the program continues to be ambitious and at the cutting edge. ■

JoAnn Inserra, MS
Norwalk Hospital, Norwalk CT

The Code of Ethics: A Further Application

Often we find that issues arise in our clinical lives for which we are hard pressed to come up with clear answers. This article is the second in a series examining the use of the Code of Ethics as a guide.

Recently, a staff physician had her purse stolen. She described the thief and reported that she had seen the woman leaving our office. Security asked if we had seen anyone answering the description provided by the doctor. We had; she had been scheduled for our 1:30 appointment.

We were asked to provide her name, address and phone number. One colleague suggested that complying would be a breech of confidentiality.

That sent me to our Code of Ethics, Section II-5, which states that genetic counselors strive to "maintain as confidential any information received from clients, unless released by the client." This is important because trust is a basic element of the counselor-client relationship. But what does it mean to "receive from" a client?

Sometimes It's Clear

The answer is relatively straight forward in the context of a counseling session. Verbal, or nonverbal communications, which include body language and photographs, can be "received." We can even easily include written communications from a client. Test results on tissue samples can also be considered received "from" (in the sense of coming out of) the client. At this time, none of us would ask for or expect to have the results of client tests released without written permission. Section II-5 certainly applies here.

Sometimes It's Not

Information *about* a client, however, is received from other sources, such as referral center or support service. If we believe this information belongs to the client, we are safe in assuming that a release has been given. So we still have received the information from the client (albeit in a round about way). This would also apply to section II-5. Obviously, we did not have a release from our client to give her name and address to Security.

Saying we "strive" to maintain confidentiality suggests there may be exceptions. For example, we discuss counseling sessions with members of our team. They could be considered professional extensions of the counselor and obligated by the same Code. Also, without identifying individuals, we discussed client issues with peers and present findings and problems to colleagues. This is appropriate in light of Sections I-2 and I-4 (striving to continue our education and training, and knowing our limits). Would Security be included in the category of authorized personnel? Staff members in our center could not agree on this point. If the answer is not clear, Section II-5 must apply.

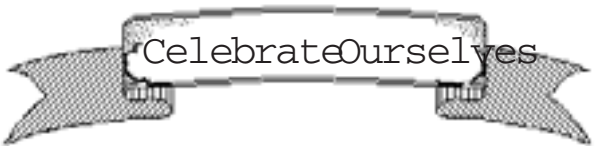
Section IV-6 tells us to strive to adhere to laws and regulations of society. We don't know that our client has plans to or has broken any laws. The need to cooperate with Security regarding the theft of a purse does not outweigh the importance of maintaining the counselor-client trust.

The Genetic Services Committee is planning to address this issue in greater detail over the next several months. It will be important to look at our individual responses to my situation to consider the range of approaches. ■

Susan Schmerler, MS
St.. Josephs Hospital Medical Center, Paterson NJ

CDC Director Meets with NSGC Board

José Cordero, MD, MPH, Assistant Director for Science, Division of Birth Defects and Developmental Disabilities, Centers for Disease Control (CDC) presented at the October NSGC Board meeting. Dr. Cordero, known to many of us because of his work in teratology and public health, brought the Board up to date on CDC projects concerning the prevention of birth defects and developmental disabilities. Key among these are the Folic Acid and Prevention of Neural Tube Defects Projects and an FAS Surveillance Program. Dr. Cordero invited members of NSGC to contact the Division for more detailed information. Those interested in funding opportunities through the CDC should be aware that current priorities include the prevention of disabilities associated with sickle cell disease, fetal alcohol syndrome and neural tube defects. Grants are often awarded through State Departments of Health. Not many grant applications request funds for development of educational materials geared to professional or lay audiences. This type of project might benefit from collaboration between the CDC and NSGC. ■



JEMF Supports Sabbatical Year for Study of Family Counseling

The second Jane Engelberg Memorial Fellowship was awarded to **Deborah L. Eunpu.**

Debbie will use the $25,000 award to support a one year sabbatical at the Marriage Council of Philadelphia, an affiliate of the University of Pennsylvania. This program is nationally recognized for its integrated approach to family counseling and its high caliber faculty. The program provides an intensive one-year experience in coursework and clinical casework in marriage and couple's therapy.

Her proposal, "The Use of Psychodynamic, Couple and Family Therapy Theories and Techniques in Genetic Counseling," will permit her to explore ways in which genetic counseling practices are influenced by the systems theory approach. Debbie's proposal stemmed from her interest in the long-term effects of genetic disorders on families which cannot typically be resolved in the traditional genetic setting.

The product of her sabbatical experiences will provide materials with a systemic family approach, adding to the developing library of genetic counseling literature that incorporates psychotherapeutic issues.

She graduated from Sarah Lawrence College in 1977, spent 10 years at Children's Hospital of Philadelphia and six at Albert Einstein Medical Center as Director of Genetic Counseling in their Center for Developmental Medicine and Genetics.

