Genetic Alliance Celebrates 25 Years of Innovation
Research for a Cure

The Alpha-1 Foundation is proud of our collaboration with the Genetic Alliance on:

- Accomplishing the passage of GINA
- Encouraging family testing for genetic conditions
- Advocacy for newborn screening
The Council for American Medical Innovation congratulates the Genetic Alliance on its 25th Anniversary!

For over two decades, the Genetic Alliance has been an invaluable leader in health advocacy. The organization's endless efforts to promote open dialogue, informational exchange and novel partnerships have brought issues affecting the genetic community to the national stage.

The Council for American Medical Innovation is proud to recognize this significant achievement. We look forward to working with Genetic Alliance to transform public health over many years to come. Again, congratulations!

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About The Council

The Council for American Medical Innovation (CAMI) is a partnership of leaders from research, medicine, academia, education, labor, business, health care and policy focused on promoting a national policy agenda on medical innovation.

www.americanmedicalinnovation.org
At Genzyme, we are united in our commitment to the treatment of rare diseases.

Genzyme is proud to celebrate 25 years of innovation in genetics and health with Genetic Alliance.
Knowledge is power.

Explore diagnostic questions.
Investigate therapeutic directions.

New insights into DNA. Greater understanding of genetic variation. Every day, scientists use Illumina sequencing technology to increase the pace of research. Now this technology is available to physicians through Illumina’s Individual Genome Sequencing (IGS)* service—the only one of its kind. Obtain valuable genetic data. Uncover the underpinnings of disease.

To learn more visit everygenome.com

*This laboratory test was developed, and its performance characteristics were determined, by the Illumina Clinical Services Laboratory (CLIA-certified, CAP-accredited). Consistent with laboratory tests, it has not been cleared or approved by the U.S. Food and Drug Administration. You will not receive from Illumina medical results, or a diagnosis, or recommendation for treatment. Please note that Illumina does not accept orders for Individual Genome Sequencing services from the following states: Florida, New York, and Rhode Island.
We congratulate Genetic Alliance on 25 years of advocacy! Whether your goal is to leverage the web to connect with your audience, create a dazzling new website or simplify your business processes, ProjectMiso can help with quality and innovation you can see and feel. With excellent one on one service, we will guide you every step of the way and help you to the top.

Our outstanding expertise in internet technology and design enable our clients to raise awareness for noble causes from health and genetics to education, women’s rights and world peace. Let ProjectMiso help you make a difference.

We congratulate Genetic Alliance on 25 years of advocacy!

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Mohr Davidow

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and the tools for scientific discovery

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We enable people with life-altering conditions to lead better lives.
Genetic Alliance 25th Anniversary Celebration

Newborn Screening Coding and Terminology Guide
Data Standards for Electronic Reporting

http://newbornscreeningcodes.nlm.nih.gov/
Promotes and facilitates the use of electronic health data standards in recording and transmitting newborn screening test results

Genetics Home Reference
Your Guide to Understanding Genetic Conditions
Provides consumer-friendly information about the effects of genetic variations on human health

Kleiner Perkins Caufield & Byers is proud to support Genetic Alliance
Amicus congratulates the Genetic Alliance on your 25th anniversary.

Thank you for your dedication to increase the awareness and understanding of genetic diseases and for serving the patient community.

Amicus is developing pharmacological chaperones, a novel, first-in-class approach to treat a broad range of diseases including lysosomal storage disorders and diseases of neurodegeneration.
Genetic Alliance Registry & BioBank

• A centralized, clinical data registry and sample biorepository that enables translational research

• Offers training tools for creating successful registries and biobanks

• Visit the GARB Toolbox for relevant discussions, videos, webinars, weekly tips, worksheets, and other resources for developing an effective registry or biobank

www.biobank.org
Genetic Alliance 25th Anniversary Celebration

Pulmonary Fibrosis
FOUNDATION

We proudly salute the Genetic Alliance for 25 wonderful years of innovation and progress in raising awareness of genetic diseases in Washington, D.C. and throughout the world.

We are inspired by your work and grateful for your support of the Pulmonary Fibrosis Research Enhancement Act (H.R. 2505, S. 1350) in the 112th Congress.

The Genomic Medicine Institute
At Geisinger Health System,
A partner in the discovery of rare disease genes,
Congratulates the Genetic Alliance on its 25th anniversary

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David H. Ledbetter, PhD, FACMG
Executive Vice President
Chief Scientific Officer

W. Andrew Faucett, MS, CGC
Director, Policy and Education
Genomic Medicine Institute
CHDI Foundation, Inc. is a privately-funded, not-for-profit, biomedical research organization that is exclusively dedicated to **rapidly discovering and developing therapies that slow the progression of Huntington’s disease** (HD). Our scientists work closely with a network of more than 600 researchers in academic and industrial laboratories around the world in the pursuit of these novel therapies, providing strategic scientific direction to ensure that our common goals remain in focus. This helps bridge the translational gap that often exists between academic and industrial research pursuits and that adds costly delays to therapy development. In its role as a collaborative enabler, CHDI seeks to bring the right partners together to identify and address critical scientific issues and move drug candidates to clinical evaluation as rapidly as possible. Our activities extend from exploratory biology to the identification and validation of therapeutic targets, and from drug discovery and development to clinical studies and trials. More information about CHDI can be found at [www.chdifoundation.org](http://www.chdifoundation.org).

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**Genetic and Rare Diseases Information Center (GARD)**

Provides timely access to experienced information specialists who can furnish current and accurate information—in both English and Spanish—about genetic and rare diseases.

A new resource for primary care providers that facilitates early diagnosis of the wide range of disorders that present with muscle weakness...

ChildMuscleWeakness.org

The National Task Force for the Early Identification of Childhood Neuromuscular Disorders
For more information: Holly@ChildMuscleWeakness.org

El Camino and Genetic Alliance: Proud partners in providing genomic-based medicine

Congratulations to Genetic Alliance on your 25th anniversary!

