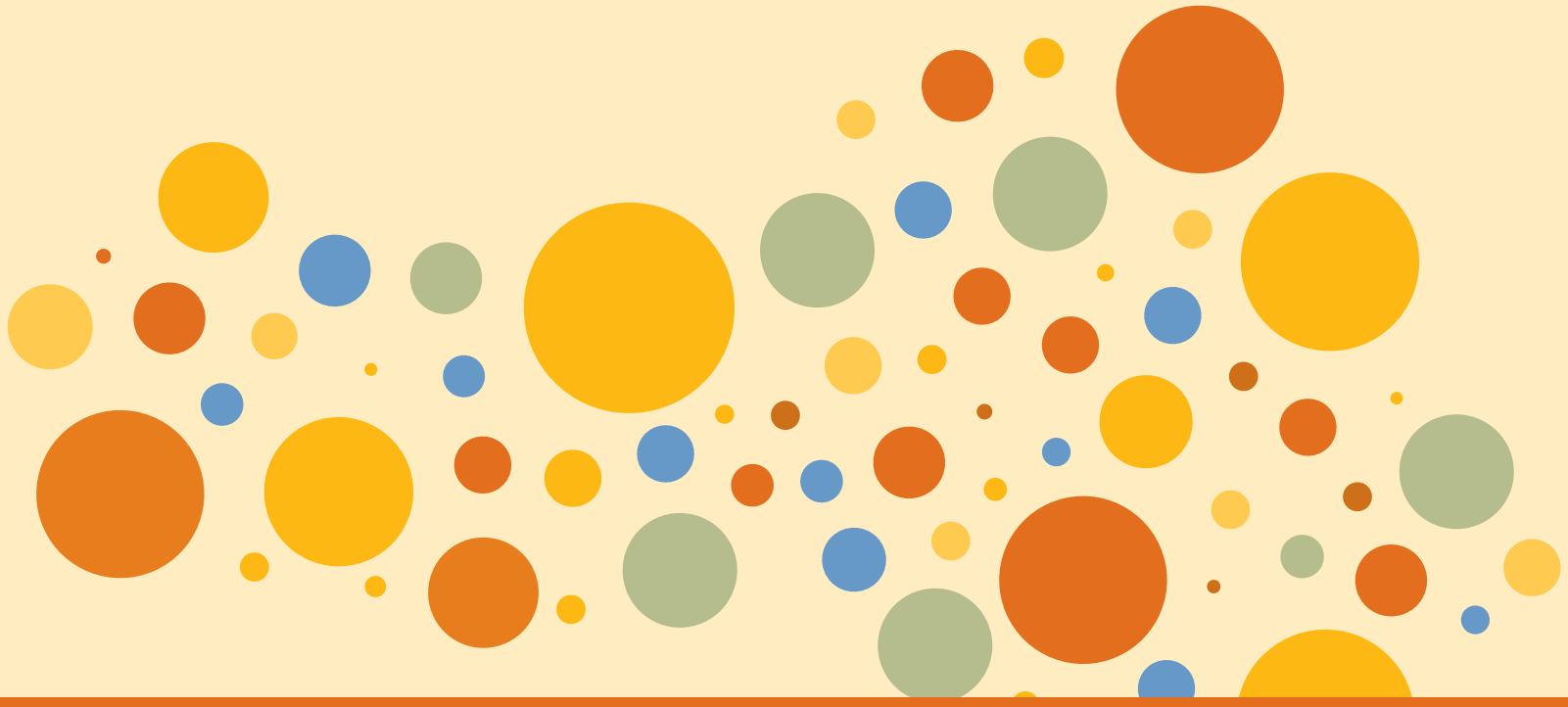
The Year of the

ADVOCATE



GENETIC ALLIANCE

2007 ANNUAL CONFERENCE

CONTENTS

WELCOME TO THE YEAR OF THE ADVOCATE!

We give you a weekend of deep learning, rich interaction, and fun! We celebrate advocates this weekend—a community of individuals joined together to change the world around us.

Dig into the workshops, the talks, the dinners, the networking—even the Pilates and the morning reflection times! Don't forget to check out all of our new offerings, and take advantage of the old standbys—the awards dinner, the singing, "after hours" networking, and the staff raffle. Board and staff are here to serve you, so please look for their ribbons if you need help or information. Engage with one another and be awed by the dedicated individuals and organizations around you, as I am each year.

We look forward to hearing and seeing Joanna Rudnick and Newt Gingrich, our plenary speakers. We celebrate our award winners—Abbey Meyers, Ronald Zuker, Amy Marcus, and Affymetrix. We are delighted to have our founder, Joan Weiss, with us for the awards dinner.

Most especially, thank you for all you bring to the conference—it is a clear instance of the whole being much greater than the sum of its parts!

Happy conference,

Sharon Terry, President & CEO

Friday, July 27th

8:00 AM – 5:30 PM

GENETIC ALLIANCE INSTITUTE FOR ADVOCACY

Organizations in Action Glen Echo
Facilitated by **Elizabeth Burden** and **Lisa Wise**
Featuring **Jonathan Martin,** National Marfan Foundation and
Catherine Burzio, Chromosome 18 Registry & Research Society

Transformational Leadership in Action Forest Glen
Facilitated by **Gene Early** and **Genetic Alliance Board of Directors**

Research in Action White Flint Amphitheatre
Moderated by **Steve Groft,** Office of Rare Diseases, NIH and
Claire Driscoll, National Human Genome Research Institute, NIH

5:30 PM – 6:15 PM

Conference Orientation & Conference Fellows Meet and Greet
White Flint Amphitheatre

6:15 PM – 7:15 PM

Exhibits Gallery and Reception (cash bar) Adjacent to Grand Ballroom

7:15 PM – 9:30 PM

Opening Dinner Grand Ballroom
State of the Alliance: **Sharon Terry,** Genetic Alliance
In the Family: **Joanna Rudnick,** Kartemquin Films

10:00 PM – 12:00 AM

Genetic Alliance 'After Hours' (cash bar) Linden Oak

KEY: Education Track | Policy Track | Research Track | Service Track

Saturday, July 28th

7:15 AM – 8:00 AM

Pilates Brookside A

Morning Reflection (secular) Brookside B

8:00 AM – 7:00 PM

Exhibits Open Adjacent to Grand Ballroom

8:00 AM – 9:00 AM

Networking Breakfast Grand Ballroom

9:00 AM – 12:30 PM

Curbside Consult Brookside B

Collaboration in Researching Genetic Conditions: **Steve Groft,** Office of Rare Diseases, NIH