Special Projects Fund to Create Support Group Manual

The 1993-94 Special Projects Fund was awarded to **Molly Minnick** and **Kathleen Delp** for their proposal entitled "A Manual for Conducting Support Groups for Parents who have Interrupted Pregnancies Secondary to Fetal Anomalies." They have co-facilitated a support group at Michigan State University for six years.

This project will create a two part manual for professionals interested in facilitating support groups for parents who have interrupted pregnancies. The collaborative effort will include both professionals and parents completing surveys about available support groups, how they are conducted and how parents have been affected by participating.

On the Road Again

On October 18, **Betsy Gettig** was invited to speak at the annual meeting of the Institute of Medicine, National Academy of Sciences, Washington DC. She was on the agenda with noted geneticists, including David Botstein and Francis Collins. She spoke for 20 minutes about the status and future of genetic counseling. After her presentation, C. Everett Koop, who was sitting in the front row, complimented Betsy for her talk, introducing himself as "Chick Koop." "I always wondered what the 'C' stood for," mused Betsy. Now she knows.

NSGC goes to NABT

NSGC exhibited at this year's National Association of Biology Teachers (NABT) National Convention, held in Boston, November 17-21.

A new banner announcing us as *the leading voice, authority and advocate for the genetic counseling profession* flew over the booth as hundreds of brochures were distributed. Interest was high since many are using sophisticated materials and curricula to teach genetics and the Human Genome Project. Several could identify a genetic counselor in their community. This key audience was enthusiastic about taking materials and information back to their students, our future colleagues.

Thanks to **Mona Inati, Ed Kloza, Marisa Ladoulis, Barbara Lerner, Michele Murray, Karen Treat, Janice Stryker, Mary-Francis Garber, Karen Greendale** and **Judith Tsipis** for their time, commitment, energy and expertise.

Kloza in NERGG

Edward Kloza, NSGC's Past President II, was elected co-director of the New England Regional Genetics Group.

NSGC Strategic Plan is Focus of Presentation

NSGC Executive Director **Bea Leopold** presented "If It Ain't Broke, Fix It: Strategic Planning from a Position of Strength" at the American Society of Association Executives 11th Annual Management Conference in New York on December 7.

The 1½ hour presentation was based on the successful completion of the first phase of NSGC's strategic plan.

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COUNSELING ISSUES

Counseling difficulties arise because so few studies are available for predicting whether UPD for a particular chromosome is associated with a specific clinical course. UPD for chromosome 15 and partial UPD for chromosome 11 have known consequences, but little data is available for UPD for other chromosomes. UPD for chromosome 16 appears to be related to intrauterine growth retardation (IUGR) but has good outcome for most newborns. Kalausek, *et al.* recently reported on four cases of UPD for chromosome 16 in which three had IUGR, and the fourth had an unexplained fetal demise. One child had an imperforate anus but otherwise all showed normal development.

UPD — COMMON?

The frequency of UPD is not currently known, although it is safe to say that it is surprisingly high. Prior to the first reports of human UPD in 1988, few genetic professionals considered a role for UPD in the causation of human disease. The discovery of placental mosaicism for trisomies led to investigations which uncovered UPD. Balanced familial translocations in a child with developmental impairments also have revealed situations involving UPD.

Currently, whole chromosome UPD has been demonstrated for chromosomes 7, 9, 14, 15, 16, 21 and X, as well as partial (or segmental) disomy for chromosome 11 in Beckwith-Weideman syndrome. However, not all UPD leads to distinct clinical entities, and it may be that UPD for some chromosomes produces no effect or may have a beneficial effect. The best examples of UPD causing aberrant development are Prader-Willi syndrome (PWS) and Angelman syndrome (AS). These patients have no deletion of the critical region on chromosome 15, but have maternal or paternal UPD, respectively. The PWS and AS story illustrates how the imprinting of the genome (presumably during germline formation) influences gene expression.

IMPRINTING IS KEY

One cannot understand UPD without understanding the concept of genomic imprinting. It appears that methylation of some deoxycytidine nucleotides

Case 1: Restricted Patient Information

Denise Simonsen, MS, UC Irvine, Orange CA

Mrs. H. was referred for genetic counseling after two amniocentesis procedures revealed trisomy 16 mosaicism. The first amniocentesis at 17 weeks revealed that 43 of 46 colonies were 46,XY, and three colonies from two primary cultures were 47,XY,+16. The 19 week amniocentesis revealed 76 of 77 colonies were 46,XY, and one colony had one cell with 47, XY,+16 and two with 46,XY.

The couple was counseled about the test results and the limited information available about mosaic trisomy 16. They were informed that the most likely explanation was placental mosaicism.

We discussed recent reports in which confined placental mosaicism for trisomy 16 had been associated with IUGR in the fetus, but malformation and mental retardation were unlikely. The couple was offered fetal blood sampling, but declined and elected to monitor the pregnancy with serial ultrasound. Asymmetric growth retardation and an unusual placental lake were observed.