We are thrilled to be collaborating with you on the first ever hospital-based curriculum for practicing physicians, starting this fall.

From your friends and colleagues at:

El Camino Hospital
THE HOSPITAL OF SILICON VALLEY
GENOMIC MEDICINE INSTITUTE
Congratulations on Your 25 Years of Significant Accomplishments and Contributions to the Genetic and Rare Diseases Community!

From Your Partners and Colleagues at the NIH Office of Rare Diseases Research

23andMe is pleased to congratulate Genetic Alliance on 25 years of innovation.

The mission of the Moebius Syndrome Foundation is to provide information and support to individuals with Moebius syndrome and their families, promote greater awareness and understanding of Moebius syndrome, and to advocate for scientific research to advance the diagnosis and treatment of Moebius syndrome and its associated conditions.
Congratulations to Genetic Alliance for 25 years of excellence in advocacy, education, and action in the field of genetics.
Thank you to the generous sponsors who made our 25th Anniversary Celebration possible!

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  - A Sanofi Company
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- RainDance Technologies
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Thanks to our supporters at all levels!

Organizations

1p36 Deletion Support & Awareness
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    Faces Autism Support Group
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    Immune Deficiency Foundation
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New England Regional Genetics Group (NERGG)
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Thanks to our supporters at all levels!

Individuals

Delphine Ally
Diane Baker
Joseph Battle
Christy Baxter
Scott Berns
Tara Biagi
Greg Biggers
Natasha Bonhomme
Jeffrey Botkin
Susan Boudreaux
Rhianna Campbell
Lisa Wise & Cameron Cohen
Amelia Chappelle
Paul Daniel
Mary Davidson & Matt Seiden
Gene Early
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Kelly Edwards
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Meredith Salisbury
Ted Snelgrove
Elizabeth Stark
Pamela Sun
Sharon & Patrick Terry
Wendy Uhlmann
Chris Vocke
Gerard Vockley
Andrea Williams
Mary Ann Wilson
Emily Winn-Deen
Joan & Stanley Weiss
Joan understood that support groups sustain struggling families while energizing research that improves lives.

— Mary Davidson

**Joan O. Weiss**

Joan founded the Alliance of Genetic Support Groups – now Genetic Alliance – in 1986 with great vision and foresight. She provided a home for hundreds of disease advocacy organizations to work together to solve the challenges inherent in genetic conditions. Joan is a social worker, teacher, and a leader in the National Association of Social Workers.
Mary brought skill and passion to advancing Genetic Alliance into policy, research, and services.

— Sharon Terry

Mary Davidson

Mary became Genetic Alliance’s second executive director in 1996. She is a social worker and has advocated for the well-being of individuals all over the world. She has worked in the Peace Corps, led nonprofits in developing nations in Central America and Asia, and has counseled individuals and families.
Rodney Howell has led the transformation of modern newborn screening and saved countless lives.

— Dr. Jennifer L. Howse
President, March of Dimes

Rodney Howell

As chairman of the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children, Dr. Howell led the effort to establish national guidelines for newborn screening, making it one of the most effective public health programs in the modern era.
We salute Abbey and Peter for innovative leadership on behalf of the rare disease community.

— The National Organization for Rare Disorders Staff

**Abbey Meyers and Peter Saltonstall**

In the early 1980s, patients and families affected by rare diseases called for a healthcare system that was fair to all, but few were listening. Abbey Meyers helped change that dramatically. Now, Peter L. Saltonstall continues to provide innovative leadership toward access to safe, effective treatments for all.
Michele is an amazing innovator, finding ways to focus on the individual and the family.

— Sharon Terry

Michele Lloyd-Puryear

Michele has championed the needs of men, women, and children for her entire career, first at Genetic Services Branch of MCHB, and now with the Office of Rare Diseases Research, NIH. She has worked in family history, newborn screening, genetic services, and translational research with enormous energy and care.
A courageous leader, Sharon embodies the openness and innovation to which she invites the whole world.

— Greg Biggers

**Sharon Terry**

Through fierce determination and unfailing openness, Sharon refocuses conversations on what matters. From her pioneering work on pseudoxanthoma elasticum, a condition that affects her children, to her leadership and creation of global partnerships as part of Genetic Alliance, she sees beyond limitations to the solutions that will make our world a better place.
He is a solution hunter who arms himself with facts.

— Sharon Terry

Erik Fatemi

Erik’s deep knowledge about and commitment to the promise of biomedical research has had a profoundly beneficial influence on the federal budget process through his work in Senate appropriations. His unstinting commitment to public service and support for healthcare and medical research benefits us today and will benefit our grandchildren tomorrow.
Terri’s creativity enables her to harness the power of collaboration & innovation to drive transformation.

— Beth Meagher

**Terri Cooper**

Dr. Cooper develops innovative strategies and new discovery models for research communities to advance treatments emerging from drug and device development pipelines. Focusing on specific indications and populations, she combines the power of health IT with novel collaborative approaches integrating industry, not-for-profit and patient perspectives to inform drug development and treatment of disease.
Greg engages the citizenry, empowering them with energy, transcending the ordinary – a true luminary.

Greg Biggers

Greg’s leadership at Genomera has put personal health back in the hands of patients. At Genomera, everyday people can do everyday science that can impact the health of individuals and communities. He has transformed a vision of crowd-sourced personal health into an exciting reality poised to make disruptive change.
James has been key to developing family health resources that deliver the “right” information at the “right” time and in the “right” manner.

— Michele Lloyd-Puryear

James O’Leary

James is the CHIEF INNOVATION OFFICER for Genetic Alliance! He is quick to find solutions, create novel pathways and see where we can increase energy and save resources. He has created a customizable family health history guide and is now throwing his energy into connecting consumers to smart services.
Amy inspires others to work together to achieve large, complex goals.

