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

Bill Demby

SEPTEMBER 1997

Alliance ALERT

Partnership Needs Resources: The Partnership for Genetic Services Pilot Program is collecting educational and informational materials for educational initiatives in medical schools and managed care organizations. In addition to highlighting information from genetic support groups, the resource packet will include information about peer support contacts, social service networks, community resources and supportive equipment. We need your help to ensure that this packet includes the best current selection of materials. Please contact Nachama Wilker at the Alliance.

Genetic Defect Found That Doubles Risk of One Type of Inherited Colon Cancer: A genetic defect in the APC gene located by researcher Bert Vogelstein of the Howard Hughes Medical Institute at Johns Hopkins University is thought to be the cause of about one in four cases of inherited colon cancer in Ashkenazi Jews. The research suggests that six percent of the American Jews of Eastern European descent harbor this genetic defect and have a 16 percent to 30 percent chance of getting colon cancer during their lifetime. Their chance of getting colon cancer is double that of other Ashkenazi Jews. A new blood test for the defect, which is rare in people who are not Jewish, is available only through the Johns Hopkins Medical Center in Baltimore. Doctors claim that this discovery is a step forward because a positive test result can alert people to the need for regular colon examinations called colonoscopies. There are potential risks, however, that arise from taking this or any genetic test. Because the psychosocial impact on the individual and family members cannot be predicted, pretest genetic counseling is always recommended. There is also the risk of potential misuse of genetic information by health insurers and employers until protective Federal legislation is passed.

American Bar Association Recruiting Disability Lawyers: The ABA's Commission on Mental and Physical Disability Law has launched a national effort to identify lawyers with special skill, sensitivity and interest in disability law. Lawyers are being recruited on the basis of personal experience with disability as well as legal experience. The ABA plans to develop a disabilities lawyers database, create a website, and publish a directory of legal groups that specialize in disabilities law. To join this project, call 202-662-1570 or obtain a Disability Lawyers List Survey Form by Internet at www.abanet.org/disability/home.html.

Charitable Air Medical Transport System in Place: Mercy Medical Airlift is a nonprofit charity that operates air medical transport programs for the benefit of patients nationwide. Through its National Patient Air Transport HOTLINE (NPATH), patient referrals are made to over 45 sources of long distance air medical transport help. Through its "Special Lift" program, large numbers of patients can be flown to and from research or experimental testing programs. MMA works in support of the Air Care Alliance, a league of public benefit volunteer pilot organizations providing air medical transportation for needy ambulatory outpatients. To contact the NPATH Hotline, call 1-800-296-1217.

The Alliance ALERT is a publication of the Alliance of Genetic Support Groups

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New SSI Law Threatens Benefits for Children with Disabilities: The new law requires that children have a "medically determinable physical or mental impairment which results in marked and severe functional limitation" in order to be eligible for the Supplemental Security Income Program (SSI). The Social Security Administration determines the level of severity and eligibility for "childhood disability." The cases of more than 260,000 children will be reviewed. Children most likely to lose benefits are those with mental retardation, tuberculosis, diabetes, organic mental disorders, autism, multiple sclerosis, cerebral palsy, epilepsy, brain injuries and burns.

Identification of Tuberous Sclerosis Gene: The second TSC gene (TSC1) on chromosome 9 has been identified by researcher David Kwiatkowski at Boston's Brigham & Women's Hospital in Boston. In the August 8 issue of Science, he states, "Identification of the TSC1 gene now permits a variety of studies into its biochemical function, which hopefully will lead to specific therapies." The National Tuberous Sclerosis Association (NTSA) has supported research to locate TSC genes since 1984.

Alliance Speaks Out for Consumers: Betsy Anderson, former Alliance VP of Consumers, represented the Alliance at an American Enterprise Institute forum on "Risk, Regulation and Responsibility: Genetic Testing and the Use of Information" on September 4, 1997. Currently Executive Director of the Federation for Children with Special Needs, Betsy participated in a panel discussion on "Genetic Privacy and Research", representing the consumer perspective. Diverse constituencies from the genetics community gathered to address consumer and research perspectives about privacy protection, pending legislation, and research access to tissue samples and live participants.