The pregnancy was unremarkable for exposures. Mrs. H. developed hypertension and baby H was delivered by emergency C-section at 36 weeks. He weighed 3 pounds, 7 ounces, and was 16 inches with Apgars of 7 and 9. Hypospadias was noted. At 5 1/2 months, the family and their pediatrician felt that he was developmentally normal and his catch-up growth has been good.

The placenta, cord blood and parental bloods were sent to Dr. Dagmar Kalousek (University of British Columbia) for research purposes. The couple understood that there would be no direct benefit to them from the study. Cytogenetic examination of the placenta revealed that the trisomy 16 was confined to the chorionic plate and chorionic villi and not found in the amnion. There was no evidence of a co-twin. DNA analysis of parental and cord blood revealed that baby H had uniparental disomy (maternal hetero-disomy) for chromosome 16.

The couple was informed that the studies had confirmed the confined placental mosaicism. They were also told that there is no evidence that their son had any trisomic cell lines, but that the analyses were limited. We did not inform them that their son had uniparental disomy because we did not have any evidence at that time that it would have had any effect on his growth or development. The couple has declined any further genetic evaluation at this time.

may be the primary epigenetic event. It remains to be seen whether other DNA modifications besides methylation play a role in imprinting. The reason UPD of some chromosomes has more severe consequences is likely related to particular imprintable genes on chromosomes or to the number of imprintable genes located on those chromosomes. In addition, UPD can lead to recessive diseases even when genes on a particular chromosome are not imprinted in the germline, as was the case in the original UPD report in humans in which a child had cystic fibrosis (and growth impairment) derived from UPD of chromosome 7. The child inherited both chromosome homologues from the CF carrier mother.

TIP OF THE ICEBERG

It is possible that the cases of UPD uncovered to date represent only the "tip of the iceberg" and many other previously unexplainable syndromes may be related to UPD. Examples of clinical entities which may also be candidates for UPD studies are primordial dwarfism, symmetric and asymmetric gigantism, abnormal phenotypes with balanced chromosomes, poly/asplenia, Russell-Silver syndrome and unexplained fetal demise.

Patients with unexplainable clinical findings who carry a familial (apparently) balanced translocation or patients with recessive diseases when only one parent can be demonstrated to be a carrier may also be candidates for UPD investigations. The detection of UPD using DNA markers requires that the parents have DNA studies simultaneously to determine the parent of origin for chromosomes inherited by a child. ■

Fryburg, et.al. *Prenatal Diag*, 12:157-162, (1992).

Kennerknecht I and Terinde R. *Prenatal Diag*, 10:539-544, (1990).

Kalousek, et al. *Am J Hum Genet*, 52:8-16, (1993).

Post JG and Nijhuis JG. *Prenatal Diag*, 12:1001-1007, (1992).

Williams J, et.al. *Prenatal Diag*, 12:163-168, (1992).

Case II: Full Patient Information

Katie Leonard, MS, Baylor College of Medicine, Houston TX

Mrs S. was referred to our center due to advanced maternal age. The result of her CVS showed 15 cells from 5 primary cultures with trisomy 16. At follow-up counseling to discuss the CVS results, Mr. and Mrs. S. were told that the most likely explanation for the result was confined placental mosaicism. An amniocentesis was recommended to clarify the CVS results.

The result of the amniocentesis showed 46,XX in 30 cells from two primary cultures. This rules out 10% mosaicism at the 95% confidence level. In light of these results, we discussed the recent studies which indicate an increased risk for IUGR in the fetus when there is confined placental mosaicism. We recommended that Mrs. S. have serial Level II ultrasounds throughout her pregnancy to watch for any complications. This information was also discussed with her referring obstetrician.

The issue of uniparental disomy was discussed with the parents at the initial follow-up counseling session. It was explained that in one study, 4 of 9 children with similar CVS and amniocentesis results had two number 16 chromosomes from their mother and none from their father. All of these children had IUGR at birth. Other children in the same study also had IUGR and normal biparental inheritance for chromosome 16. The couple was told that at this time it is not known whether or not uniparental disomy for chromosome 16 will have any effect on the child. Parental bloods were requested for research purposes. The couple was interested in participating in this research, but have not had their blood drawn. We have also requested tissue samples on Mrs. S.'s placenta and amnion at the time of the baby's birth.

COUNSELING PROTOCOL

Counselors might consider these questions when confronted with UPD cases:

- *Should we inform patients of pseudomosaicism for trisomy 16 on amniocentesis because of the possibility of confined placental mosaicism and IUGR?*
- *Should we recommend serial ultrasound for all patients with trisomy 16 mosaicism on CVS or amnio?*
- *Should we inform parents when uniparental disomy 16 is detected even though it might be a benign condition?*
- *Does fetal uniparental disomy 16 have an adverse effect on postnatal development?*
- *Is it the trisomic placenta, the uniparental disomy, or both that affects intrauterine growth and development?*

— D. Simonsen

✍✍✍ LETTERS TO THE EDITOR ✍✍✍

SEMANTIC CLARIFICATION

TO THE EDITOR:

The information on databases for genetic materials *(PGC 15:3)* was interesting. I am not familiar with all of the titles listed. However, you should be aware that some of the listed databases are not databases at all. For example, Grateful Med is a communications software that permits one to interact with MEDLARS (Medical Literature Analysis and Retrieval System) of the National Library of Medicine (NLM). Comp-U-Serve is also not a database, but a communications software that permits interaction with private vendor databases as well as various private vendors. I know the logistics of screening information for accuracy prior to publication may be difficult. However, that should be done so workers unfamiliar with given areas are not lead astray.