— Diane Baker

**Amy Comstock Rick**

Leading a multidisciplinary team through the public process of decision-making around our government’s embryonic stem cell research policy, Amy oversaw a journey of profound implications that was open, thoughtful, intense, and ultimately successful. She is focused and unflappable in the face of the turbulence that accompanies meaningful change.
Clarissa rises to every challenge, leading all of us with aplomb and grace.

— Christine Vocke

Clarissa Sellers

As PXE International Research Coordinator, Clarissa leads our epidemiological study, all while working on her master’s degree in public health at George Washington University. Collaborative, skilled and efficient, she leads us all through a maze of challenges to a quality result. We are delighted by her passion and inventiveness!
Dr. Clifford Reid played a pivotal role in making large-scale human genome sequencing a reality.

Clifford Reid

Complete Genomics CEO Dr. Clifford Reid introduced the first high-quality, affordable whole human genome sequencing and bioinformatics service; freeing customers from buying expensive instruments, computing or data storage equipment or investing in specialized personnel. Customers focus on biological discovery instead. Customers send DNA samples and receive high quality, research-ready results.
Colleen B. Zak is a passionate patient advocate who has altered how this disease is viewed.

— The ARPKD/CHF Alliance

Colleen Zak

Inspired by the love of a child affected by Autosomal Recessive Polycystic Kidney Disease & Congenital Hepatic Fibrosis, Colleen B. Zak, RN, founded the ARPKD/CHF Alliance. From anguish and despair, to faith in things hoped for and certain of things not seen (Hebrews 11.1), improving the lives of those affected.
Fabry disease is a family disease. Through FSIG, Jack has created a global Fabry family.

— John M. King 
Senior Product Director, Fabrazyme 
Genzyme Corporation

Jack Johnson

Jack is the driving force behind Fabry Support and Information Group (FSIG). His own family's long history with Fabry inspired him to create an organization to unite people affected by Fabry to learn from and support one another. FSIG started as a "kitchen table" support group and evolved into an indispensable resource for hundred of families.
The Sies demonstrated remarkable vision in establishing the Linda Crnic Institute for Down Syndrome.

— Ed McCabe, MD, PhD
Executive Director
Crnic Institute

Anna and John Sie

Anna and John J. Sie are world-class innovators and philanthropists. They recognized the need for a comprehensive institute to move knowledge rapidly from laboratory to clinic to improve the lives of all people with Down syndrome. Today the institute is set to eradicate the condition’s medical and cognitive ill effects.
Francis manifests the essence of the innovator his every action: presence, creativity, and vision.

— Sharon Terry

Francis Collins

Francis is a physician-geneticist noted discoveries of disease genes and his leadership of the international Human Genome Project. He is the director of the National Institutes of Health. He understands the anatomy of problems and invents effective solutions. He is ever vigilant in keeping human health foremost in his work.
Dr. Dimmock is a wonderful doctor. He took time with us to explain what was wrong with our child and how we could get the answers to his problems.

— patient

David Dimmock

Dr. Dimmock is a respected clinical biochemical geneticist, known for the first use of exomic sequencing to change management, in a child with immune dysfunction. He has spearheaded clinical implementation of whole genome sequencing in a CLIA/CAP regulated environment with ethical oversight ending diagnostic odysseys for children with genetic disorders.
We all benefit from patient advocates as tireless and dedicated as Dan Greenberg.

— Karen Litwack

Dan Greenberg

Dan Greenberg and his wife led the charge to discover the genetic basis of Canavan disease, and have fought to clarify the legalities of gene patenting. Dan helps the Chicago Center for Jewish Genetic Disorders develop and promote comprehensive Jewish genetic disease resources for families, clergy, healthcare professionals and the Jewish community.
Darren’s passion for digital biology continues to help advance medical genetics and accelerate disease research.

— Roopom Banerjee
President and CEO
RainDance Technologies

Darren Link

Darren Link has dedicated his career to helping scientists answer important biological questions in human health and disease research. As co-founder at RainDance Technologies, Darren has developed a microdroplet-based platform that enables geneticists to sequence select regions of the genome and better understand the underlying genes associated with medical conditions.
Dr. Rade Drmanac is a truly dedicated pioneer of affordable massively parallel human genome sequencing for 25 years.

— Dr. Clifford Reid

Rade Drmanac

Complete Genomics CSO Dr. Rade Drmanac developed the novel DNA biochemistry and nanoarray technology used in its highly accurate, large-scale, and affordable human genome sequencing service. It’s now sequencing thousands of genomes for National Cancer Institute, Inova Translational Medicine Institute, Erasmus Medical Center and others to enable genomic medicine.
Joan is highly esteemed for her innovative, enabling work in the diagnosis of rare genetic diseases.

— David Meeker, MD
COO, Genzyme Corporation

Joan Keutzer

For over 15 years, Joan Keutzer – VP, Scientific Affairs, Genzyme Corporation – has devoted her time and boundless energy to developing the scientific tools required to support the diagnosis and treatment of patients with lysosomal storage disorders. She has demonstrated the feasibility and utility of newborn screening and developed technology that makes diagnosis faster and easier.
If you don’t stand for something, you’ll fall for anything.
— Martin Luther King

Chikezie Maduka
Chikezie Maduka is a member of the National Community Committee of the Center for Disease Control Prevention Research Centers, representing the University of Maryland Prevention Research Center. He is also a member of the Special Interest Group on Genomics of the National Community Committee. He is Co-Chair of the Seat Pleasant-University of Maryland Health Partnership and an Innovator of the Seat Pleasant Community Genomics Group.
Cheryl’s dedication to her vision has grown a global support system for people with JSRDs.

**Cheryl Duquette**

The Joubert Syndrome & Related Disorders Foundation has grown from 8 families in the USA to over 750 worldwide. Much of this is attributed to the tireless efforts of Cheryl Duquette. Her presence at countless medical meetings sparked passion in researchers across the continent. Without her vision, families would not share the supportive benefits the Foundation offers today.
Dr. Gunay-Aygun is an effective, innovative leader for ARPKD/CHF research.

