"Shoot for the moon; the worst that can happen is that you end up somewhere in the stars."

Bill Demby

This Alert is a vehicle of communication between the Alliance and its constituency. It is our goal to provide timely and useful material in a readable, easy access format. Feel free to send in your announcements, share your ideas, or ask for help.

Jayne Mackta, Editor

Medical File Privacy Standards Proposed by HHS: In late September, Health and Human Services Secretary Donna Shalala proposed guidelines that would protect the confidentiality of medical records. The proposed standards would require that doctors, hospitals, insurance companies and medical researchers release individual records only when necessary for medical treatment and reimbursements or when requested by law enforcement authorities. Consumer Alert: The U.S. Senate Labor & Human Resources Committee will hear testimony on privacy and confidentiality of health information on October 28, 1997. If you or a family member have been negatively affected by the unauthorized disclosure of personal identifiable medical information and would like to contribute to written or live testimony for this important hearing, please contact Mary Davidson, Alliance Executive Director, ASAP.

Alliance Board and Staff Launch Membership/ Organizational Campaign: Member surveys, enclosed in the September Alert, are the first step in a year-long organizational campaign to increase membership and member services and to involve more members in Alliance activities. Your cooperation in faxing or mailing completed surveys to the Alliance is greatly appreciated. The second step will be an Alliance leadership meeting in February of Board and Partnership Steering Committee members to review survey results, evaluate membership services, project future organizational objectives and share in a final review of the Partnership Program’s Quality Genetics Services Guidelines. A third and culminating step will be a September 1998 Membership Conference to bring together Alliance members — support groups, consumers and professionals — to approve organizational recommendations and to strengthen partnerships in all areas of genetics. REMEMBER TO SEND IN YOUR COMPLETED SURVEYS AS SOON AS POSSIBLE.

New Alliance Service: In response to repeated requests by helpline callers and conference attendees for a full collection of member brochures, we will offer Alliance brochure packets at the NSGC and ASHG meetings in Baltimore in late October. You can place an order at the meetings or through the Alliance office. There will be a nominal charge to cover the costs of handling and shipping. This convenient, well organized packet of brochures will provide much needed information, resources and contact information to healthcare professionals, teachers, primary care providers and researchers.

New Staff: Toni Pollin, M.S. and Nancy Hsu, B.A., joined the Alliance staff in September. Toni, a genetic counselor, is in charge of the Resource Referral Center. Questions regarding the helpline, the Alert and our website should be directed to her. Nancy, a recent Washington University graduate, plans to enroll in a genetic counseling graduate program next year and will be at the Alliance as an administrative assistant/graduate intern until then. Also, the Alliance, now serving as a field placement for the NIH/Hopkins Graduate Genetic Counseling Program, has genetic counseling graduate student Lisa Pike Buchanan interning at the office one day a week. She answers helpline calls and is undertaking a helpline needs assessment.

Insurance Resources Needed NOW: Helpline callers are requesting assistance with a range of social security, disability and health insurance problems, including plan enrollment, reimbursements for care, coverage for chronic conditions, contesting disqualified claims, temporary financial support and others. Contact Nancy Hsu at the office, if you have resources, organizations, contacts or suggestions to help us meet this growing need.

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Consumers in Demand: Consumer participation is being sought for meetings, panel discussions and speaking opportunities on a range of policy and ELSI issues: genetic testing, patenting and licensure, privacy, discrimination, research, quality genetics services, managed care and health care delivery and living with genetic disease. To meet this growing need and to facilitate a louder, more prominent consumer voice, the Alliance is calling on interested consumers to register for the Consumer Policy Network. Contact Mary Davidson by phone, fax or e-mail: MedAlliance@aol.com.

A Sad Farewell and Hearty Welcome to Alliance supporters at MCH: The Alliance sends Dr. Jane Lin-Fu, Chief of the Genetic Services Branch at the Bureau of Maternal and Child Health, heartfelt thanks and best wishes for a retirement as full and challenging as the career she is leaving behind. “Retiring” has never characterized her work. The Alliance salutes Dr. Jane Lin-Fu as a tough, unyielding advocate for children and families living with genetic diseases and welcomes the new Chief, Dr. Michelle Puyear, a PhD geneticist and pediatrician. To John Gallicchio, Deputy Chief of the Genetic Services Branch, we extend congratulations for his promotion to Deputy Director of the Division of Children with Special Health Care Needs. The Alliance looks forward to continued collaboration with Maternal and Child Health to promote the special healthcare needs of people living with genetic diseases.

Quality Guidelines, Another Step Forward: On September 19, the Partnership brought together twenty Mid-Atlantic consumers to review and expand the Guidelines for Quality Genetics Services. Sponsored by MARHGN, the meeting developed an initial consensus of seven critical indicators of quality service: availability of service, access to genetics services, access to specialists and skilled providers, collaboration of consumers and providers, continuity of coverage and care, access to current information and research, and increased provider understanding of genetic conditions. These guidelines will be used as the basis for assessing genetics service delivery in the Partnership's managed care pilot sites. A similar meeting is being planned in Seattle for later this year.

