Hemophilia Breakthrough on PBS

The PBS television network's series, The Infinite Voyage, is on frontiers in science. A PBS crew filmed Alliance member George McCoy for the Voyage episode on frontiers in biology, aired nationally on PBS on June 1, 1988.

Mr. McCoy has hemophilia, a genetic blood clotting disorder. In March, 1987, he became the first person to be treated experimentally with a synthetic, recombinant DNA based Factor VIII blood clotting protein. The research project is being performed by physicians at the University of North Carolina at Chapel Hill. The experimental treatment has been successful during its first year. The genetically engineered protein has so far been safe and therapeutic. A second year of study on an improved version of the protein is planned.

Genetics Institute, Hyland Laboratories, and Baxter Travenol collaborated in development of the protein. The product was tested in mice for safety, and hemophilic dogs for efficacy before the FDA approved the use of a human subject. They chose UNC-Chapel Hill as the pilot site for clinical trials in humans due to the University's long history of research and treatment in hemophilia.

see HEMOPHILIA, page 2, column 2

Alliance Endorses Survey

A project is now underway to learn more about the service needs of individuals with genetic disorders and their families. It builds on the partnership between genetic support groups and health professionals who are providing services to genetic clients. This project is seeking information from members of genetic support groups about their experiences with genetic services and resources, their organization's programs and their recommendations for enhancing the training of professionals.

One questionnaire is being sent to leaders of some of the voluntary genetic organizations across the country that have a service component. A second questionnaire is going to a sample of the members of some of the organizations that belong to the Alliance of Genetic Support Groups. Results of these surveys will be used in developing educational materials that document the unmet service needs of genetic clients and suggest strategies for improving the coordination of genetic and community services.

Directors of this study are Dr. Rita Black, Associate Professor, Columbia University School of Social Work, and Mrs. Joan O. Weiss, Senior Social Worker,

see SURVEY, page 2, column 2
Editorial Comment

We hope you've been wondering why you haven't gotten your issue of Alliance lately. Well, you can stop bothering your postman. We've had a few technical, production-related problems, and have been unable to meet our publication timetable. It looks like we're up and running again, and we intend to stay that way. After all, we've missed you, too!

Please note that we have changed our name. We are now the Alliance of Genetic Support Groups, no longer the Alliance of Genetics Support Groups. A geneticist turned grammarian challenged our use of the plural, and none of us were able to defend it. Thus, we are renamed.

The field of genetics is rapidly gaining the interest of the general press and the public at large. Breakthroughs in genetics research get headlines in daily newspapers. It's not unusual to read or hear about the latest discovery of a genetic marker for a particular genetic disorder, or even that a genetic basis has been found for a disease not previously thought to be genetic. Issues related to gene mapping and sequencing projects; creation of a "super mouse" at Harvard; and concern for the environmental consequences of experimentation in genetic engineering get air time on national radio and television news shows. But have you noticed that this growing news coverage has so far largely omitted the viewpoints of people and families directly affected by genetic disorders themselves?

Congress, governmental policy-makers, the press, and the general public are talking about genetic issues without hearing from the segment of our population most intimately involved with genetics—you and me. Are we making the necessary efforts to participate in this process? Do we write to our governmental leaders or to the press to explain how much we depend on continued research and treatment for genetic disorders? We need to become educated about the complicated scientific and social issues raised by advances in genetic knowledge. Then, we need to be active in expressing our viewpoints as professionals and as people and families affected by genetic disorders. If we don't speak up for ourselves, the major decisions may be made without us...and we may not like the results!

--The Editor.

HEMOPHILIA, from page 1, column 1

In the late 1960s, medical researchers developed a method of preparing freeze-dried concentrates of clotting proteins from human plasma. Since that time, freeze-dried concentrates have been used extensively to control bleeding in people with hemophilia. Although the concentrates have been effective, they also have had drawbacks. They are created by pooling the plasma of many different donors. This pooling means that there can be significant risk of exposing the hemophiliac to such blood-borne viruses as those which cause hepatitis or AIDS. Recent advances in blood processing technology have reduced that risk, and safe new blood products are becoming more widely available.

PBS included the story in its series because researchers involved in the recombinant DNA project hope that the knowledge gained from this breakthrough will help lead to a better control or even, someday, a cure for hemophilia by means of gene therapy.

SURVEY, from page 1, column 2

Division of Medical Genetics, The Johns Hopkins Hospital. The funds to conduct this study have been provided by the March of Dimes Birth Defects Foundation and the Bureau of Maternal and Child Health, Department of Health and Human Services.

Future issues of this newsletter will report on the findings of this project.
From the President

Fortune smiles seldom enough on any of us that a glimmer of its presence on the horizon is by necessity treated with the same eagerness and anticipation as if the actual prize were at hand. Such a glimmer exists in our world today. Some of it may be there of our own doing, our own planning and work; some of it, more likely most of it, may be by reason of an evolution, a counter-thesis long overdue which is now awakening portions of the collective conscience of our society.