— The ARPKD/CHF Alliance

Meral Gunay-Aygun

Meral Gunay-Aygun is the primary investigator for “Clinical Investigations into Autosomal Recessive Polycystic Kidney Disease and Congenital Hepatic Fibrosis,” at the National Institutes of Health, a study generated by the ARPKD/CHF Alliance. Her thoughtfulness, kind and gentle manner, dedication, drive and hard work give hope to thousands of ARPKD/CHF families.
Carol Terry is truly the matriarch of the Wilson Disease Association. She is my inspiration.

— Mary Graper
WDA President

Carol Terry

After a horrifying journey that led Carol to her diagnosis of Wilson disease, she was instrumental in founding the WDA 28 years ago. Carol is still an active board member today, and her dedication is unparalleled and contagious. The Wilson disease community is fortunate to have a tireless advocate like Carol.
Anu is visionary. She constantly seeks opportunities and innovative solutions for advocacy-driven research initiatives.

— Liz Horn, Genetic Alliance Registry & BioBank

**Anu Acharya**

Anu Acharya is the Founder & CEO of Ocimum Biosolutions, a global genomics outsourcing partner for discovery, development and diagnostics. Since founding the company in 2000, she has piloted it to great heights through strategic international acquisitions, capital raises for equity investments and launch of innovative products, solutions and services.
Your imagination is your preview of life’s coming attractions.

— Albert Einstein

Andrew Evans

With the availability of the personal genome, Andrew grasped the opportunity to promote radical self-awareness by making genomic data more intimately accessible. His invention of the SNPTips™ web browser extension was straightforward, useful, easily distributable, and usable!
No mountain is too high for him to climb, no obstacle too difficult to overcome.
— Sarah E. (Sally) Everett, Esq.

John Walsh
With two other patients who also had COPD due to Alpha-1 Antitrypsin Deficiency, John Walsh created the Alpha-1 Foundation to fund research for a cure, and AlphaNet, a patient service organization which hires “Alphas serving Alphas,” a unique business model that has been widely studied by other not-for-profit organizations.
Gene’s leadership enables complex systems to stay focused on what matters most.

— Kemp Battle

**Gene Early**

Recognized as a skilled leadership and organizational development advisor, Gene engages global thought leaders to deepen their understanding of themselves and the systems they serve. His faith in (and commitment to) the power of individuals to transform themselves encourages systems to innovate to effect lasting and sustainable change.
Marianne Genetti brings attention to problems with diagnosis, the need for a medical specialty in diagnosis, and a means to support those in need of diagnosis. Through In Need Of Diagnosis, Inc. (INOD), Marianne Genetti has brought to light those once-invisible problems in the field of diagnosis.

— Katherine Massey

**Marianne Genetti**

Those who suffer with illnesses that have eluded diagnosis are not just underserved, they are invisible. Through In Need Of Diagnosis, Inc. (INOD), Marianne Genetti brings attention to problems with diagnosis, the need for a medical specialty in diagnosis, and a means to support those in need of diagnosis.
Kathleen Bogart’s doctoral research on the lack of facial expression has been featured in *The New York Times* and in documentaries. Her efforts to educate the public that people with Moebius syndrome who have no facial expression can read the facial expression of others has helped dispel incorrect myths.

Kathleen’s research has dispelled long-held myths about people with Moebius syndrome.

— Vicki McCarrell, President, Moebius Syndrome Foundation
When I think of an ordinary person doing extraordinary things, I think of Mike Cohn.

— Patricia Wood, President, NBIA Disorders Association

**Mike Cohn**

Even though NBIA makes it hard for Mike to walk and talk, he eagerly volunteers. He has organized some of our most creative fundraisers: a CD of original songs, an auction of guitars signed by musicians and a calendar featuring our families. We don’t know what we’d do without him.
His astounding intellect is surpassed only by the reach of his mighty and loving heart.

— Kemp Battle

**Patrick Terry**

Pat is a serial innovator. Co-founder of PXE International, Genomic Health, the Personalized Medicine Coalition and more, he seeks to disrupt the status quo to create new pathways for solutions. He has birthed numerous foundations and companies, all dedicated to reducing suffering in this world.
These are our PKU Heroes. They made it possible for people with PKU to live up to their greatest potential and pursue their dreams.

— National PKU Alliance

Richard Koch & Charles Scrver

Dr. Richard Koch and Dr. Charles Scrver are pioneers who have graced the PKU community with extraordinary and legendary service for decades. Both have dedicated their lives to treating and helping others understand PKU. Their work has led to PKU going from a severe developmental disability to a treatable condition.
Gemma’s work impacted people with osteogenesis imperfecta all across the United States and even internationally. Her work began in earnest when she wrote two articles published in Redbook Magazine in 1968 and 1970 that were instrumental in starting the OI Foundation. She provided critically needed support for families when little information was available on OI families.

Because of Gemma we have a Foundation that provides help and hope to people living with OI.

**Gemma Geisman**

Gemma’s work impacted people with osteogenesis imperfecta all across the United States and even internationally. Her work began in earnest when she wrote two articles published in Redbook Magazine in 1968 and 1970 that were instrumental in starting the OI Foundation. She provided critically needed support for families when little information was available on OI families.
Rose works as a connector. She inspires just by being.

**Rose James**

Rose James, a member of the Lummi tribe and University of Washington Bioethics faculty, began her career in cancer pathology. She is the innovator behind the Launching Native Health Leaders program that creates opportunities and connections for tribal college students. She has literally redefined what it means to contribute and succeed.
Dr. Váradi is a key figure in Hungary to translate basic science to application in genetics.