Howard J. Allen, PhD
Dept Gynecologic Oncology
Roswell Park Cancer Institute
Buffalo NY

AUTHORS' REPLY:

Thank you for your clarification of terminology. Because one purpose of the survey was to gather as complete a list as possible, the question regarding services was open ended. Therefore, the responses included a variety of computer services, many with which we were also not familiar. Future Perspectives' *articles might summarize some of the less known services. Any users willing to share their knowledge about specific services are invited to contact the GeneBytes authors.* ■

Barbara Pettersen, MS
Jannell Sloan, MS
Co-Chairs, Patient
Literature Subcommittee

WHO IS RESPONSIBLE?

TO THE EDITOR:

Re: Using Genetic Support Groups Wisely *(PGC 15:3)*. We applaud the effort of genetic counselors to familiarize themselves with local support resources to aid in patient adjustment. The Alliance of Genetic Support Groups stands ready to make referrals to national groups when a local resource is unavailable or not known.

One aspect of the article troubles us. Ms. Lerner points out that one basic tenet of genetic counseling is nondirectiveness. She then takes a directive stance by suggesting that genetic counselors make decisions about the quality of the referral group.

The Alliance has struggled with this issue. With rare exceptions, the value of a group can only be determined by the individual or family. Thus, in making a referral, we have concluded that it is up to the individual *and* the group to make a judgment, *not* the health care professional. This does not mean that the counselor should not learn about the group before a referral is made.

The Alliance stands ready to offer technical assistance for member organizations, and we strongly recommend that a genetic professional serve on their Boards of Directors and help them, when requested. ■

Joan Weiss, MSW, LCSW
Alliance of Genetic Support Groups

REVIEW ACKNOWLEDGED

TO THE EDITOR:

Thank you for the excellent review of *Precious Lives, Painful Choices: A prenatal decision-making guide (PGC 15:3)*. I know I represent many parents in saying that the work of genetic counselors is highly valued. *Precious Lives* is both a tribute to genetic counselors and a resource for them. Since the book is not available through normal channels, copies can be ordered from Wintergreen Press, 3630 Eileen Street, Maple Plain MN 55359. 612-476-1303. ■

Sherokee Ilse

Student Corner

A new school year is in full swing, and we have already begun to hear from some students attending training programs. We would like to hear from all of you. If there is other exciting news related to your training program, this is the place for the students or program directors to share the information. Theses noted include:

- UNIVERSITY OF CALIFORNIA - IRVINE. Lori Williams, Use of FISH to investigate allele loss in Tuberous Sclerosis. 714-456-5791 (day); Lisa Mullen, Segregation analysis of cleft lip with or without cleft palate in Vietnamese families. 714-456-5791 (day).
- UNIVERSITY OF MINNESOTA. Paula Winter, Confidentiality concerns involved in a family study of breast cancer. 612-331-6046 (eve); Vickie Matthias Hagen, Application of FISH to the study of sex chromosome mosaicism. 612-626-1900 (day) ■

— Rich Dineen, MS and
Bonnie Hatten, MS

Research Network

I'natl NF Network Funded

Children's Hospital of Boston has received support from the National NF Foundation to initiate a worldwide Clinical Coordinators' Network, providing a forum to address clinical issues related to NF1 & NF2.

Contact Gretchen Hehn Schneider, MS, Childrens Hospital, Fegan 10, 300 Longwood Ave, Boston MA 02115; Fax# 617-277-5933. ■

Pre-Implantation Genetic Analysis for CF

A Demonstration Project, designed to reduce the likelihood that couples at 25% risk for CF will have affected children, has been launched by IVF America. The project combines in vitro fertilization with genetic testing of embryos, giving high risk couples the opportunity to ascertain the diagnosis before the pregnancy is established. Most costs will be defrayed by IVF America.

The project is recruiting 40 couples. To refer couples or receive information, contact Niecee Singer, IVF America, 201-736-7200. ■

Natl Registry for Hearing Impairment in Omaha

The Center for Hereditary Communication Disorders at Boys Town National Research Hospital has been awarded a contract to establish and maintain a National Hereditary Hearing Impairment Resource Registry to assist research in hereditary communication deafness. Families or researchers may contact Dr. Paul Ing, Boys Town National Research Hospital, 555 North 30th St, Omaha NE 68131; 1-800-835-1468. ■

GeneBytes

Pedigree Drawing Made Easier

Finally there's a pedigree drawing program that is powerful but easy to use. Cherwell Scientific's Cyrillic will meet the needs of most genetic counselors. This Windows-based product is a vast improvement over other pedigree programs. Most genetic counselors want to draw professional-looking pedigrees for publications, slides and overheads. Cyrillic accomplishes this task nicely. We were able to produce a publication-quality printout of a pedigree within 30 minutes of installing the software.