We see its promise in the continuing strength and vitality of many of our organizations which, born of personal need, now serve to forward a public cause. We see it in the legislative arena, which recognizes the long dormant strength of the millions of Americans affected by the many forms of genetic disease that exist. We see it in the recognition of public policy, which no longer looks simply at the plight of a child who is adversely affected, but rather at the growth of an individual throughout the span of his or her life. We see it ever so slowly in the media, which has begun to recognize the fact that stereotypes are not appropriate ways of talking about people. We see it in the recognition by public agencies of the movement toward self-help and the personal responsibility which that implies toward our own care and our own well being. We see it in the increased awareness by the general public that we will not accept the role of second class, disenfranchised citizens.

Some of what we see comes in the form of laws. Some of what we see comes in the form of rhetoric of public officials. Some of what we see comes in the softening of lines which have long divided our community from the rest of the American people. But mostly what we see is an emerging ability of those of us who have come from specific constituent organizations to recognize our commonalities and realize that we have been too long divided, that we have been too long distracted by those divisions from the goals we all seek and from the means to achieve those goals.

We are perhaps now at a point in time when this glimpse on the horizon can be pulled a bit closer. We are perhaps at a point when we can strive by mutual effort to achieve for our own constituencies greater progress than any of us alone is able to do. If there has ever been a time when the right pegs were in the right places, when the forces needed to impact upon our society have been strong enough and when the means and resolution to achieve these things has ever existed, now is indeed the time. The enhancement of the quality of the lives of those whom we serve is finally being recognized -- not as a gift of charity to be accepted as alms or a result of advocacy leading to legislation -- but as a right that exists for each and every American and is ours, not to ask for, but to demand and expect.

We as representatives of the millions of Americans affected by genetic conditions must now recognize that the role we have to play must not be a timid one. Policy cries to be structured and directed, and we must impact as best we can on that process. Public acceptance and knowledge is ours to influence if we so choose and we are judicious and perseverant. The many organizations that you see listed in this newsletter and the many others with whom they interact are a part of the process that can and will result in the drawing nearer of that glimmer on the horizon of a fortune too long withheld, of a promise too long denied.

Whatever work there is left to be done -- indeed there are volumes of that-- that which is within our grasp right now is the means by which each of us can contribute more effectively to our overall cause. As you look around you at colleagues and coworkers and organizational counterparts, realize that these are not your adversaries; these are allies, resources that with thoughtful collaboration will make each of our jobs that much easier and each of our goals sooner realized. What we have to gain far outweighs anything we might risk and what we need to do demands that we try.

--Greg Weigle, President.
In the previous issue of Alliance, this column was initiated "to help our readers become informed consumers of genetics services." In this issue we will begin with a brief discussion of some basic service needs of persons who have a genetic disorder and/or families of affected persons and what resources should be available to meet these needs.

When the diagnosis of a genetic disorder is first given to you as an affected adult or the parent of an affected child, you must begin to adapt to a new reality. Many factors impact upon this process, including the personalities of all the people involved; the age of the affected person at time of diagnosis; severity of any related disability; availability of medical care; economic conditions and size of the family; you and your family's understanding of and attitude toward the genetic disorder; and the attitudes of health care deliverers and the community at large.

Even though for each person and family the experience is unique, certain feelings, reactions and service needs may be similar to those of others who have learned to cope with the presence of a genetic condition and its implications. The needs to be addressed by you and your family may fall into many areas, such as medical support, information, therapy, behavior management, self-help, social and emotional progress, recreation, living arrangements, family support, finances, legal services, and vocational rehabilitation.

Availability of suitable medical care is usually the first and main concern. Where can you locate a physician who is knowledgeable about your genetic condition? Is the diagnosing physician knowledgeable enough to be the primary care physician (the one who will provide ongoing care?) If not, how can you or your doctor find such a resource? How close is the nearest knowledgeable health care to your home? Are there groups which can help you or your family to make good use of the needed clinical care?

Local or national medical societies, as well as genetic support groups, either single- or cross-disorder groups, (such as the Alliance of Genetic Support Groups or the National Organization for Rare Disorders), can assist with location of health care providers, educational materials, or other important health care information. Medical schools and local hospitals usually have a public relations or community service division which can also help locate experienced medical care providers.

Suitable medical care is only one part of the total picture. Other concerns emerge which reveal the need for a broader, more comprehensive approach to care. Comprehensive care for a genetic disorder has the goal of supporting satisfying and productive lives for affected people and their families. Clinical treatment, research, and psychosocial services are all necessary components of comprehensive care. It involves not only the physician but also in many cases, a genetic counselor, who will assist in understanding the disorder and its inheritance patterns; a nurse, who is important as an educator as well as a caregiver; and the clinical social worker, whose role includes supportive counseling and the identification of resources for other psychosocial needs.

Other types of professionals may have a role in the ongoing care and education of you and your family. Vocational rehabilitation counselors, psychologists, psychiatrists, or other professionals may be of value at various times.