— Anonymous Hungarian scientists

András Váradi

Dr. Váradi, a protein biochemist from Budapest (Hungary), initiated a project to study the function of ABCC6 protein in 2001; mutations in the ABCC6 gene are associated with pseudoxanthoma elasticum. His intention is to develop allele-specific correction therapy for different ABCC6 mutants.
Wendy & Rachel Kaplan

SustainAbility Farm is Rachel's dream brought to life through the work of both Wendy and Rachel. From Rachel’s facilitated/typed request to spend her life on a farm, to planting seeds, borrowing a greenhouse, persuading Planting Fields Arboretum to support Rachel's dream, and to invite other people with developmental disabilities and their direct support workers to join in - all within a 10 month period of time. They sell the organic produce through a CSA. Each person chooses how they own a piece of the work: tilling the field, planting, weeding, measuring the rows, unspooling string (lots of unspooling), picking, delivering bags of vegetables and selling at the farm stand. Rachel leads a self-determined life, and she, her Mom, and the other members of the NY Self-Determination Coalition encourage others to direct their lives in a meaningful way.
The drive, enthusiasm, and energy of the Genetic Alliance staff are inspiring.

— Andy Golden

Genetic Alliance Staff

There is never a dull moment at Genetic Alliance! Each member of the staff brings diverse expertise to the organization – from biology to finance to film – which contributes to a collaborative, productive, and fun work environment.
Never doubt that a small group of thoughtful, committed citizens can change the world.
— Margaret Mead

Caroline Lieber

Under the leadership of Caroline Lieber, three graduate students in the Joan H. Marks graduate program in Human Genetics at Sarah Lawrence College founded GenetAssist, an organization created to provide global access to genetic counseling. Caroline’s professional mission is to introduce more students to genetic counseling through various educational outreach programs.
I’ve been honored to work with Rhonda in our efforts to bring treatment to people with Gaucher.

— Richard Moscicki, MD  
Chief Medical Officer  
Genzyme

Rhonda Buyers

For over 17 years, Rhonda has been an integral part of the National Gaucher Foundation. Through her leadership, it became a major advocacy organization. Her vision and desire to help others paved the way for support, financial and educational programs to assist thousands of people with Gaucher disease and their families worldwide.
Len’s vision has made possible innovations that will positively impact healthcare for generations to come.

— D. Craig Brater, M.D.
Dean, Indiana University School of Medicine

Leonard Betley

Leonard J. Betley has an unprecedented ability to stimulate innovation within Indiana’s life science community. His leadership at the Richard M. Fairbanks Foundation resulted in a $10 million gift that created the Fairbanks Institute for Healthy Communities, an organization enabling research that can help speed the development of personalized medicine.
A lady with a will of iron, Helen-Ann will never take “no” for an answer.

— Creighton Phelps, Ph.D.
National Institute on Aging

Helen-Ann Comstock
Helen-Ann Comstock founded AFTD in 2002, when there was little recognition of this “other” form of dementia. Today, AFTD is the nexus for both care and the cure in the FTD community, funding research, coordinating a growing network of caregiver support groups, educating clinicians and providing respite for family members.
Her work has been described as an “impressive story of the revolution that genomic analysis is bringing to the clinic.”

— Francis Collins
Science, Vol. 331 no. 6017

Elizabeth Worthey
Dr. Worthey was first author on a landmark report where genome sequencing and analysis of a child suffering from a rare, devastating bowel disease identified a novel mutation leading to diagnosis and subsequent successful treatment. The innovative tools developed are now being used to end diagnostic odysseys in other children.
Participating in HFA’s Voices Campaign was a defining moment in my life.

— Community Member from Tennessee

Kimberly Haugstad

Kimberly Haugstad, Executive Director, Hemophilia Federation of America created the Voices Campaign, a united community effort to bring members together in peer support and education. Launched in 2009, the advocacy initiative is aimed at raising public and legislator awareness of the bleeding disorders community through video and written stories.
Your leadership putting such an event together has changed our childrens’ lives forever.

— Blanca Edna Garcia
Fabry mother and patient

Jerry Walter

Jerry founded the National Fabry Disease Foundation (NFDF) to increase assistance and support to people with Fabry disease. His advocacy work is truly innovative—one of the best examples of his innovation is the Charles Kleinschmidt Fabry Family Weekend Camp at Victory Junction, a weekend where Fabry kids and their families learn from each other and have a lot of fun!
Dr. Vanier is an exceptional leader, and is truly passionate about medicine and the transformative power of genomics.

**Vance Vanier**

Navigenics offers a more complete picture of each person’s health by combining lifestyle, health and family history with personal genetic information. Navigenics allows individual access to personalized genetic information and tools through a secure, online account, as well as support from a board-certified Navigenics genetic counselor – helping doctor and patient make the most of these genetic insights.
Sue’s knowledge, sense of humor and ability to work with people is famous. She will always be an important part of TSA.

— Judit Ungar
TSA President

Sue Levi-Pearl

During her 30 year tenure, Sue Levi-Pearl left an indelible mark on the national Tourette Syndrome Association. Her focus on research helped TSA set a high standard for everything science-related. Sue retired in 2009, but her contributions, enormous efforts, energy and wisdom that she directed towards TSA and its families will remain eternally.
Jeanne has lit the fire for the next generation through her work with science teachers.

Jeanne Chowning

Jeanne Chowning, Director of Education at the Northwest Association for Biomedical Research, works with science teachers to find innovative ways to introduce topics such as genetics, research ethics, stem cells, bioinformatics, and biobanking. Her programs and resources inspire kids to consider careers in biomedicine and research in ways they never imagined possible.
Jared’s heart and creativity for Kammy’s Kause inspires me.
— Jackie Leich

Jared Hiner

Eight years ago Jared Hiner had an idea to start Kammy’s Kause as a way to provide financial support to the 4p- Support Group while honoring his daughter, Kamdyn. His tireless dedication has lead to a well respected annual event. He truly makes a difference and continues to raise awareness about 4p- and Wolf-Hirschhorn Syndrome.
Kemp energizes innovation by modeling inward exploration, outward adventure, and upward inspiration.

— Gene Early

Kemp Battle
Kemp brings entrepreneurial vision, organizational savvy, and operational diligence to his clients, whether they be in the Fortune 50, start-ups, or non-profits transforming major sectors of society. He is a master story-teller, active catalyst in the arts and culture, and discriminating architect of social spaces that enliven the human spirit.
Children with SMA are bright individualities and have the same right, as well as others, to live without genetic conditions which the science can correct today.