Typical symbols, such as multiple partners, miscarriages, stillbirths and terminations, are easily selected, and users can create new symbols. In addition, the user can select from multiple shading and fill patterns to identify disease/marker status, with an option of printing a legend at the bottom of the pedigree. New family members can be added at any time. The standard numbering system (Roman numerals for generations, Arabic for individuals within a generation) is used, but the user must remind the software to update the numbering when individuals are added or removed.

A dialog box, which pops up with the click of a mouse, makes for easy entry of individual data, such a gender, adoption status, disease and marker information, names and proband status. Information can be selectively displayed on the pedigree.

Database Capabilities As Well

Besides the basic ability to draw a pedigree, Cyrillic can be used to manage a DNA database. We did not evaluate this capability in detail, but the simple tests we ran were handled with ease. Users can readily enter and display DNA markers and phenotypic information in standard formats. Cyrillic will handle the needs of all but the largest DNA research projects. The program can handle 1,000 family members, 50 markers per family, 250 markers per chromosome and 200 genetic disease definitions. Data can be exported directly into linkage programs, such as MLINK and LIPED. Conversely, data can be imported from other standard file formats, such as DOS text.

A useful feature of Cyrillic when creating reports or articles is the ability to export the pedigree image directly into Microsoft Word for Windows. In addition, the image can also be exported into standard Windows graphics programs to enhance the image for slides, overheads and publications.

Systems requirements for Cyrillic are Windows 3.1 or later, Windows-compatible monitors and printers, 1 Mb of disc space and a minimum of 2 Mb of RAM (We found it works better with 4 Mb). A math co-processor is necessary only for heavy use with linkage programs. Cyrillic is available from Cherwell Scientific Publishing Co., 156 Auburn Place, Brookline MA 02146; 617-277-4200. ■

Note: Version 1.1 will be released soon. Enhancements include the ability to import data from PedDraw and ASCII files, import marker data from a text file and save the pedigree with the personal data removed.

Robert Resta, MS

■ RESOURCES ■

BOOKS

The Student with a Genetic Disorder

edited by: Diane Plumridge, Robin Bennett, Nuhad Dinno and Cynthia Branson

published by: 1993, Charles C. Thomas (Springfield IL) 365 pp.

reviewed by: Jennifer M. Lee, MS, Obstetrics & Gynecology Consultants of the Southwest, PA, Fort Worth TX

This well-written reference introduces the reader to basic human genetics and the principles used to diagnose genetic disease. In the initial chapters, basic concepts such as inheritance and variability are explained succinctly for the non-genetics professional. The authors also provide a synopsis of the roles special education and therapy play in the life of a child with a genetic disorder.

The remainder of this reference is devoted to explanations of the specific characteristics and needs of children with a variety of genetic conditions. These explanations emphasize the child's *abilities* rather than the disabilities and provide the reader with an appropriate "first glance" of any one particular condition.

Each description includes a list of physical features, an explanation of characteristic features, a brief discussion of the impact of the condition on the child's cognitive and behavioral functioning and illustrations, where appropriate. Finally, additional resources and references are provided with each description.

In light of the author's focus on the normalcy of children with genetic conditions, the drawings emphasizing abnormalities seemed inappropriate. These sketches detracted from the well-written and accurate descriptions of children with various genetic conditions. Photographs would have served the purpose better.

This would be an appropriate textbook for an introductory genetic class for non-genetics professionals in various disciplines because of its emphasis on the educational and therapeutic needs of children with genetic conditions. It would also be useful for pediatric genetic counselors who are often asked to explain the educational and emotional impact of a condition to parents and other professionals. Additionally, the descriptions provided could be easily adapted for use as a checklist for appropriate counseling and referral. ■

Brothers and Sisters: A Special Part of Exceptional Families, Second Edition

authored by: Thomas H. Powell and Peggy Ahrenhold Gallagher

published by: 1993, Paul H. Brooks Publishing Company, (Baltimore MD) 291 pp.

reviewed by: Debra Duquette, MS, Hutzel Hospital, Detroit

Resources regarding needs of siblings of disabled persons have been scant. Therefore, it is often difficult for genetic counselors to provide resources for parents who ask about their other children's possible reactions to and concerns with the diagnosis of their sibling. For adult siblings who seek genetic counseling, information about recurrence risks, carrier screening and prenatal diagnosis can be provided. However, genetic counselors must attend to the psychosocial aspects of the sibling relationship that may be too easily forgotten.

Brothers and Sisters is devoted to that relationship, specifically the unique contribution of siblings to the family system. The book addresses the special challenges siblings of disabled persons face. This second edition contains new information about current research regarding siblings' needs, their adjustment patterns, available community services as well as practical advice. It is written for both professionals and families, especially parents and older children.

