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

Bill Demby

September 1996

This ALERT functions as 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

ALLIANCE FESTIVITIES: Reservations for the 10th Anniversary Benefit Gala on Saturday, October 12 at the Park Hyatt in Washington, DC can be made through the office: 1-800-336-GENE. Tickets are $125 per person. There will be a membership meeting from 10 AM - 12 Noon at the Sheraton City Center Hotel. The hands-on computer workshop and technical sharing session will follow at the Park Hyatt from 1:15 - 5:30 pm. Please finalize your plans as soon as possible as there are many events scheduled in Washington, DC that weekend. Contact the Alliance for a program announcement or hotel information. We look forward to celebrating with you.

Informed Consumers: The September 1996 issue of Consumer Reports on Health focuses on the pros and cons of genetic testing. The lead article advocates the inclusion of family trees in personal and medical records as a way to monitor the presence of hereditary disorders and determine the appropriateness of undergoing genetic testing. Such a recommendation will necessitate a significant change in the way physicians do business. The article cites a study of the medical records of almost 9,000 people which discovered references of family history in only 4 percent. For those whose history indicates a high risk for a particular disorder, the editor warns that it does matter where the testing, if available, is done. His advice is to seek ‘... the genetics division of a university-affiliated medical center, which is likely to be staffed by highly qualified physicians, technicians, and genetic counselors.’

Gene for E.D.A. Found: An international research team has identified the location and structure of the gene that causes the rare disorder known as anhidrotic ectodermal dysplasia or E.D.A. It is just one syndrome in a group of 150 related genetic syndromes known as the ectodermal dysplasias in which two or more of the ectodermal structures (i.e., hair, skin, teeth, glands, and nails) function abnormally. While the gene for E.D.A. has been successfully isolated, researchers do not yet know how the gene functions. The study was supported by grants from a number of sources, including the National Foundation for Ectodermal Dysplasias, which serves more than 1,600 people in 32 countries who are affected by ectodermal dysplasias. The Foundation is located at 219 East Main, P.O. Box 114, Mascoutah, IL 62258-0114.

Important RP Breakthrough: Geneticists have isolated the first gene to cause X-linked retinitis pigmentosa (RP). Called the RPGR gene, it accounts for the most common form of X-linked RP, which is thought to represent about 15 percent of all RP cases and is the severest form of the disease. Visual impairment begins in the first decade of life, leading to severe or complete blindness often by the third or fourth decade. Significant funding was provided by the Foundation Fighting Blindness, Executive Plaza 1, Suite 800, 11350 McCormick Rd., Hunt Valley, MD 21031.

New Role: Elizabeth Thomson, who did an outstanding job as Acting Chief of the ELSI Program, Division of Extramural Research at the National Center for Human Genome Research for the past two years, will continue with the program as Assistant Director of Clinical Genetics Research. We know she will do well and continue to make highly significant contributions to the ELSI effort.

Story Models Needed: The Alliance Partnership for Genetics Education Steering Committee will be meeting in October to plan pilot projects in MARHGN, GLaRGG, and PacNoRGG. We are especially interested in learning about techniques and programs utilized by support groups to facilitate consumers in the telling of their stories and the life impact of their genetic disorder. Contact Nachama Wilker or Donna Foster at the Alliance office: 800-336-GENE. Thank you.

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

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