Vitaliy Matyushenko

Vitaliy Matyushenko, President of Foundation Children with Spinal Muscular Atrophy, who is fighting for changing an understanding of Rare Diseases conception in society and Governance around Ukraine. Furthermore, he is focusing on disseminating information on best practice care according to the international guidelines among doctors and gives his time to advocacy for individuals and rare diseases groups.
To the staff of the Office of Rare Diseases Research, NIH, Steve Groft is “Mr. Rare Diseases”.

Stephen Groft
Since 1982, Stephen Groft has been an innovator for rare diseases. For example, he collaborated to bring about a genetic and rare diseases information center used by patients and their loved ones, a clinical research network where researchers and patient advocacy groups work together, and the NIH undiagnosed diseases program.
Genetic Alliance 25th Anniversary Celebration

Inspiring, encouraging, compassionate, and optimistic – these words only begin to describe Sue Lomas.

— Dr. Katy Phelan

Susan Lomas

Susan Lomas has dedicated herself to growing the Phelan-McDermid Syndrome Foundation from its inception, in 2000. First as volunteer and now as Interim Executive Director, Susan strives to provide family support, raise awareness and facilitate opportunities for translational science by building an alliance with other disease groups.
Mickey: a dedicated pioneer in improving health through enhanced diagnosis and prevention of devastating diseases.

**Mickey Urdea**

Tethys offers a solution to prevent type 2 diabetes. The PreDx® Diabetes Risk Score (DRS), a multi-marker blood test, provides an accurate, personalized risk score for diabetes diagnosis within five years. Utilization of PreDx DRS will help reverse the growing diabetes epidemic, lower healthcare costs and improve intervention outcomes.
Jan’s leadership was integral to creating physician awareness of Stickler Syndrome through the “Get The Word Out” campaign.

— Pat Houchin
Stickler Involved People

Jan Helfer

Jan created a campaign to introduce specialty physicians to “this zebra.” Strategies included development of brochures and slide presentations, mailings to nearly 8000 professionals, presentations to medical schools, exhibiting at physician and genetic counselor conferences, and publishing articles and PSAs in medical journals. The Stickler word definitely got out!
Joyce Hooker

Joyce Hooker, long-time Mountain States Genetics Regional Collaborative Center Project Manager, has remained the glue keeping this region together. Her work has created a foundation upon which we will continue to succeed for many years. Joyce has a unique ability to engage, inspire, and connect with people, and through this she has certainly changed lives.
Thanks to Kelly, when our doctors hear hoofbeats they now know it’s a WAGR zebra coming.

— Rhonda Sena
WAGR parent

Kelly Trout

Kelly Trout, International WAGR Syndrome Association co-founder, medical consultant and board member, brought our mission to the medical community by collecting information about WAGR/11p Deletion Syndrome individuals and collaborating and sharing the information with medical professionals. Thanks in part to Kelly’s work, today WAGR families are participating in an NIH clinical research study.
Without hesitation, Lisa generates solutions that work for everyone and soon become the new standards.

— Jim Pollack
NEST DC

Lisa Wise

Lisa is a Renaissance woman. She has left her mark on many nonprofits and companies. She continues to innovate: in healthcare, in housing, in design, in service to others. The gift of her wit and commitment, and her office dogs Lucy (RIP), Shaggs and Jackson keep Genetic Alliance going!
Medical genetics must be a household word for all obstetrician-gynecologists, both for prenatal diagnosis and for female reproductive disorders.

**Joe Leigh Simpson**

Joe Leigh Simpson, M.D., currently at Florida International University College of Medicine, has been a pioneer in the development of reproductive genetics as a special field of study within clinical genetics. Having mentored many current leaders in this area, he’s a dedicated educator and committed to women’s health and maternal-fetal medicine. His team was the first to achieve definitive non-invasive detection of fetal trisomy18 and trisomy 21 by analysis of maternal blood.
“PASSIONARY” = someone inspired passionately to improve the world, volunteering beyond ordinary responsibilities; change-agent; inspirational difference-maker

Simone Sommer

Dr. Sommer is a passionary, advocate, and entrepreneur dedicated to improving the lives of people with rare cancers. While seeking a treatment for her only child’s rare cancer, she developed a method to improve the diagnosis, treatment and research strategies for rare cancers and founded a nonprofit to pilot this vision.
Ana and Isa demonstrate the power of storytelling for global change around critical health issues.

**Anabel Stenzel & Isabel Stenzel Byrnes**

Twin sisters Anabel Stenzel and Isabel Stenzel Byrnes, centerpiece characters of the documentary “The Power Of Two,” survived a lifelong battle with cystic fibrosis through miraculous double lung transplants and have emerged as authors, athletes and advocates for organ donation and those with CF in the U.S. and Japan.
Andrea Williams is transforming the world of sickle cell disease care and advocacy with her collaborative approach, personal passion and global perspective. The Founder and Executive Director of Children’s Sickle Cell Foundation, Inc., she enjoys developing novel approaches to addressing the needs of children living with sickle cell disease.

Andrea redefined the meaning of “consumer engagement.” Her passion, creativity, and dedication have and will open doors for families throughout the health community.

— Natasha Bonhomme, Genetic Alliance

Andrea Williams
Andrea M. Williams is transforming the world of sickle cell disease care and advocacy with her collaborative approach, personal passion and global perspective. The Founder and Executive Director of Children’s Sickle Cell Foundation, Inc., she enjoys developing novel approaches to addressing the needs of children living with sickle cell disease.
The Advocates Partnership Program is a small effort you make in the world of advocacy, but what it means to us is HUGE!