Fundraising and Friendraising: **Elizabeth Burden**

How to Talk to Affected Individuals and Their Families About Genetic Disease: **Heather Ferguson,** Genetic Alliance

Taking Care of Yourself in the Caretaking: **Brigid Guttmacher,** Counselor

9:00 AM – 10:30 AM

WORKSHOPS: Session I

Improving the Quality and Accuracy of Your Educational Materials: Using Tools Developed by the Access to Credible Genetics Resources Network Brookside A
Joseph McInerney, National Coalition for Health Professional Education in Genetics
Kate Reed, National Coalition for Health Professional Education in Genetics
Meredith Weaver, University of Maryland

The Heredity Project: Promotion Through Genetic Testing Oakley
Vicki Park, University of Tennessee Health Sciences Center

Resource Repository: One Stop Shopping Great Falls
Karen White, Genetic Alliance
Audrey Gordon, Progeria Research Foundation

Regulation of Genetic Testing Glen Echo
Janet Woodcock, Food and Drug Administration
Kathy Hudson, Genetics and Public Policy Center

How to be Effective Research Advocates Forest Glen
Deborah Collyar, Patient Advocates in Research
Joyce Graff, VHL Family Alliance

Widening Perspectives on Advocacy: From Visionary Leaders to a Coalition of Grassroots Organizations Linden Oak
Martha Carvalho, Brazilian Genetic Alliance, Associacao X Fragil do Brasil

Saturday, July 28th continued

10:30 AM – 11:00 AM

Coffee Break Adjacent to Forest Glen/Glen Echo

11:00 AM – 12:30 PM

WORKSHOPS: Session II

Popular Education in Genetics and Reproductive Health: Perspectives from the Latino/a and Asian American Communities in New York City Brookside A
Andel Nicasio, Dominican Women's Development Center
Jesus Sanchez, Dominican Women's Development Center
Deborah Hong, Charles B. Wang Community Health Center

Keep It Simple: Conveying Disease Information to Your Target Audience
Linden Oak
Paula Raimondo, University of Maryland
Meredith Weaver, University of Maryland
Kate Reed, National Coalition for Health Professional Education in Genetics

Preterm Birth and Birth Defects Great Falls
Siobhan Dolan, Einstein Medical Center & Sarah Lawrence College

Making Clinical Trial Information Available to Your Members Forest Glen
Robert H. Shelton, Private Access

Registries, Databases, and BioBanks Glen Echo
Liz Horn, National Psoriasis Foundation
Connie Lee, Angioma Alliance

Genetic and Rare Diseases Information: Information for the Public Oakley
Janine Lewis, Genetics and Rare Diseases Information Center

12:30 PM – 1:30 PM

Buffet Lunch Grand Ballroom
Optional Regional and Issue-Based Networking

1:30 PM – 3:00 PM

WORKSHOPS: Session III

Evenings with Genetics: A New Community Seminar Series Brookside B
Susan Fernbach, Baylor College of Medicine

Educating Policymakers: Promoting Cardiovascular Disease Prevention and Muscular Dystrophy Registries Forest Glen
Apryl Brown, Detroit Medical Reserve Corps
Patricia Furlong, Parent Project Muscular Dystrophy, Genetic Alliance Board of Directors

KEY: Education Track | Policy Track | Research Track | Service Track

1:30 PM – 3:00 PM

WORKSHOPS: Session III continued

Managing Incidental Findings Linden Oak
Jordan Paradise, University of Minnesota Law School
Suzanne Sobotka, University of Minnesota Law School
Bonnie LeRoy, University of Minnesota

Networks: Tools for Transformation Glen Echo
Sharon Terry, Genetic Alliance
Amelia Chappelle, Genetic Alliance

How Do We Serve Ourselves in the Midst of Serving? Oakley
Brigid Guttmacher, Counselor

Race, Testing, Treatment, and Cost: Does It Matter? Brookside A
Penny Kyler, Genetic Services Branch, MCHB, HRSA
Vence Bonham, Jr., National Human Genome Research Institute
Ursula Tsosie, Urban Indian Health Institute
W. Nicholson Price, Graduate Student, Columbia University

3:00 PM – 3:30 PM

Coffee Break Adjacent to Forest Glen/Glen Echo

3:30 PM – 5:00 PM

WORKSHOPS: Session IV

Disease InfoSearch: How Can I Get the Word Out About My Organization? Glen Echo
Lisa Forman Neall, National Center for Biotechnology Information, NIH
Helen Travers, Genetic Alliance & Genzyme
Karen White, Genetic Alliance
Heather Ferguson, Genetic Alliance

Can Nurses Close the Gap in Genetic Knowledge Delivered by Healthcare Providers? Oakley
Dale Lea, National Human Genome Research Institute, NIH
Kevin Lewis, Colon Cancer Alliance

The Imperative of Understanding the Genetics of Minority Populations Brookside A
Joseph J. Jacobs, Abbott Molecular

Issues in Newborn Screening: False Positive Screens and Carrier Identification Forest Glen
Natasha Bonhomme, Genetic Alliance
Penny Kyler, Genetic Services Branch, MCHB, HRSA
Andrea Williams, Children's Sickle Cell Foundation

Saturday, July 28th continued

3:30 PM – 5:00 PM

WORKSHOPS: Session IV continued

Facilitating Parent-Primary Care Physician Partnerships in Genetics Linden Oak
Paula Goldenberg, Children's Hospital of Philadelphia
Donna McDonald-McGinn, Children's Hospital of Philadelphia
Lisa Jennings, Northeast Velo-Cardio-Facial Syndrome Support Group

The Impact of Living with a Genetic Condition in 2007 Brookside B
Sandy Gordon, Trimethylaminuria Foundation

6:00 PM – 7:00 PM

Reception (cash bar) Adjacent to Grand Ballroom
Featuring Live Music from **Mark Puryear and Paul Watson**

7:00 PM – 10:00 PM

Awards Banquet Grand Ballroom
Hosted by **Suzanne Richard,** Open Circle Theater
Honoring Award Recipients: **Abbey Meyers, Ronald Zuker, Amy Marcus,** and **Affymetrix**
Closing: **Francis Collins,** National Human Genome Research Institute, NIH