Partnership Seeks Consumers to Participate in Great Lakes Project: The Partnership is currently recruiting consumers—within 50 miles of Chicago, Detroit, Lansing or Dayton—to join our effort in bringing a greater understanding of living with a genetic condition to medical students in these areas. If you are interested in participating in this project, please contact Nachama Wilker by phone, fax or e-mail at nwAlliance@aol.com as soon as possible.

Martha Volner, Assistant Director/Health Policy Analyst, leaves the Alliance: After nearly nine years of dedication to the mission of the Alliance of Genetic Support Groups, Martha Volner is pursuing new opportunities. Through analysis of policy, insurance and legislative proposals and her advocacy of disability and healthcare issues, she helped the Alliance become a strong consumer voice in the genetics community. On the helpline, Martha addressed the questions and concerns of callers, linking them to support groups, genetic counselors and appropriate community resources. The Alliance staff and Board and the many people whose lives she has touched over the years extend best wishes for continued success.

A Consumer Reports from Washington State: Alliance member Lynne Schauble testified at a recent public hearing held by the Washington State Department of Health. In close collaboration with Alliance staff both before and after the meeting, Lynne reports: “It is encouraging to have the state public health organization invite input from a broad spectrum of interested groups and individuals as partners in the development of this plan (Statewide Genetics Education Plan), recognizing the complexity of the issues, as well as the comprehensive needs of those families and organizations.” Of particular note is the proposal to incorporate genetics education in the Washington State Essential Learnings Curriculum. For more information, contact the Alliance office or Debra Lochner Doyle, Washington State Genetics Coordinator, at 206-464-7752.

The Alliance Receives Three Grants in One Month: The Community Partnership Foundation at SmithKline Beecham has awarded $80,000 to the Alliance to support development and dissemination of the Consumer Guidelines to Quality Genetics Services. MCH Genetics Services Branch approved two grants: one to cover core operating expenses, including the helpline and an Information Resource Center, which will be staffed with fulltime professionals. The second MCH grant will support the activities of the Partnership Program in MARHGN. A fourth grant application to support Partnership activities in PacNORGG and GLaRGG is pending final notification by NIH/ELSI.

Volunteer Protection Act Passes: On June 18, 1997, President Clinton signed the Volunteer Protection Act, which aims to protect volunteers of associations, nonprofit groups, and government entities from liability.
National Standards Urged for Health Plans: On September 24, three nonprofit HMOs, Kaiser Permanente, HIP Health Insurance Plans, and Group Health Cooperative of Puget Sound, joined with two consumer groups, Families USA and the American Association of Retired Persons, to advocate for increased national regulation of managed health care. The current state regulatory patchwork creates delivery problems for both providers and consumers. You can obtain a summary of preliminary principles for consumer protection by calling Families USA at 202-628-3030.

McKusick Receives Lasker Medical Research Award: A Johns Hopkins medical geneticist and editor of Mendelian Inheritance in Man and Online Mendelian Inheritance in Man, Dr. Victor McKusick received this award because of his pivotal role in shaping medical genetics as a distinct medical specialty. Dr. McKusick has been on many support group medical advisory boards, has been a strong supporter of the development of the Alliance, and was a long-term mentor to Joan Weiss, Alliance Founding Director.

Smith Honored: On September 1, Ann C. M. Smith, Vice President for Professionals of the Alliance, received the Alumni Honorary Degree from her 1973 undergraduate Alma Mater, Colorado College. Among many other accomplishments, Ann Smith, a genetic counselor, has played a key role in research on Smith-Magenis syndrome and helped found PRISMS, the support group for families.

Misleading Advertising Concerns ASRM: The American Society for Reproductive Medicine (ASRM) issued a press release on September 15 in response to advertisements for the movie "GATTACA". One ad shows a healthy, smiling baby described as a "Baby Made To Order." Representing ASRM, J. Benjamin Younger, M.D., states, "The advertising campaign, albeit creative and provocative, features what appear to be advertisements for a legitimate company's services, not a movie. We urge Sony to change their advertising to make it clear this is only a movie and that the scenarios portrayed in the ads are not real."

European Biotech Law Makes Progress: In July, the EU Biotechnology law passed its first major committee vote. If passed, this law would allow genes, gene sequences, and biotechnologically engineered products to be patented. In the U.S. many scientists and consumer groups continue to urge the U.S. Patent Office to deny such patents in order to prevent interference with research progress.

Note New Address: The Mid-Atlantic Regional Human Genetics Network (MARGHN), one of the ten regional genetic networks of the Council of Regional Networks for Genetic Services (CORN), has moved effective October 1. The new address is MARGHN, Center for Human and Molecular Genetics, University of Medicine and Dentistry of New Jersey, New Jersey Medical School, Doctors Office Center, Suite 5400, 90 Bergen Street, Newark, NJ 07103-2714; Tel: 973-972-3302; Fax: 973-972-3310; Gisela Rodriguez, MSW, Coordinator.