— Megan O’Boyle, Advocate 2010

Advocates Partnership Program

In 2005, Genetic Alliance and the American Society of Human Genetics hosted the first Advocates Partnership Program at the ASHG conference. Since then, the program has expanded to the annual meetings of the nation’s leading health professional societies, offering advocates an unparalleled opportunity to gain insight into a variety of professions while networking with advocates and professionals from around the globe.
Lucy lights up a room, and through dance returns forgotten grace to people with Parkinson’s.

— Phyllis Richman

Lucy Bowen McCauley

Lucy Bowen McCauley, Artistic Director of Bowen McCauley Dance, offers free weekly dance instruction and extraordinary performance opportunities for individuals with Parkinson’s Disease - Dance for PD. Combining dance technique, Company repertoire and an understanding of the Disease, she shows students that they can still experience the joy of movement.
Colleen Hammond

Many see needs in this world and hope that someone takes action; some see the need and take the action. This is what Colleen Hammond did for the cystinosis community. As she watched her young son struggle for life, she knew that without a strong advocacy group his life would not reach its full potential. So in a converted pantry she founded the Cystinosis Research Network (CRN). CRN is a leader in family support, medical advocacy, and research funding. What began as a dream for one woman became an organization that represents all those afflicted with cystinosis with honor and grace. All of the cystinosis community is proud and grateful for the vision that Colleen Hammond had and continues to have.
Always on the forefront of HAE research, Dr. Zuraw’s contribution to this rare disease community is inestimable.

— Janet Long, Executive Vice President, US HAEA

Bruce Zuraw

Bruce L. Zuraw chairs the US Hereditary Angioedema Association Medical Advisory Board. He was instrumental in the implementation of its Scientific Registry, which collects clinical samples and patient-provided data. Dr. Zuraw is a pioneer of HAE research, a global HAE educator, and a dedicated advocate for HAE patients.
When we are afraid, she steadies us. When we are lost, she leads us home.

— Kemp Battle

**Diane Baker**

When a system is in need of truth and honesty, couched in love and compassion, Diane delivers. In her work as the program director of the genetic counseling program at the University of Michigan, and as longtime Council member of Genetic Alliance, she has a powerful influence on the world.
Our students live transdisciplinarity – they are our future! — Wylie Burke

Center for Genomics and Healthcare Equity Trainees

The University of Washington Center for Genomics and Healthcare Equality trainees define the future of the field by pursuing complex issues – pharmacogenetics, epigenetics, return of results, community engagement, data sharing, health policy - in a multidisciplinary environment. All are committed to working through the lenses of social justice, partnership, and sound science.
Believe in our kids.

Dup15q Alliance recognizes the individuals affected by Chromosome 15q Duplication Syndrome as innovators. It is the affected individuals who have inspired the creation of the Dup15q Alliance International Registry, which will help drive research and targeted treatments for Chromosome 15q Duplication Syndrome and related disorders.
Elizabeth Hunt

Liz’s devotion to the American Indian community in Santa Clara County is apparent in every way. She works tirelessly to eliminate health disparities, which has resulted in the creation of award-winning programs influencing national healthcare policy. Her amazing attitude has lifted the spirit of the community.

Never, never, never, never give up!!
My optimism for life carries through my work.

Frank Lehmann-Horn

Through an innovative partnership with Dr. Lehmann-Horn in Germany the Periodic Paralysis Association has been able to offer extensive genetic testing to patients affected with Periodic Paralysis. His generous nature and caring attitude have enabled hundreds of families to be correctly diagnosed and treated thereby enhancing their quality of life.
The PacBio technology represents a great leap forward. It will enable a new level of information and understanding.

— Roger Kornberg
Nobel Laureate
Stanford University

Stephen Turner
Pacific Biosciences pioneered PacBio RS, the world’s first single molecule, real-time DNA sequencer. By harnessing nature’s own highly evolved sequencing engine (the DNA polymerase) in combination with advances in nanophotonics, chemistry, enzymology, optics and bioinformatics, the RS system determines an organism’s precise genetic code in real-time, enabling more complete information to be extracted from DNA in less time and at lower cost than other methods available.
With every image, you feel his hands on your temples steadying your gaze, tempting you to see beyond difference... in others and in yourself. — Joanna Rudnick

**Rick Guidotti**

Rick is founder of Positive Exposure, a nonprofit organization that challenges stigma associated with difference by pioneering a new vision of the beauty and richness of genetic diversity. Rick’s photo essay “Redefining Beauty” in Life Magazine won the Genetic Alliance’s “Art of Reporting” award. Rick has empowered this community to new heights!
Dr. Raphael Schiffmann is a leader in his field. His research on neurogenetic disorders has lead to many pivotal changes in therapy.

Raphael Schiffmann

He investigated the natural history, pathogenesis and treatment of Fabry disease, an inherited disorder of lipid catabolism. Dr. Schiffmann’s research concluded with the approval of enzyme replacement therapy for this disorder by association with the European Union and many additional countries. He received the Merit Award from the National Institute for his great work.
Stephen is a catalyzing force, actively reshaping how we do science today.

Stephen Friend

Stephen Friend, Co-Founder and Director of Sage Bionetworks, is both a teacher and a learner in the best sense. Ever curious, ever imaginative, ever hopeful that we can transform our research practices and forge a new productive path. He has inspired remarkable partners to join him on the journey.
Shantanu brings courageous leadership and a spirit of possibility to a world hungry for hope.

**Shatanu Gaur**

As an eighth grade student, Shantanu Gaur won the Genetic Alliance essay contest—beginning a career of energetic leadership and innovative thinking. Today, as the youngest member of the Genetic Alliance Council and co-founder of a medical technology company, he helps heal people by combining innovation in culture and medicine.
Paul’s deep knowledge, clear communication, and focus on what works are gifts to the legislative process.

— Sharon Terry

**Paul Williams**

The creation of a safe and trustworthy oversight process for genetic testing requires helping diverse stakeholders rise above their vested interests and embrace the broader goal of long-term public benefit. Paul’s clear-eyed survey of the landscape, the players, the issues and the consequences are guiding this journey with integrity and realism.
Penny’s dedication ensures that community priorities reach the national stage and national successes reach communities.