Many of these services are presently available in certain geographic areas. Major hospitals, social service agencies, other community agencies and individual private practices are a few resources that one might expect to explore.

see WHEEL, page 5, column 1
How do you and your family identify your needs? What can you do to find quality services? What can you do if they're not available where you live? Will you and your family automatically receive referrals when needed?

Subsequent issues of the Squeaky Wheel will focus on these questions and the professionals who are involved with the delivery of care to families affected by genetic disorders.

(Some information for this article was taken from the News Digest published by the National Information Center for Handicapped Children and Youth, November, 1985 issue.)

--June Vavasseur, MPH, for the Family Services Committee of the Alliance of Genetic Support Groups.

Conference Calendar

June 16-18, 1988
Maple Syrup Urine Disease Symposium
Location: Ephrata, PA
Contact: MSUD Foundation
Mr. and Mrs. D.G. Koons
1045 Piketown Rd.
Harrisburg, PA 17112
(717) 469-7167

June 22-25, 1988
Inward Bound/Outward Growth
Location: Saint Paul, MN
Contact: Spina Bifida Assn. of America
700 Rockville Pike, Suite 750
Rockville, MD 20852
(800) 621-3141

June 22-25, 1988
Tenth Annual Prader-Willi Syndrome Association Conference
Location: Louisville, KY
Contact: Prader-Willi Syndrome Assn.
5515 Malibu Dr.
Edina, MN 55436
(612) 933-0113

June 23-26, 1988
Fourth Annual Cornelia deLange Syndrome Convention: Communication and Families-Opening Doors to Understanding
Location: Chicago, IL
Contact: Cornelia deLange Syndrome Foundation
60 Dyer Ave.
Collinsville, CT 06022
(800) 223-8355

July 8-10, 1988
International Symposium on the Marfan Syndrome
Location: Baltimore, MD
Contact: Program Coordinator
Office of Continuing Education
Johns Hopkins Medical Institutions
Turner 22, 720 Rutland Ave.
Baltimore, MD 21205
(301) 955-2959

July 10-13, 1988
Heritable Disorders of Connective Tissue and Skeletal Dysplasias
Location: Baltimore, MD
Contact: Program Coordinator
Office of Continuing Education
Johns Hopkins Medical Institutions
Turner 22, 720 Rutland Ave.
Baltimore, MD 21205
(301) 955-2959
Meet ASHG

The American Society of Human Genetics (ASHG) is a nonprofit organization, dedicated to promoting contact among investigators in the field of human genetics; to encouraging and integrating research in human genetics; and to dealing with other issues related to human genetics.

Membership in the ASHG is open to anyone residing in the Americas who is interested in research or problems pertaining to human genetics. Corresponding membership is available to persons outside the Americas. Membership has grown from 220 in 1948 to 2,800 in 1986. ASHG membership represents such fields as biochemical genetics, cancer genetics, clinical genetics, cytogenetics, differentiation and development, gene mapping and linkage, genetic counseling, molecular genetics, genetic epidemiology, population genetics, and prenatal diagnosis.

The ASHG publishes a monthly research record, The American Journal of Human Genetics, which relates to heredity in humans and to the applications of genetic principles in medicine, psychology, anthropology, social sciences, and areas of molecular and cell biology relevant to human genetics. Other publications include a biennial Membership Directory, a brochure on Careers in Human Genetics, and a softcover directory of graduate-level training programs, Guide to Human Genetics Training Programs in North America.

Alliance Goals

The Alliance of Genetic Support Groups is dedicated to fostering a partnership among consumers and professionals in order to enhance education and service for and represent the needs of families and individuals affected by genetic disorders.

Education--

The Alliance is working to increase awareness about genetic disorders...

by developing and disseminating information to enhance public and professional awareness of available resources and referrals, and...

by encouraging communication among support groups and enhancing awareness of cross-disability similarities and identifying resources.

Services--

The Alliance is working to improve the availability and appropriateness of genetic services...

by identifying gaps in existing services, and...

by developing model programs or recommendations to fill those gaps.

Representation--

The Alliance is working to represent the common needs and concerns of its constituency...

by providing a forum for identifying common issues, and...

by establishing communications channels with government agencies, professional groups, service providers, and other consumer organizations.

Memberships/general correspondence to:
ALLIANCE OF GENETIC SUPPORT GROUPS
38th and R Streets, NW
Washington, DC 20057
(202) 625-7853

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Information in this newsletter may be reprinted with credit given to source.
Display Table for Members

The Alliance of Genetic Support Groups will have a table in the display area at the National Society of Genetic Counselors Meeting, followed by the American Society of Human Genetics Conference, both to take place in New Orleans in October, 1988. At our table any genetic support group that is a member of the Alliance may display its brochure or other pertinent literature. We will be contacting each of our member organizations at a later date to determine interest in this opportunity for educating interested health professionals about your particular voluntary organization and the genetic disorders which you represent.

Alliance Symposium Announced

The Alliance of Genetic Support Groups announces a symposium to explore strategies of "Empowerment" through knowledge and skill-building for individuals, families, and professionals involved with genetic disorders.

Date: November 4-5, 1988
Place: Washington, DC
Stay tuned to your mail for more information!