The book is divided into two sections. The first provides an overview of the importance of siblings in the family system, incorporating current research and personal accounts. The second outlines strategies to assist siblings, including:

- methods of relaying information;
- facilitating social interaction and teaching between siblings;
- developing community programs; and
- helping adult siblings deal with their concerns.

A useful appendix provides a bibliography, categorized by the type of disability. The authors also provide sources of community services and support for siblings and parents.

A strength of this book is the generous use of quotations and photographs from siblings to

highlight important points. These personal notes were often more enlightening than the research. The book provides thought-provoking questions for genetic counselors to ask about sibling relationships. It also outlines sibling concerns at different life-cycle stages with respect to topics such as themselves, their affected sibling, friends and the community.

There are some weaknesses of this book. Although it covers many issues, some important ones, such as the death of a sibling, are not discussed. The book briefly addresses genetic counseling, but only in the context of adulthood. In addition, some of the information provided about genetics is inaccurate. The need for further research and resources for siblings is apparent. Even with its imperfections, this book provides a great deal of practical advice and is one of the best resources available regarding siblings. ■

Resources

Support Groups

Long QT Syndrome

A new foundation, the Sudden Arrhythmia Death Syndromes Foundation, directed by cardiologist G. Michael Vincent, provides education to both the medical and lay communities. For information, call 800-786-7723.

An information pamphlet is available for families and gene linkage studies are being conducted at several medical centers. Contact Jennifer Robinson, MS, LQTS Registry, PO Box 653, University of Rochester Medical Center, Rochester NY 14642-8653; 716-275-5391. ■

Pseudoxanthoma Elasticum

PXE, a rare, inherited disorder of connective tissue, may present as small, bilateral, yellowish bumps in the folds of the arms, groin, knees and armpits, eye problems including a sensitivity to light or calcium deposits in the artery walls, causing leg pain.

This disorder can be autosomal dominant or recessive.

The National Association Pseudoxanthoma Elasticum (NAPE) has recently relocated from Albany NY to 1884 Cherry St, Denver CO 80220-1146. ■

Nager and Miller Syndromes

The Foundation for Nager and Miller Syndromes is an international support group dedicated to helping those affected by these two rare genetic conditions which involve severe facial and limb anomalies, but not intellect. This clearinghouse links families. A medical advisory board assists professionals. A research project, scholarships to a craniofacial summer camp and a 16-page newsletter is also available. Contact: Margaret Ieronimo 708-724-6449. ■

Meeting Manager

1994

Jan 20	*All in the Family - Special Problems Posed by Genetic Research in Human Subjects;* Bethesda, MD. Contact: E. Feingold, 301-496-7531
Feb 28 - Mar 2	*Human Genome Project: Commercial Applications;* CHI, San Francisco. 617-487-7989
March 7-8	3rd Annual Perinatal Substance Abuse Conference, *Twenty-One Years: FAS Comes of Age (1973-1994)*, Madison WI. Contact: Georgiana Wilton, 608-262-6590 or 800-462-5254
Apr 7 - 8	NSGC Region IV Educational Conference, Indianapolis IN. Contact: Kathy Delp, 616-732-8664
Apr 29 - May 2	Parent-to-Parent Conference, *Weaving the Future from the Fabric of our Past*. Asheville NC. Target Audience: Parents and Professionals. Contact: Family Support Network of North Carolina, 1-800-TLC-0042
May 7-10	NSGC Region VI Educational Conference, Asilomar Conference Center, Pacific Grove CA. *Breaking Down Barriers to Genetic Services - Financial, Cultural and Psychological Issues.* Contact: Stefanie Uhrich, 206-543-3767, Robin Bennett, 206-548-4030 or Steven Keiles, 213-857-2074

BULLETIN BOARD

MEMBERSHIP CHAIR NAMED

Karen Copeland has agreed to fill the Membership Committee post vacated by Bonnie LeRoy, who became President-Elect in October. She will complete the remaining year of the two year term. ■

LOST IN ATLANTA

An 8 1/2 x 11 blue spiral notebook with extensive notes from the *ABCs of Cancer Genetics* Short Course. Return to Debbie Raymond, 23 Melrose Ave, Norwalk CT 06855. ■

COURSE DATE CHANGED

Applications are still being accepted for the ELSI-funded "Train the Trainer Course," to be held in Chicago April 20-24. Participants will be trained to conduct the course, "Humanizing Genetic Testing: Clinical Applications of New DNA Technologies," geared for primary care health professionals. All expenses will be covered. For an application, call Beth Fine, 312-908-7713. ■

VOLUNTEERS NEEDED

Interested in communications or critiquing publications? Contact the new Publications Committee.

If you are interested in evaluating brochures for grammar, layout, literacy levels, second language translation or content, your help is needed. Also, this committee will be providing feedback and a "stamp of approval" to NSGC members wishing to write letters of criticism, comment or support to the media about its portrayal of genetic issues.