10:00 PM – 12:00 AM

Genetic Alliance 'After Hours' (cash bar) Linden Oak

KEY: Education Track | Policy Track | Research Track | Service Track

Sunday, July 29th

7:00 AM – 7:45 AM

Morning Service (all faiths welcome) Brookside B
Led by **Francis Collins**

Pilates Brookside A

7:45 AM – 8:30 AM

Networking Breakfast Grand Ballroom

8:00 AM – 10:30 AM

Exhibits Open Adjacent to Grand Ballroom

8:30 AM – 10:00 AM

WORKSHOPS: Session V

Teaching and Learning Together Linden Oak
Joann Boughman, American Society of Human Genetics
Greg Feero, National Human Genome Research Institute, NIH

Community Centered Family Health History Oakley
James O'Leary, Genetic Alliance
Claudia Petruccio, Institute for Cultural Partnerships
Alejandra Gepp, National Council of La Raza

Collaboration, Education, and Test Translation (CETT) Glen Echo
Andy Faucett, Emory University
Melissa Dempsey, University of Chicago
Vicky Whittemore, Tuberous Sclerosis Alliance
Patricia Furlong, Parent Project Muscular Dystrophy, Genetic Alliance Board of Directors

Case Study of Newborn Screening Information Integration Brookside B
John Adams, OZ Systems
Lura Daussat, OZ Systems

Transition to Self-Management for Individuals and Families Forest Glen
Paula Goldenberg, Children's Hospital of Philadelphia
Christie Falco, Duke University

10:00 AM – 10:30 AM

Coffee Break Adjacent to Grand Ballroom

10:30 AM – 12:30 PM

Closing: Keynote Address Grand Ballroom
Genetic Diagnostics as the Base of a 21st Century Intelligent Health System:
Newt Gingrich, Founding Director for Center for Health Transformation
Transformational Leadership: **Sharon Terry,** Genetic Alliance

• • • Workshop Descriptions

Institute for Advocacy

Friday, 8:00 AM – 5:30 PM

ORGANIZATIONS IN ACTION Glen Echo

This daylong comprehensive, hands-on training will give advocacy group leaders the skills to grow their organizational capacity. This year's Organizations in Action workshop will focus on organizational assessment, strategic planning, and communication techniques that are effective with all audiences including providers, members, patients, politicians, and the media.

TRANSFORMATIONAL LEADERSHIP IN ACTION
Forest Glen

Advocacy organizations and other nonprofits have historically embraced the "hero model." The leadership has been heroic—and borne great burdens. Though initially quite successful, this model has caused a number of problems: lack of incentive for others to bear the burden, difficulty with succession in the organization, burnout for the leaders, isolation for the leaders, and a narrowness the organization cannot afford. Even the national media has noticed that the baby boomer leaders have burned out and we are suffering from a lack of leadership for the nation's nonprofits.

It is time to move to a model of transformational leadership—leading from one's strengths in a collaborative way. This style of leadership liberates profound skills and abilities in the individual, but also creates a connection with the other potential leaders in the organization that bring into focus the answers to the questions "What matters? What is essential?" This style of leadership also liberates the skills of those around the leader, and pushes organizations to the next level.

Further, the essence of community—understanding why we are gathered to help a disease, a cause, an issue—is nurtured in this new style of leadership, resulting in an ability to work productively across multiple agendas, unifying the effort.

This workshop will provide a hands-on experience with proven tools to transform the way you lead. Bring yourself, your vision, your passion, and your problems. There will be no time for whining, there will be no space for limiting one's potential, there will be nothing but truth.

RESEARCH IN ACTION White Flint Amphitheatre

From Bench to Bedside to Practice: A Practical Course – Moving Toward Treatment

Sponsored by the Office of Rare Diseases, NIH, DHHS

8:30 AM Welcome and Overview

Steve Groft, Director, Office of Rare Diseases, NIH & **Claire Driscoll,** NHGRI, NIH

8:45 AM Diagnosis

The Testing Landscape: What Labs Need to Do – **Bin Chen,** CDC

9:05 AM So You have the Gene, What Now?

Overview of the Pipeline – Translation 101 – **William Gahl,** Clinical Director, NHGRI, NIH

Nuts and bolts and comparative genomics – **Elliott Margulies,** NHGRI, NIH

Large-scale screening – **Jim Inglese,** NHGRI, NIH

10:15 AM Shifts in the Testing Paradigm

Collaboration Education and Test Translation Program – **Andy Faucett,** CDC & Advocates

10:45 AM Break

11:10 AM The Power of One
Challenges of social science research into rare genetic conditions and innovative methodological approaches - **Barbara Biesecker,** NHGRI, NIH & **Janine Janosky,** University of Pittsburg

11:45 AM Discussion of Morning Issues

12 Noon Lunch and Networking

1:00 PM Finding and Capturing Phenotypes
Phenotype/Genotype Correlations in large-scale populations – **Teri Manolio,** NHGRI, NIH

IT Infrastructure Needs – **Jim Ostell,** NCBI, NLM, NIH

2:15 PM Rare Disease Clinical Research Network
Challenges and Portability – **Steve Groft,** ORD, NIH and Members of the Network

2:45 PM Break

3:00 PM eyeGENETM
About the National Eye Institute Genotyping Network – **Santa Tumminia,** NEI, NIH

3:20 PM Multi-Advocate Collaboration and Public Private Partnership
Ron Bartek, Friedreich's Ataxia Research Alliance (FARA) & **Guy Miller,** Edison Pharmaceuticals/ University of Pennsylvania (invited)

3:40 PM Facilitated Discussion of the Opportunities and Challenges of Accelerating Research
Alan Guttmacher, NHGRI, NIH

4:30 PM – CLOSING

Workshops: Session I

Saturday, 9:00 – 10:30 AM

Education Track

Improving the Quality and Accuracy of Your Educational Materials: Using Tools Developed by the ATCGRN Brookside A

This session will introduce participants to two tools designed to guide users in assessing the quality and completeness of information contained within educational materials. We will discuss how they have been used to determine the quality of educational materials and will demonstrate ways in which they have been used to improve materials. Participants will be asked to use the tools to assess their own materials. We encourage individuals with varying levels of experience to attend and to bring one of their organization's short educational booklets or brochures.

Education Track

The Heredity Project: Promotion Through Genetic Testing Oakley

The Heredity Project is an educational initiative to help the public understand the relationships between genes, common diseases, and healthcare. We will look at how genes travel through families and how different kinds of risk interact to cause health problems. Recent research in type-2 diabetes will serve as an example of the contribution of genes to overall risk. During the workshop, participants will be invited to use wireless remotes to provide their input, which will be used for further development of the project.