$3 Million Research Agreement: The National Down Syndrome Society (NDSS) and the National Institute of Child Health and Human Development (NICHD) have formed a partnership to support Down Syndrome research. The NDSS has added $600,000 to $2.4 million in funding from NICHD. Individual researchers and organizations conducting research in areas involving Down Syndrome will be the recipients of this funding. The target date for the first grant awards is late 1998.

"A Question of Genes: Inherited Risks" airs on PBS: Oregon Public Broadcasting is televising a two-hour narrative film on September 16 that explores moral, ethical, and medical questions arising from the opportunities and questions posed by genetic technologies. The film, produced, directed, and written by Noel Schwerin, takes viewers on a journey into the lives of families and individuals in the midst of genetic testing. Seven case studies examine issues ranging from the dilemmas of prenatal testing for parents to those genetic research companies must face. The program is sponsored by SmithKline Beecham and the Department of Energy. Visit the web site at www.pbs.org/gene, and check local listings to confirm date and time.

Legislation Updates
- The Genetic Justice Act was introduced by Senator Tom Daschle of South Dakota and Congresswoman Nita Lowey of New York prior to adjourning for the August recess. Hearings will be held when Congress returns.

- The Slaughter Bill remains pending, and hearings will be held in the fall.

- The House Science Committee has released legislation proposing a prohibition of federal funds for research on the cloning of humans.

- Congress will decide funding for the National Institutes of Health, including the National Human Genome Research Institute, in September.

Tools for Nonprofit Organizations
Developing the Nonprofit Board — Strategies for Educating and Motivating Board Members by Maureen Robinson, Director of Education, National Center for Nonprofit Boards, 16 pages, 1994. $16. Provides the framework for evaluating whether an organization is doing an adequate job of educating and investing in one of its most important assets — the board of directors. Filled with practical advice on board development. Call the National Center for Nonprofit Boards for a complete publications catalog: 1/800/883-6262
**Recommended Reading**

*Stones from the River* by Ursula Hegi: Reviewed by Kimberly Northup

This disturbing novel follows a young German girl's lifelong struggle to come to terms with her genetic condition, achondroplasia. Trudi Montag battles with rejection from her mother and from neighbors, schoolmates and villagers in Germany during the reign of Adolf Hitler. Her story is one of courage and spiritual growth despite and because of her small stature. The book provides insight both on how one who is visibly different feels, and also on how others' reactions can affect his or her self image. The well crafted story allows us to empathize with Trudi by paralleling her differences with those of others to demonstrate the cruelty of prejudice. The symbolism of the persecution of the Jews by the Nazis and of Trudi by society is wonderfully done. The author does not directly provide genetic information on this disorder; however she does dispel many myths. A touching story, *Stones from the River* provides more emotional appeal than scientific data and offers penetrating insights into the nature of prejudice within the framework of German life before and during World War II.

**New Alert Spotlight: "Request 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.

**New Stickler Syndrome Research:** A 5 year study by NIH to connect molecular with clinical findings. If you wish to be considered for participation in this study, contact Dr. Doug Wilkin at 301-435-3497 or by e-mail at dawilkin@nhgri.nih.gov.

**Family Study of Macular Degeneration:** Researchers are studying the role of heredity and other factors in the development of age-related macular degeneration (AMD). Study goals are identification of the gene or genes for AMD and the development of more effective treatments and preventative measures. Participants must be at least 45 years old, both with and without a history of AMD and must meet certain other criteria. Contact Principal Investigator Johanna M. Sedon, MD at Mass. Eye and Ear Infirmary or Harvard Medical School.

**New Clinical Trial for Osteogenesis Imperfecta, Types III and IV:** Dr. Joan Marini at the National Institute of Child and Health Development at the National Institutes of Health is currently enrolling infants under the age of 2 years for a long-term study which will include natural history, growth plateau, and genetic components. Infants who have not yet had collagen evaluation or developed dermal fibroblasts are especially sought. Please call Beth Hopkins, RN, at 301-496-0741 for further information.