If you are willing to commit four hours per month, call Nancy Steinberg Warren, 513-599-4475.

GRANTS AVAILABLE TO MEMBERS

■ **HELP SPREAD THE WORD: NEW GRANT FUNDS SPEAKERS' TRAVEL**

Laboratories for Genetic Services (LGS) of Houston has created a travel grant, the LGS/NSGC Speakers' Travel Fund, to facilitate members' access to educating health care professionals and the public about genetic counseling and genetic services. The grant provides matching funds for active members in good standing of the NSGC to speak to various approved groups, e.g. medical and nursing societies, patient support groups, educational institutions, public health associations, special interest groups and political committees. Funds cannot be used to attend genetics meetings. This grant was created in memory of Kurt Fenolio, a genetic counselor who died in 1992. Annual funding of $3000 has been committed by LGS. Information will be forwarded to members in the Winter Membership mailing. Applications may be requested from Caroline Caskey at LGS; 713-798-9500.

■ **3RD JEMF APPLICATIONS AVAILABLE**

The Jane Engelberg Memorial Fellowship (JEMF) award is available to genetic counselors in good standing of NSGC who are Board certified by ABMG or ABGC or Board eligible by ABGC . The JEMF supports study, research, writing or exploration of new interests geared to enhance present skills, develop new skills, contribute to the body of knowledge in the field or expand professional roles. Applications for the 1994-1995 Fellowship are now being accepted. If you have an exciting idea, but need assistance to turn it into a proposal, you are encouraged to call any member of the JEMF Advisory Board, or Katherine Schneider, the first JEMF awardee. Copies of the successful proposals are available from Judith Benkendorf, 202-687-8810. The deadline for submissions is May 2.

The JEMF Advisory Board
Judith Benkendorf, MS, Barbara Bernhardt, MS, Barbara Bowles Biesecker, MS, Karen Greendale, MA and Audrey Heimler, MS

■ **MOVE YOUR SPECIAL PROJECT FROM YOUR DESK TO REALITY**

NSGC's Special Projects Fund provides funding of up to $2000 to one or more genetic counselors for project(s) that focus on the future of the genetic counseling profession and/or the provision of genetic services. Projects will be reviewed on the basis of their merit and strength as well as their vision of the future. Proposals are due by May 15.

Special Projects Committee
Michael Begleiter, MS, Beth Buehler, MS, Linda Lustig, MS, Elizabeth Otto, MS, Bev Tenenholz, MS and Pat Ward, MS

Written or faxed requests for guidelines, eligibility and applications for the JEMF and SPF grants are being accepted by NSGC's Executive Office. Awards will be announced at the 1994 Annual Business Meeting in Montreal.

■ EMPLOYMENT OPPORTUNITIES ■

■ *These classified listings represent the most recent additions to the NSGC JobConnection service. Members and students interested in complete or regional information may receive a computerized printout, at no charge, by contacting the Executive Office. Printouts are mailed on the first and third Monday of each month. This service is strictly confidential.*

NEW HAVEN CT: Immediate opening for Genetic Counselor.
RESPONSIBILITIES: Varied PN counseling & consultation with pts & physicians in tertiary care facility.
CONTACT: Miriam S. DiMaio, MSW, Dept Genetics, Yale Univ School Medicine, PO Box 3333, New Haven CT 06510; 203-785-2661. EOE/AA.

AUGUSTA GA: Immediate opening for BC/BE Genetic Counselor or RN w/ repro genetics experience.
RESPONSIBILITIES: Coordinate & participate in preconceptional & PN GC; oppty to participate in clinical & rsrch activ.
CONTACT: Paul G McDonough, MD, Medical College of Georgia, Section of Reproductive Genetics, CJ-119, Augusta GA 30912-3360; 706-721-2828 or 3832. EOE/AA.

LEXINGTON KY: Immediate opening for BC/BE Genetic Counselor.
RESPONSIBILITIES: Join active PN genetics clinic w/ diverse caseload: all aspects of preconception & PN coun: CVS, amnio, triple profile, teratology, malformations. Tchg oppty available.
CONTACT: Anjana Pettigrew, MD, University of Kentucky College of Medicine, Dept Pathology, Lexington KY 40536-0093; 606-257-4089. EOE/AA.

BALTIMORE MD: Immediate opening for BC/BE Genetic Counselor.
RESPONSIBILITIES: Expanding perinatal svcs in community-based OB/GYN prog offering wide range of clinical dx & lab svcs: CVS, amnio, U/S; GC oppty in PNDx, preconceptional coun, education & mktg.
CONTACT: Josephine Hochuli, RN, BSN, St Agnes Hospital, 900 Caton Ave, Baltimore MD 21229; 800-875-8750. EOE/AA.

BALTIMORE MD: Immediate opening for BC/BE Genetic Counselor. Min one yr exp required.
RESPONSIBILITIES: Partic in all aspects of GC in univ-based fetal dx ctr; occasional ped outrch clinic responsibilities; many oppty for rsrch & tchg.
CONTACT: Carole Meyers, MD, University of Maryland at Baltimore, Div Human Genetics, Suite 400, 405 W. Redwood St, Baltimore MD 21201-1703; 410-706-5749; EOE/AA.