Workshop Descriptions

Workshops: Session I
(continued)
Saturday, 9:00 – 10:30 AM

Education Track
Resource Repository: One Stop Shopping
Great Falls
The new Genetic Alliance Resource Repository houses valuable resources on a wide range of topics, including fundraising, genetic testing, advocacy at the state and federal levels, and media strategies. Learn how to access and contribute to this treasury of information for and by the advocacy community at this interactive workshop.

Policy Track
Regulation of Genetic Testing Glen Echo
Genetic tests are the primary genetic service at this time. The regulatory system must encourage safe, accurate, and accessible genetic tests. At the same time, regulators must avoid placing too heavy a burden on those who create, manufacture, and provide genetic testing services to the public. Policymakers must strike an appropriate balance. This workshop will describe the current system and invite vigorous discussion of its strengths and weaknesses. Recent guidances, proposed legislation, and petitions will be discussed.

Research Track
How to be Effective Research Advocates
Forest Glen
You know more than you think! Even though you may not know how to extract DNA or breed mice, you can bring a critical ingredient that is missing from many research projects—the perspective of the patient. This session will discuss how you can learn enough about the science to listen intelligently and identify patient aspects the scientists may have missed. An increasing number of federally funded research projects are encouraging involvement of patient advocates—be ready to help in this critically important way!

Services Track
Widening Perspectives on Advocacy: From Visionary Leaders to a Coalition of Grassroots Organizations
Linden Oak
This presentation will provide diverse perspectives on advocacy. We will start with self-reflection, when a genetic condition is diagnosed in our own family, and move to a wider and global understanding of the realization that as advocates we play an important role in raising the issues that concern people living with a broad range of genetic conditions. The process involves increasing numbers of people from different areas and at all levels—regional, national, and international.

Workshops: Session II
Saturday, 11:00 AM – 12:30 PM

Education Track
Popular Education in Genetics and Reproductive Health: Perspectives from the Latino/a and Asian American Communities in New York City
Brookside A
Funded by the March of Dimes and the Health Resources Services Administration, the Consumer Genetics Education Network (CGEN) Project aims to provide culturally appropriate information about genetics, family health history, and reproductive health in an effort to empower ethnic minority communities and to help them make informed decisions about their health. The Dominican Women's Development Center and Charles B. Wang Community Health Center will share their unique experiences in developing grassroots, culturally appropriate educational strategies for the Latino/a and Asian American communities in New York City.

Workshop Descriptions

Education Track

Keep it Simple: Conveying Disease Information to Your Target Audience Linden Oak

The purpose of this workshop is to introduce the tool and checklist we have developed for communicating high quality health information in simple language. We will describe how we envision the checklist can be used and give examples. We will provide attendees an opportunity to work with the tools to translate complex or specialized information into plain language, and to redesign educational materials using the checklist. We encourage individuals of all different levels of experience to attend to encourage a discussion of best practices.

Policy Track

Preterm Birth and Birth Defects Great Falls

The relationship between birth defects and prematurity is complex, and it is increasingly clear that both result from the interaction of genetic and environmental factors. Addressing prematurity and birth defects simultaneously provides the impetus to help reduce underlying, shared risk factors. It sheds new light on familiar issues of fostering healthy pregnancies by providing high-quality preconception, prenatal and interconception care. At the same time, further research into the genetic factors that contribute to both birth defects and prematurity is needed and current guidelines for screening for adverse birth outcomes will be presented.

Policy Track

Making Clinical Trial Information Available to Your Group's Members Forest Glen

Many disease advocacy groups would love information and links to be available on clinical trials and research investigations that are highly relevant to their members' specific needs. Yet few organizations have the staff, time, or budget to research, set up, and maintain such a service on their website; and those who have tried have learned that the information quickly becomes dated without considerable ongoing maintenance. This workshop will consider the technical and ethical issues surrounding the need to help protect your members' personal privacy preferences, and presents an example of a new solution for disease advocacy organizations.

Research Track

Registries, Databases, and BioBanks Glen Echo

Are you interested in starting a BioBank for your organization? Are you wondering how to make it reality? Learn how to create a BioBank for your organization. This workshop will outline the steps to join the Genetic Alliance BioBank, get approval for all regulatory documentation, create a clinical questionnaire, and develop recruitment and retention strategies. This workshop will also discuss fundraising opportunities and challenges encountered.

Services Track

Genetic and Rare Diseases Information Center Oakley

During this presentation, staff from the Genetic and Rare Diseases Information Center (GARD) —a collaborative effort of the National Human Genome Research Institute and the Office of Rare Diseases, National Institutes of Health—will discuss their experience with providing the public with comprehensive, individually tailored responses to questions about genetic and rare disease concepts in English and Spanish. We will also talk about the challenges of addressing the public's ever-evolving questions and concerns about new developments in genetic technologies.

Workshop Descriptions

Workshops: Session III

Saturday, 1:30 – 3:00 PM

Education Track

Evenings with Genetics: A New Community Seminar Series Brookside B

This seminar series offers the community the most current information on healthcare, education, research, and resources regarding a variety of genetic diseases. Genetic faculty members present information in lay terms and an individual or parent of a child impacted by the condition shares his or her unique viewpoint on it. The seminars are held monthly. Attendees include families, school nurses, teachers, speech pathologists, medical/nursing students, and faculty. Positive participant evaluations, as well as extensive travel by some participants to attend the series, affirm the value of the program.

Policy Track

Educating Policymakers: Promoting Cardiovascular Disease Prevention and Muscular Dystrophy Registries Forest Glen

Policymakers are educated by knowledgeable and proactive advocates. This workshop will offer two examples of educating policymakers to focus efforts on specific conditions. We will discuss the American Heart Association's work to increase research and education for cardiovascular disease, as well as Parent Project Muscular Dystrophy's visionary program to establish an international registry in order to illustrate tools and techniques for advocates in the policy area.

Research Track

Managing Incidental Findings Linden Oak

An incidental finding (IF) is an unexpected discovery during a research study that may have clinical implications for an individual research participant, raising critical questions for researchers, advocacy groups, IRBs, policymakers, and the general public. Although beyond the scope of the research study, an IF may be of grave clinical importance and may be definitively diagnosed if followed up. We will overview critical ethical, legal, and scientific issues; present empirical research findings; and suggest recommendations for guidance.