RESOURCES
"A Question of Genes: Inherited Risks" was broadcast on PBS on September 16. A videotape can be ordered by calling 1-800-440-2651. To obtain an educator's guide, call 1-800-991-1441.

New Web Site: A Place To Remember (www.APlaceToRemember.com) offers support material and resources to those who have been touched by a crisis in pregnancy or the death of a baby.

GENETIC RESEARCH UPDATES
• Genetic Condition Linked to Age-Related Macular Degeneration: A study of 167 unrelated patients with AMD, funded in part by The Foundation Fighting Blindness, showed that sixteen percent of AMD patients had mutations in the same gene that causes Stargardt disease. This study was published in the September 19 issue of Science.

• Researchers Discover Two Genes Responsible for Retinitis Pigmentosa: The discovery of two RP-causing genes, "RPE 65" and "CRALBP" transforms the outlook for millions who suffer from inherited retinal degenerative diseases, not just RP. Researchers believe patients with mutations in these genes may respond to vitamin A therapy favorably. This study, funded by The Foundation Fighting Blindness, is to be published in the October 1997 issue of Nature Genetics.

• Gene Locus for Pseudoxanthoma Elasticum (PXE) Found: A locus for the PXE gene has been isolated and localized to the p arm of chromosome 16 by a research group at Harvard. See the October issue of Human Molecular Genetics.
REQUESTS FOR RESEARCH PARTICIPANTS
The Alliance will dedicate a corner of the Alert to announce approved genetic disease research projects. Please send information on approved studies to the Alliance, and they will be included as space allows.

• PXE International seeks additional families for its blood and tissue registry to aid investigators in cloning the gene. Contact PXE International for more information at 781-784-3817.

• Congenital Lactic Acidosis study enters Phase III of its clinical trial of dichloroacetate (DCA). Mercy Medical Airlift facilitates transportation to and from the Clinical Research Center at the University of Florida. Unless family members have not met their insurance deductible, there is no cost for participation. For more information, contact: Peter W. Stacpoole, Ph.D. M.D. at 352-392-2321.

LEGISLATION UPDATE
• Funding for NIH for the fiscal year of 1998 is still being working its way through Congress. The House version of the bill includes $211.8 million for the National Human Genome Research Institute and the Senate version includes $218.9 million.

UPCOMING MEETINGS
ANNOUNCEMENTS: Member groups wishing to submit a meeting or conference announcement to the Alert are asked to provide the following information within the body of the text so that it is readable: date, location, and contact information, including name of organization, mailing address, and telephone number. Watch for the Alliance at many of the conferences listed below. Meetings where the Alliance exhibits the brochures of member organizations begin with *.

"Mechanisms in Nutrition and Chronic Disease Prevention" • Nov. 7; sponsored by the American Health Foundation and Zepter International; New York Marriott Marquis Hotel, New York, NY. Contact Rajko D. Medenica, M.D. Ph.D. at 1-888-777-6604.

Turner's Syndrome Society of the United States National Conference • Nov. 7 - 9; Hotel Sofitel, Minneapolis, MN. Contact the Society, 1313 5th St., SE, Suite 327, Minneapolis, MN 55414; TEL: 612-379-3607.

International Centennial PXE Conference • Nov. 7 - 9; Bethesda, MD. Contact PXE International, Inc. 23 Mountain Street, Sharon, MA 02067; TEL: 617-784-3817; email: pxe@tiac.net or http://www.med.harvard.edu/programs/PXE. Co-sponsored by the National Association of Pseudoxanthoma Elasticum.

International Down Syndrome Research Conference on Cognition and Behavior • Nov. 20 - 22; Amelia Island Plantation, just north of Jacksonville, FL. Contact the National Down Syndrome Society, 666 Broadway, 8th floor, New York, NY 10012; TEL: (800) 221-4602; e-mail: infot@ndss.org.

First National Symposium on Angelman Syndrome • Nov. 29; Brussels, Belgium; organized by The Angelman Foundation Belgium. For information, contact Dr. B. Dan, Avenue du Parc 147, 1190 Brussels, Belgium; FAX: +32 2 4773287.

"TUSKEGEE: Can Past Lessons Guide Researchers in the Future?", a conference sponsored by the Applied Research Ethics National Association (ARENA) • Dec. 7; Sheraton Hotel, Boston, MA. Contact ARENA at 617-423-4112.

New England Regional Genetics Group 20th Annual Meeting • Dec. 4 - 5; The New England Center, University of New Hampshire, Durham, NH. Contact Joseph Robinson, Coordinator, P.O. Box 670, Mt. Desert, ME 04660; TEL: 207-288-2704.

First Non-Ketotic Hyperglycinemia Family Get-Together • Dec. 20; Clearwater, Fl. For more information contact Mary Lou Chandler at 407-578-1789 or Karen Carter at 813-799-4977.

5th Joint Clinical Genetics Meeting sponsored by the American College of Medical Genetics and March of Dimes • February 27-March 1, 1998; Los Angeles, CA. Contact: ACMG, 9650 Rockville Pike, Bethesda, MD 20814-3998; Tel: 301-530-7127.