DETROIT MI: Immediate opening for BC/BE Genetic Counselor.
RESPONSIBILITIES: Join active team in large, diverse, rapidly expanding repro gen ctr: CVS, amnio, MSAFP, dx U/S, teratogens, novel fetal therapy. Oppty for rsrch, publications.
CONTACT: Eric Krivchenia, MS or Mark Evans, MD, Div Reproductive Genetics, Hutzel Hospital, 4707 St Antoine, Detroit MI 48201; 313-745-7067. EOE/AA.

NEW BRUNSWICK NJ: Immediate opening for BC/BE Genetic Counselor. CV and 3 ltrs recommendation required.
RESPONSIBILITIES: Join team of 3 GCs, OB nurse, PhD cytogeneticist, MD/PhD clinical geneticist/ molecular biologist in rapidly grow comprehensive genetics ctr: multifaceted position w/ PN emphasis: CVS, amnio, MSAFP, triple screen, U/S, teratogens, specialty & satellite clins, family support grps, educ oppty.
CONTACT: Debra-Lynn Day-Salvatore, MD, PhD, Director, Institute for Reproductive & Perinatal Genetics, St. Peters Medical Center, MOB 4410 New Brunswick NJ 08903; 908-745-6678. EOE/AA.

MINEOLA NY: Immediate opening for BC/BE Genetic Counselor. Exp pref but not req.
RESPONSIBILITIES: Emphasis on PN coun for amnio, triple marker scrn, DNA testing, U/S abnormalities; Ped coun; Educ med students & residents; Prader Willi syndrome support grp.
CONTACT: Liz Hegarty, MS, Div Genetic Counseling, Winthrop University Hospital, 259 First St, Mineola NY 11501; 516-663-2657. EOE/AA.

YOUNGSTOWN OH: Immediate opening for BC Genetic Consultant. 1-2 yr exp preferred.
RESPONSIBILITIES: Work closely w/ Med Dir Genetics at Tod Childrens Hosp: coun pts & families re: inheritance patterns & recurrence risks in PN & peds; assist geneticists w/ fam hx & development of pedigrees.
CONTACT: Ann Marie Ondo, Human Resources Dept, Western Reserve Care System, 345 Oak Hill Ave, Youngstown OH 44501; 216-740-4902. EOE/AA.

NASHVILLE TN: Immediate opening for Genetic Counselor.
RESPONSIBILITIES: Range of genetic svc in academic ctr; wide range of GC oppty in general, PN, amnio, teratogens, MSAFP scrng.
CONTACT: D.N. Singh, PhD, Div Medical Genetics, Meharry Medical College, Nashville TN 37208; 605-327-6399. EOE/AA.

GOING UNDERCOVER ON RAINY DAYS

A limited number of bright blue folding umbrellas embossed with NSGC's logo are now available at a reduced price. Send $8.50 for each umbrella to the Executive Office and your umbrella will be mailed within one week by second day priority mail. Your check, payable to NSGC, must accompany your order.

national society
of genetic
counselors, inc.

nsgc

233 Canterbury Drive
Wallingford, PA 19086-6617

Parting Thoughts

December 31, 1993 marks the retirement of our dear friend and colleague, Natalie Paul, from her position with the Professional Education Division, March of Dimes National Birth Defects Foundation.

During her 23 year tenure with the March of Dimes, Natalie has held nearly every position in the division, including a stint as Assistant Director of Professional Education. Many genetic counselors know Natalie for her able editing and her indefatigable staffing of the March of Dimes exhibits at our conferences, along with husband, Bill. Most of all, we know her as a true supporter of the profession.

In fact, not only was the March of Dimes the very first exhibitor at an NSGC meeting, but many of us have Natalie to thank for her assistance and support in seeing our first professional manuscripts published in the Birth Defects Original Articles Series, which contained the proceedings of our early Education Conferences. During her career, Natalie edited over 150 books, two birth defects compendia and two newsletters.

This past fall, in honor of Natalie's retirement, her 50th wedding anniversary and because for the first time Natalie would be unable to join us in Atlanta, NSGC sent Natalie a gift which was gratefully acknowledged with the following words to our organization,

> "...We have known each other since your beginnings in 1978 in San Francisco and through all of these years my love and great admiration for you has steadily grown. You knew what you wanted and were strong enough to achieve it. Congratulations! Look where you are now! I know that you will continue your great work and believe me when I say that we (Bill and I) will never forget you, your many kindnesses and great friendship. I pray that our paths will cross again soon."

Natalie, so do we, many times over. If Natalie's feeling for NSGC spring from the pride of "watching her child grow up," then suffice it to say that we, as a professional society, would be less of what we are today if it weren't for Natalie's care and nurturing. ■

Judith L. Benkendorf, MS
Washington DC