Research Track

Networks: Tools for Transformation Glen Echo

Traditional organizational design emphasizes rigid hierarchical levels while paying little attention to informal relationships within and between organizations. Recently, social networks have widely captured the attention of managers and executives. Organizational network analysis attempts to discover and analyze dynamic social networks to further strategy and performance. After truly understanding a network, organizations can more easily make appropriate connections to fulfill needs and reach goals. Genetic Alliance is applying organizational network analysis to fulfill our mission of capacity building within the genetics community.

Services Track

How Do We Serve Ourselves in the Midst of Serving? Oakley

With days filled caring for family, work, advocacy, and everything else life brings, there is little time to reflect on challenges met, goals accomplished, or our own needs. How do we take care of the caring part of ourselves when we are so busy caring for others? This workshop provides an opportunity to learn and support each other by sharing challenges and solutions.

Services Track

Race, Testing, Treatment and Cost: Does It Matter? Brookside A

Over the last three years, the Maternal and Child Health Bureau/Health Resources and Services Administration (MCHB/HRSA) and the National Human Genome Research Institute's (NHGRI) community-based grants have heard from consumers regarding testing, treatment, and cost. In 2000, MCHB/HRSA focused on the inclusion of underserved and under-represented communities in order to develop a literate genetics citizenry; NHGRI focused on the participation of minorities in genomic research. Two questions have arisen: "Do my genes tell me who I am?" and "Does it make a difference?" This panel will explore these issues from a minority perspective and elucidate the process of informed decision-making.

Workshops: Session IV

Saturday, 3:30 – 5:00 PM

Education Track

Disease InfoSearch: How Can I Get the Word Out About My Organization? Glen Echo

Learn how to reach the public with information about your organization and the conditions your organization represents using Disease InfoSearch, a free service of Genetic Alliance. This workshop will show you how easy it is to partner with Disease InfoSearch to highlight the most up-to-date and accurate information about your condition.

Policy Track

Can Nurses Close the Gap in Genetic Knowledge Delivered by Healthcare Providers? Oakley

Participants will learn the results of a recent survey of Genetic Alliance members' perceptions of provider knowledge in genetics. Only 17 percent of respondents indicated that their healthcare providers' knowledge of genetics was "good to excellent." Can nurses close the gap in genetic knowledge delivered by health care providers? Presenters will share personal stories about past and present nursing provision of genetic services. Discussion about whether nurses have played a role in providing participants with genetic services will follow. The Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics will be presented to further discussion.

Policy Track

The Imperative of Understanding the Genetics of Minority Populations Brookside A

This session will focus on the need for disenfranchised populations to become more involved and proactive in the research and clinical application of genetics in order to help address health disparities. Documented genetic profiles of patients on the Navajo reservation will be discussed. Some of the issues to be raised include cultural barriers to participation in research, traditional peoples' knowledge of genetics, possible dispelling of stereotypes, and strategies to enhance genetic understanding.

Services Track

Issues in Newborn Screening: False Positive Screens and Carrier Identification Forest Glen

False-positive results and carrier identification are two familiar challenges associated with newborn screening (NBS). Now, advances in technology and an increase in information revealed about families create new stumbling blocks for its implementation. This session will discuss the role consumers play in the advancement of the newborn screening system, the importance of having healthcare professionals and policymakers understand the viewpoints of parents, and the need for educating the public about NBS.

• • • Workshop Descriptions

Services Track

Facilitating Parent-Primary Care Physician Partnerships in Genetics Linden Oak

Building partnerships with a primary care provider may improve health care management for patients with genetic syndromes through improved referrals, surveillance of co-occuring conditions, syndromic preventive health care, and additional syndrome-specific anticipatory guidance topics. This workshop will focus on patients with the 22q11.2 Deletion (DiGeorge Syndrome, Velo-Cardio-Facial Syndrome) as an example of a genetic syndrome, patient health care needs, and challenges to syndrome-specific primary care. The current health care guideline project of the International 22q11.2 Deletion Syndrome Foundation will also be discussed.

Services Track

The Impact of Living with a Genetic Condition in 2007 Brookside B

Being hit with a rare genetic disorder is like being hit by a stray bullet. In this session we'll discuss the physical, emotional, and financial impact of living with a genetic disorder today for both adults and children. We'll also focus on what it means to be "different" and some of the ramifications for our communities and country, as well as the relevance for our national equality and disparity policies.

Workshops: Session V

Sunday, 8:30 – 10:00 AM

Education Track

Teaching and Learning Together Linden Oak

Achieving genetic literacy is a challenge for students, advocates and healthcare professionals alike. This interactive workshop will explore mechanisms and resources for accessing and promoting the effective use of genetic information that may be utilized by advocacy groups. The workshop will include discussions of the American Society of Human Genetics Mentor Network and GenEdNet, a genetics education clearinghouse; efforts by the National Human Genome Research Institute and others in genetics education for healthcare providers and the public; and resources in genetics and family history directly available from Genetic Alliance as well as other websites that may be used by individuals or groups.

Education Track

Community Centered Family Health History Oakley

Effective community-based programs often lose their focus when translated to a national audience. Tools produced for a specific community are modified to have broad appeal, frequently missing many of the populations they were originally designed to reach. In this session, we will discuss methods for keeping the community focus in a national initiative, using the Community Centered Family Health History project as a model. The session will conclude with an open forum discussion.

Research Track

Collaboration, Education, and Test Translation (CETT) Glen Echo

Is your organization developing clinical genetic testing? Do you have an active collaboration with the clinical lab and researchers? Learn about the CETT (Collaboration, Education and Test Translation) Program. This workshop will showcase the importance and benefits of collaboration between the laboratory, the researcher, and the advocate for groups considering testing. We will also address groups that have testing who might want to use the CETT Program to build a useful collaboration.

Research Track

Case Study of Newborn Screening Information Integration Brookside B

The mission of newborn screening is best realized by a comprehensive system that assures screening, follow-up, diagnosis, and intervention for each child. Both newborn hearing screening and bloodspot screening must demonstrate accountability and effective care. The state of Alaska is enhancing its electronic information system to collect data, track patient journeys, and monitor outcomes. At the end of the presentation, participants will recognize how effective information management can be the safety net assuring better outcomes in the pre-analytic, analytic, and post-analytic phases.