**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 •.


**California's Newborn Screening Program: Past Accomplishments and Future Possibilities** • Oct. 8 - 9; Hyatt Regency Hotel, Los Angeles, CA. Call the Department of Health Services, Newborn Screening Program: 510-540-2709.

**Prader-Willi Syndrome Service Providers Conference** • Oct. 16 - 18; Nashville, TN. Contact Prader-Willi Syndrome International Forum, 40 Holly Lane, Roslyn Heights, NY 11577; TEL: 1-800-358-0682 or 516-621-2445.

**Healthy Mothers, Healthy Babies National Biennial Conference** • Oct. 20-22; Baltimore, MD. Contact HMHB, 409 12th St. SW Washington, DC 20024-2188 or call the Conference Hotline: 202-863-2441.

**Living and Learning with Fetal Alcohol Syndrome** • Oct. 24 - 25; Ann Arbor, MI. Sponsored by Parents Supporting Parents (313-662-7291) and the Family Empowerment Network: 1-800-462-5254.
Sixth Annual Conference of the HHT Foundation International • Oct. 24-26; Downtown Atlanta Ramada, Atlanta, GA. For more info, write: HHT Foundation International, PO Box 8087, New Haven, CT 06530 or call 1-800-HHT-NEIW; email: hhtinfo@hht.org.

Fundamentals of Phototherapy Workshop for Nurses and Technicians • Oct. 24 - 26; Scottsdale, AZ. Sponsored by National Psoriasis Foundation, 6600 SW 92nd Ave., Suite 300, Portland, OR 97223-7195. Contact Julie Honse, 1-800-723-9166, ext. 11.

ISONG Educational Conference — “Hand in Hand: Genetic Nursing Education and Practice Roles and Managed Health Care” • Oct. 27 - 28; Radisson Plaza Lord Baltimore Hotel, Baltimore, MD. Contact Dale Halsey Lea, Program Committee Chair, International Society of Nurses in Genetics, at 207-883-4131.

EB Awareness Week is Oct. 27-Nov. 2. Contact the DEBRA office at 212-513-4090 for materials.

Advantages in DNA Diagnostics: Maximizing Genomic Data for Accelerated Discovery of Therapeutic Targets and Expanded Clinical Application • Oct. 27 - 28; Washington, DC. Contact: NMHCC, Inc. 1-888-44-NMHCC.

• National Society of Genetic Counselors, Baltimore, MD, October 28-November 1, 1997.

• American Society of Human Genetics, Baltimore, MD. October 28-November 1.

Neurofibromatosis Symposium (in conjunction with the American Society of Human Genetics) • Oct. 28, 7-10 pm; Convention Center, Baltimore, MD. Contact The National Neurofibromatosis Foundation, 95 Pine Street; 16th floor, New York, NY 10005; email: NNFF@aol.com; Attn: Fran Morris.

National Organization for Rare Disorders/Exceptional Parent Annual Conference • Oct. 30 - Nov. 1; Arlington, VA. Call NORD for more information: 1-800-447-6673.

The Noonan Syndrome Support Groups Meeting • Nov. 1; Baltimore, MD. Contact TNSSG, PO Box 145, Upperco, MD 21155; TEL: 410-239-6926; email: wandar@bellatlantic.net.

Reviewing and Revising the Expedited Categories of Research • Nov. 6; Hyatt Regency, Bethesda, MD. Sponsored by Public Responsibility in Medicine and Research and Tufts University School of Medicine. Contact PRIM&R, 132 Boylston St., Boston, MA 02116; TEL: 617-423-4112.

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.

National Perinatal Association Annual Clinical Conference and Exposition • Dec. 4-7; Tampa, FL. Contact: NPA Office at 813-971-1008.

"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.

"Ethical Research in an Ethical Society", a conference sponsored by Public Responsibility in Medicine and Research and Tufts University School of Medicine, Dec. 8 - 9, 1997; Sheraton Hotel, Boston, MA. Contact PRIM&R at 617-423-4112.