— James O’Leary
Genetic Alliance

Penny Kyler

As both an occupational therapist and an ethicist, Penny has always seen the forest and the trees. Her unfailing dedication to serving the needs of individuals and families has provided momentum for the Health Resources and Services Administration and its grantees to drive national programs to the local level.
She always sees, and inspires others to see, the essence of what is possible.

— Kemp Battle

Molly Corbett Broad

Molly is one of American Higher Education’s greatest innovators and visionaries. Whether managing complex university systems in Arizona and California, serving as President of The University of North Carolina or reinventing the American Council on Education, Molly inspires institutions and their leaders to bring forth their deepest purpose.
Anyone who would go out and do something about discrimination is a hero.

— Gordon Cummings
Past President, NF Midwest

Dave Evans

Dave Evans was thrown out of a local water park because customers thought that his tumor-like growths were contagious. Dave sued those who humiliated him on principle. He stood up to discrimination and ignorance for everyone with genetic differences. As he so eloquently put it “I deserve to be here.” We ALL deserve to be here.
Bill gave CDO the technological administrative capability most organizations dream of!

Bill Ort

After Chromosome Disorder Outreach’s website was essentially destroyed by hackers a few years ago, Bill Ort quietly rebuilt our site from the bottom up. The new site includes a mathematical algorithm which allows CDO administrators to catalog and scan our database records in only seconds. Bill Ort is an unsung hero to CDO families!
The beauty of collaboration between older and younger generations is that we combine strength with wisdom.
— Brett Harris

Penelope Moore

Dr. Penelope J. Moore, associate professor, Iona College, is recognized for promoting awareness about the importance of family health history. She has forged valuable partnerships with national Geriatric Education Centers, local government, private, and non-profit organizations, to expand a focus on intergenerational communication about health history.
Melodie is a catalyst for improving access for psoriasis patients. She is an incredible practitioner.

— Jennifer Cather

**Melodie Young**

Melodie Young, MSN, RN, A/GNP-C, is a dedicated nurse practitioner, researcher, educator, mother, wife, colleague, friend, and long time psoriasis patient advocate. She is a leader in her field, developing numerous educational programs for medical professionals. She works tirelessly to improve the lives of her patients.
Marie has been my “running buddy” to change the landscape of newborn screening and genetic services.

— Michele Lloyd-Puryear

Marie Mann

Marie is truly a leader in maternal and child health and is an innovator. In the midst of the technologic changes within the newborn screening laboratory, she captured that momentum to bring changes to the newborn screening system. This required the ability to partner, listen, and respond with sensitivity and the ability to bring politically diverse groups and individuals to consensus.
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Diet is key for celiac disease treatment; however, a support group is just as beneficial.

— Pat Murphy Garst

Pat Murphy Garst
Innovator Pat Murphy Garst of Des Moines, Iowa, organized the Midwest Celiac Sprue Association in 1977. Searching for a network to share knowledge of celiac disease, she promoted education, research and awareness. The organization evolved nationally into the Celiac Sprue Association which networks as the largest celiac disease organization.
Over the years I have seen Natasha bloom to embody patience and collaboration, both essential for successful partnerships, but also essential for the diffusion of any innovation.

— Michele Lloyd-Puryear

**Natasha Bonhomme**

Natasha has been called the “immune system” for Genetic Alliance. She is able to discern and alleviate stresses in the system. Her work has spanned culture, newborn screening, and strategic development. Able to hold two sides of any situation in her view, she brings balance to innovation.
Her focus on trusted practices brings vision and flexibility to problem solving strategies.

— Diane Baker

Kelly Edwards

Although the destination of our genomics journey is still being defined, the light provided by bioethics will guide the research, governance, and integration into daily life of this paradigm-changing field. Kelly’s commitment to partner genomic progress with humanity, justice, and community involvement is transforming this environment.
Jennifer is well respected within the dermatology community. I am honored to be her colleague and friend.

— Melodie Young

Jennifer Cather

Jennifer Cather, MD, is a woman of absolute integrity, who genuinely cares about her staff, her patients and her colleagues. Constantly seeking out the most challenging and complex dermatology patients, she is the perfect combination of generosity, brilliance, humility and humor.
Instead of only witnessing pain and suffering, Michael organizes compatriots and initiates positive, sustainable change.

— Simone S. Sommer, MD, MPH

Michael Marcus

Michael S. Marcus, MSW, Program Director at Older Adult Services, is a tireless advocate dedicated to reducing poverty, hunger and homelessness. His involvement in innovative programs to improve the lives of aging populations as program director at four foundations, and founder of Health-Care-For-All and the National Caregiver Initiative, has resulted in thousands of seniors receiving billions of dollars of services.
Bob has worked tirelessly for over 10 years to bring effective treatments to Pompe patients.

— John McPherson
Senior VP, Biologics Department
Genzyme Corporation

Robert Mattaliano

The scientific leadership of Bob Mattaliano – Group VP, Biologics Development, Genzyme Corporation – has been instrumental in the development of treatments for Pompe disease. His innovative approach to problem solving has surmounted technical hurdles and fostered key collaborations between Genzyme and leading academic researchers working on Pompe.
Under Steve’s respected leadership, the National MPS Society has grown into a respected advocacy, awareness and funding organization.

— Barbara A. Wedehase
Executive Director,
National MPS Society

Steve Holland

Steve fights tirelessly to ensure that one day there will be effective treatments for all MPS diseases. With his leadership the National MPS Society supports research, raises awareness and provides critical support for families affected by MPS diseases.
Isabelle and Kyle Mosca inspire other siblings of special needs children to see the awesomeness in awareness.

Isabelle & Kyle Mosca

Isabelle wrote and Kyle illustrated *Adventure to Autism Planet*, a children’s book which is now read in schools all over the country on Blow Bubbles 4 Autism Day each April to teach children tolerance, patience and autism awareness. Thanks to the twins, people now spread healing bubbles of hope and support to those facing autism each year.