Services Track

Transition to Self-Management for Individuals and Families Forest Glen

Now more than ever people with genetic illnesses are surviving and living longer lives. This means that there is an ever-growing population of children and adolescents entering and facing the challenges of adulthood. With this transition comes a multitude of issues, including switching healthcare providers, independent living, and co-occurrence later in life. In this session we will explore the common and not so common issues that arise when transitioning into adulthood with a genetic condition. The perspectives of patients, parents, and healthcare providers will be explored.

State of the Alliance Dinner

Friday, July 27th at 7:15 PM

Filmmaker Joanna Rudnick will speak about living with a high risk of developing breast and ovarian cancer and her documentary from Kartemquin Films, *In The Family*.

In the Family follows filmmaker Joanna Rudnick as she navigates the uncertain world of genetic information. With the knowledge that she has an up to 85 percent lifetime risk of breast cancer and a 60 percent lifetime risk of ovarian cancer, she is confronted with what measures to take to prevent the disease suffered by generations of women in her family.

As Joanna turns 32, she balances dreams of a family with the unnerving reality that she is risking her life by holding on to her fertility. Joanna looks to other women who carry the same mutation to help her understand the consequences of both paths: removing healthy body parts or constantly monitoring her body to try and catch cancer early. Joanna takes the audience into genetic counseling sessions, screening appointments, and intimate discussions between mothers and daughters, and husbands and wives to explore the emotional complexities of living with genetic knowledge.

In the Family is a co-production of Kartemquin Films and ITVS, the independent arm of public television. Kartemquin Films recently received the prestigious 2007 MacArthur Award for Creative and Effective Institutions. *In the Family* will be broadcast on PBS in 2008.

About Joanna

Joanna brings a personal connection to *In the Family* as a BRCA-positive young woman. Professionally, she has a background in science journalism and film production. In addition to her role as Director of Development at Kartemquin Films, she is producing *Prisoner of Her Past*—a film that traces the journey of Chicago Tribune journalist Howard Reich's attempt to uncover his mother's tragic Holocaust childhood in order to understand why she is reliving it 60 years later.

Paying Tribute to Our Heroes: Genetic Alliance 2007 Awards Banquet

Abbey Meyers, Founder, President, National Organization for Rare Disorders

Saturday, July 28th at 7:00 PM

Art of Advocacy Award

The Art of Advocacy Award pays tribute to a visionary grassroots leader who is harnessing his or her knowledge and experience to improve the quality of research, healthcare, information, and support services for a specific condition or for a coalition of grassroots organizations.

"Twenty-five years ago," says an industry representative, "this mother from Connecticut took the passion that only a loving and concerned parent can have and turned it into a vision that energized other parents of children with unmet medical needs stemming from rare diseases, that motivated physicians and other healthcare providers seeing these unusual and oft-undiagnosed disorders, and that enlightened elected public officials." In 1983, Abbey Meyers founded the National Organization for Rare Disorders (NORD), a federation of voluntary health organizations dedicated to helping people with rare "orphan" diseases, which is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and service. Leading the way through NORD, Ms. Meyers is considered to be the primary consumer advocate responsible for the passage of the landmark Orphan Drug Act of 1983.

The president of a rare disease advocacy organization states, "All of the tens of thousands of Americans with rare diseases who have benefited from the medicines developed for them under the Orphan Drug Act can be grateful that Abbey has the mind, heart, and voice to have been the ultimate advocate for them."

Genetic Alliance is proud to honor Ms. Meyers with the Art of Advocacy Award for her vision, dedication, and experience in moving the rare disease community forward. Her 25 years of tireless work has touched the lives of countless patients and families worldwide. Thank you, Ms. Meyers for everything you have done and will continue to do.

Paying Tribute to Our Heroes: Genetic Alliance 2007 Awards Banquet

Ronald M. Zuker, MD

Saturday, July 28th at 7:00 PM

Art of Listening Award

The Art of Listening Award honors a health professional who models the importance of caring, receptive professionals in the lives of individuals and families living with genetic conditions.

Ronald M. Zuker, MD, a plastic surgeon based in Toronto, has been affiliated with the Moebius Syndrome Foundation (MSF) since its inception in 1994. Known for pioneering the grascillis muscle surgery ("smile surgery") in 1997, Dr. Zuker's work has benefited countless children throughout the world living with Moebius Syndrome. His patients view him as a model physician who takes time with each patient and their family to carefully explain the procedure, answer any questions, and simply listen. A member of the MSF states, "I'm always amazed at the time he takes with each person who wants to talk to him. At our conferences, he gets down on his knees to talk to the children, plays with them and never turns away anyone who wants to talk to him. He's even missed airplane flights because he won't tell people he doesn't have the time to talk to them."

Genetic Alliance is proud to honor Dr. Zuker with the Art of Listening Award for understanding the importance of listening as an invaluable key to understanding and for his ability to ease the difficult—often frustrating—journey faced by individuals and families dealing with the uncertainties and ambiguities of a genetic condition. Thank you, Dr. Zuker.

Paying Tribute to Our Heroes: Genetic Alliance 2007 Awards Banquet

Amy Marcus, Reporter, Wall Street Journal

Saturday, July 28th at 7:00 PM

Art of Reporting

The Art of Reporting Award honors a reporter or journalist who models the special responsibility to report fairly and impartially about developments in genetics and the impact of these discoveries on people's lives.

Amy Marcus is a Pulitzer-prize winning staff writer at the Wall Street Journal. Known for articles concerning genetic issues such as rare diseases, drug discovery, and research, Ms. Marcus' articles have touched many readers. Members of the Progeria Research Foundation write, "[Ms. Marcus] does a great service to raise awareness—in a compassionate and scientifically accurate way—about the work of [genetic advocacy] organizations, why we do what we do, and our vital importance as catalysts to move science forward in order to find treatments and cures for our loved ones." A doctor at the Dana-Farber Cancer Institute states, "Most impressive [is] Amy's focus on writing a story that helped the reader understand what toll severe disease can have on everyone involved, while maintaining an honesty and depth that helps the reader understand, not just feel sorry for the [patients and their families]." A representative of Spectrum Science Communications maintains, "Through her work, she is a true advocate for rare diseases and a role model for fellow journalists."

Genetic Alliance is proud to honor Ms. Marcus with the Art of Reporting Award for modeling the special responsibility to report fairly and impartially about developments in genetics and the impact of these discoveries on people's lives. Ms. Marcus heightens public awareness and understanding of genetic conditions and the advancements in the field of genetics in a way that is sensitive to the difference between education and exploitation. Thank you, Ms. Marcus.

Paying Tribute to Our Heroes: Genetic Alliance 2007 Awards Banquet

Affymetrix

Saturday, July 28th at 7:00 PM

Art of Industry Partnership Award

The Art of Industry Award honors a for-profit biotechnology, pharmaceutical, or genetics company whose track record models the benefits of creative partnerships between consumer advocates and industry to advance understanding and treatment of genetic conditions, disorders, and diseases.

Headquartered in Santa Clara, California, Affymetrix offers a complete portfolio of tools that continue to accelerate life science research and enable scientists to develop diagnostics and tailor treatments for individual patients. The company's GeneChip® microarray technology provides researchers with a better understanding of the genes associated with adverse drug response or common, complex disorders. It is currently being used by the world's top pharmaceutical, diagnostic and biotechnology companies, as well as leading academic, government and not-for-profit research institutes.

For the past six years, Affymetrix has played a leading role in helping to prevent the misuse or abuse of genetic information and to ensure that people continue to benefit from the tremendous advances in genomic technology. Affymetrix believes that informed public discussion, meaningful application of ethical principles and thoughtful public policy must foster the constructive uses of genetic information.

Affymetrix is a co-founder and active executive committee member of the Coalition for Genetic Fairness, an influential organization established to promote genetic nondiscrimination policy. Affymetrix continues to be the lead industry supporter of the Genetic Information Nondiscrimination Act. This important federal legislation will establish a much-needed federal prohibition against genetic discrimination and will enact a national standard for ensuring the privacy of personal genetic information.

Genetic Alliance is proud to honor Affymetrix with the Art of Industry Partnership Award as the industry leader in the fight for federal protections against genetic discrimination. As the Genetic Information Nondiscrimination Act progresses through Congress, Genetic Alliance is proud to recognize the dedication of the one industry partner who has stood shoulder-to-shoulder with the consumer community for over 10 years to advocate for this sweeping national change. Thank you, Affymetrix.

Keynote Address

Genetic Diagnostics as the Base of a 21st Century Intelligent Health System

Former Speaker of the House Newt Gingrich

Sunday, July 29th at 10:30 am

Former Speaker Newt Gingrich is the founder of the Center for Health Transformation, a collaboration of leaders dedicated to the creation of a 21st Century Intelligent Health System that saves lives and saves money. He is also the founder of the Gingrich Group, a communications and consulting firm specializing in transformational change, and serves as a distinguished visiting fellow at the Hoover Institution at Stanford University in Palo Alto, California.

Widely recognized for his commitment to a better system of health for all Americans, his leadership helped save Medicare from bankruptcy, prompted FDA reform to help the seriously ill and initiated a new focus on research, prevention and wellness. His contributions have been so great that the American Diabetes Association awarded him their highest non-medical award and the March of Dimes named him their 1995 Georgia Citizen of the Year. He currently focuses on health issues in the private sector as founder of the Center for Health Transformation and is a Board member of the Juvenile Diabetes Foundation.

Newt also serves with former Senator Bob Kerrey as Co-chairman of the National Commission for Quality Long-term Care. In addition he is a member of the Advisory Board for the AHRQ (Agency for Healthcare Research and Quality) and for the National Library of Medicine. A leading advocate of increased federal funding for basic science research, in 2001 he was the recipient of the Science Coalition's first Science Pioneer award, given to him for his outstanding contributions to educating the public about science and its benefits to society.

A strong advocate of volunteerism, Gingrich has long championed the positive impact every individual can have on society. He has raised millions of dollars for charity, donating both time and money to a wide array of causes, including Habitat for Humanity, United Cerebral Palsy, the American Cancer Society, ZooAtlanta, and the Earning By Learning literacy program, which he founded.

Gingrich is the author of eleven books including Saving Lives and Saving Money, which describes the Center for Health Transformation's 21st Century Intelligent Health System.

He resides in Virginia with his wife, Callista. He has two daughters and two grandchildren.

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Nicole Exe, MS, Genetics Fellow, University of Michigan

Alyson Krokosky, Genetics and Public Health Fellow, University of Michigan

David Marshak, Genetics Fellow, Cornell University

Andria Cornell, Public Policy Fellow, George Washington University

Summer Interns 2007

Sam Hwu, Duke University

Yojiro Konno, Grinnell College

Greg Gilmore, University of California, Irvine

About Genetic Alliance

What We Do

Genetic Alliance facilitates innovation, accelerates translational research, and improves genetic services to improve human health. Standing at the crossroads of the genetics community, we are a rich nexus of advocacy and community organizations, industry leaders, and public partners. Together we increase the capacity of advocacy organizations to achieve their missions and leverage the voices of millions of individuals and families living with genetic conditions. To accomplish these goals, we catalyze networks, design and implement rigorous training courses, and create shared infrastructure.

Genetic Alliance builds a new paradigm for advocacy. With our programs, tools and technologies, advocacy organizations are revolutionizing research and treatment for genetic conditions, and supporting innovation in genetic testing and technologies.

National Consumer Center for Genetics Resources and Services

NCCGSR empowers the community through education and policy.

- Access to Credible Genetics Resources Network: Establishing standards for accurate and credible information on genetic conditions and providing tools for the development of quality materials
- Community Centered Family Health History: Engaging diverse communities in the creation of family health history resources
- Consumer-Focused Newborn Screening Projects: Generating models to maximize benefits in newborn screening

Resources

Genetic Alliance provides training in leadership, organizational development, and advocacy.

- Annual Conference: Networking and workshops
- Disease InfoSearch: Online database of advocacy organizations and resources on genetic conditions
- Institute for Advocacy: Skill-building workshops
- Resource Repository: Digital repository of shared documents
- Strategies for Success: Issue specific webinars
- Website and listserv hosting: Electronic services at a reasonable cost
- WikiAdvocacy: Tools for advocates and organizations
- WikiGenetics: Human genetics information for the layperson

"I continue to be amazed at the quality and quantity of work you are accomplishing at Genetic Alliance. Your leadership there has been a blessing to us all!"

–Audrey Gordon, Executive Director, Progeria Research Foundation

"Your programs always reflect the Alliance's strategy and vision for making change using multiple approaches to public engagement and policy development. Thank you for your excellent work." –Kay Johnson, Co-Director, Project THRIVE

Publications

Genetic Alliance develops publications for emerging and experienced stakeholders.

- G.Advocacy: Quarterly e-newsletter
- *Guide to Understanding Genetics for Patients and Professionals:* Manual of basic genetics concepts
- Monograph series: Topic specific publications, i.e. *Cancer and Advocacy*
- Public Policy Bulletin: Weekly policy e-newsletter
- Weekly Bulletin: Weekly community e-newsletter

Public Policy

Public policy must catalyze the translation of genetic information into treatments and services. We proactively engage all stakeholders through dynamic coalitions, forums, webinars, Hill events, and informational resources. Genetic Alliance directs robust policy initiatives.

Genetic Alliance educates all stakeholders. We work with our partners in industry and the healthcare professional community to lead advocates in addressing:

- Comprehensive newborn screening
- Genetic information nondiscrimination
- Open access to genetic research and information
- Personalized medicine
- Quality genetic testing
- Stem cell research

Exhibitors

The Exhibits Gallery opens at the reception on Friday, July 27th at 6:15 PM. Visit the Exhibits Gallery in the hallways adjacent to the Conference registration table and the Grand Ballroom.

Children's Alopecia Project, Inc.

Address: Post Office Box 6036, Wyomissing, PA, 19610
Tel: 610.468.1011
Email: cap4u@verizon.net Website: www.childrensalopeciaproject.org

The Children's Alopecia Project is the only 501(c)3 non-profit devoted specifically to children living with the incurable autoimmune hair loss disease Alopecia. We are raising awareness and funds to give back to the kids with our CAP kid meetings, events, and activities. We are building self-esteem and confidence so the children become stronger teens, productive adults, and the advocates of tomorrow. Visit www.childrensalopeciaproject.org to learn more about how you can help.

The Genetics and Public Policy Center

Address: 1717 Massachusetts Avenue NW, #530, Washington, DC, 20036
Telephone: 202.663.5971 Fax: 202.663.5992
Email: gppcnews@jhu.edu Website: www.dnapolicy.org

The Center helps policy leaders, decision makers, and the public better understand how the evolving field of human genetics applies to healthcare by surveying public attitudes about genetics issues, analyzing existing regulatory landscapes, monitoring transitions of genetic applications into clinical practice, positing options and likely outcomes of genetics policy, and facilitating public participation in making genetics policies.

Mercy Medical Airlift

Address: 4620 Haygood Road, Suite 1, Virginia Beach, VA, 23455
Telephone: 757.318.9174 Fax: 757.318.9107
Email: mercymed@aol.com Website: www.patienttravel.org

Mercy Medical Airlift functions as the National Patient Travel Center. Referrals and assistance with all forms of charitable medical air transportation are available through 1.800.296.1217.

NYMAC
New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services

Address: Wadsworth Center, Empire State Plaza, Albany, NY, 12201
Telephone: 518.474.7148 Fax: 518.473.1733
Email: nymac@wadsworth.org Website: www.wadsworth.org/newborn/nymac

NYMAC (New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services) was established in September 2004 as one of seven regional collaboratives in the country funded by the Genetic Services Branch in the Health Resources and Services Administration (HRSA)'s Maternal and Child Health Bureau. The charge of this group is to develop a regional approach to address the maldistribution of genetic resources in the New York/Mid-Atlantic region, which includes Delaware, the District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia. The Wadsworth Center, New York State Department of Health is the lead institution for this project.

Office of Rare Diseases, National Institutes of Health

Address: 6100 Executive Boulevard, Bethesda, MD, 20892-7518
Telephone: 301.402.4336 Fax: 301.480.9655
Email: ord@od.nih.gov Website: rarediseases.info.nih.gov

The Office of Rare Diseases was established in 1993 within the Office of the Director of the National Institutes of Health. On November 6, 2002, the President established the Office in statute (Public Law 107-280, the Rare Diseases Act of 2002). A rare disease (also called an orphan disease) is a disease or condition affecting fewer than 200,000 persons in the United States. An estimated 25 million people in the U.S. have a rare disease. he goals of ORD are to stimulate and coordinate research on rare diseases and to support research to respond to the needs of patients who have any one of the more than 7,000 rare diseases known today.

The University of Chicago Genetic Services Laboratories

Address: 5841 South Maryland Avenue, Room L035, MC0077, Chicago, IL, 60637
Tel: 1.888.824.3637 Fax: 773.834.0556
Email: ucgslabs@bsd.uchicago.edu Website: www.genes.uchicago.edu

Our molecular and cytogenetic laboratories are committed to high quality genetic diagnositics and translational research toward the development of tests for orphan diseases. Our specialty services include custom mutation analysis, comprehensive testing for Angelman syndrome, complete testing for lissencephaly, analysis of balanced translocations, and sequencing for Cornelia de Lange syndrome.

Earlier this Week

On the evening of Wednesday, July 25th, Genetic Alliance held a Gala in celebration of the Year of the Advocate at the Galleria at Lafayette Centre. Rick Guidotti unveiled the latest addition to the Positive Exposure exhibit. Genetic Alliance paid tribute to advocates Kathy Hudson and Frank Swain for their enormous dedication and drive in the fight against genetic discrimination.

Far left: from Rick Guidotti's Positive Exposure Exhibit

Middle: Kathy Hudson, Director, Genetics and Public Policy Center

Left: Frank Swain, Senior Vice President, B&D Consulting

GENETICS DAY ON THE HILL

On Thursday, July 26th, more than 100 advocates visited Congressional offices to lobby on behalf of the genetics community. The primary issue raised to Senators and their staff was genetic testing oversight. We look forward to following up with each of the visited offices and tracking our impact.

Save the Date!

Genetic Alliance
2008 Annual Conference

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July 25-27, 2008

Bethesda North Marriott Hotel and Conference Center

Metro Washington, DC

Sponsors

Office of Rare Diseases, NIH Major Conference Sponsor

Health Resources and Services Administration Major Conference Sponsor

PhRMA General Education Grant

Genzyme Institute for Advocacy

American Clinical Laboratories Association State of the Alliance Address

American Society of Human Genetics Curbside Consults

March of Dimes Fellows and General Support

Baxter Conference Tote Bags

Transgenomic Breakfast Network

BIO General Support

College of American Pathologists General Support

Expression Analysis General Support

GeneLogic General Support

Genomic Health General Support

National Society of Genetic Counselors General Support

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Genetic Alliance

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