

# GENETIC ALLIANCE ANNUAL REPORT 2013





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## Background

*Genetic Alliance engages individuals, families and communities to transform health.*

Genetic Alliance, Inc. (Genetic Alliance) was incorporated as a non-stock, nonprofit organization on October 31, 1986 within the laws of the State of Maryland.

We bring together diverse stakeholders to create novel partnerships in advocacy; we integrate individual, family, and community perspectives to improve health systems; we revolutionize access to information to enable translation of research into services and individualized decision-making.

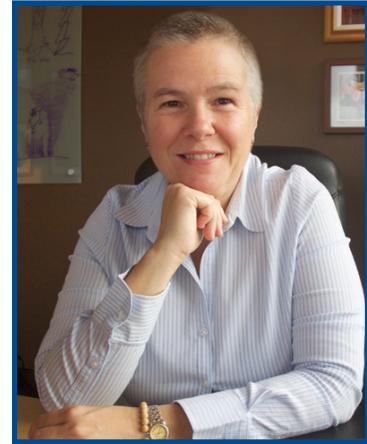
Genetic Alliance's network includes more than 1,200 disease-specific advocacy organizations as well as approximately ten thousand universities, private companies, government agencies, and public policy organizations. The network is a dynamic and growing open space for shared resources, creative tools and innovative programs.

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## Dear Friends,

Since 1986, Genetic Alliance has systematically built a world-class disease advocacy platform. We transformed from a basic advocacy organization to a leading network of key stakeholders determined to transform health through personal empowerment.

Genetic Alliance staff embody the change we want to see in the world. We commit to keeping our focus on our mission above all else. We know that we can only ask openness, transparency and commitment from the systems around us if we ask these things of ourselves.



Today, we engage an array of stakeholders in all we do, whether fostering intense, open dialogue between adversaries, convening meetings where disparate stakeholders identify common purpose, or sharing resources typically considered proprietary. Through the programs and initiatives described in these pages, we are committed to helping systems identify new solutions and pathways for the common good.

Our narrative requires that we be present to the places in the ecosystem that are ready for transformation, those that are at the vanguard of change. It also requires that we work for those who don't have this luxury. In that light, we have devised three enterprises and a number of cross-cutting programs. Read on to explore BioTrust, Genes in Life, and Expecting Health. I am confident that you will be compelled to join us on our quest for better health.

Sincerely,  
Sharon F. Terry, President & CEO

### COUNCIL

Sharon Terry, MA President and CEO

Kemp Battle Secretary and Treasurer

Managing Director, Tucker Capital Corporation; Folklorist and Writer

Greg Biggers CEO, Genomera

Kelly Edwards, PhD, MA Associate Professor, Bioethics and Humanities,  
University of Washington School of Medicine

Shantanu Gaur, MD Co-founder, SGL Medical

### STAFF

Natasha Bonhomme Vice President of Strategic Development

Mark Petruniak New Media Coordinator

Rachel Koren Program Assistant

Sharon Romelczyk, MPA Program Manager

Marybeth McAfee, MA Associate Director of Health Information

Sharon Terry, MA President and CEO

Tetyana Murza, MES Managing Director

Jeff Thomas Administrative Assistant

James O'Leary, MBA Chief Innovation Officer

Thalia Thompson, MPH, CHES Maternal and Child Health Program Manager

Mary Peckiconis, MA Office Manager

# 2013 by the NUMBERS

Only 30%



of respondents to the *Survey on Access to Care for Individuals with Genetic Conditions* had ever been referred to a support and/or advocacy group by a healthcare provider.

200

quality tools in the Advocacy ATLAS



94%

of Americans believe that participation in clinical research is essential for the advancement of medical science, but less than participate in clinical trials.



5%



650

disease advocacy organizations

10k

conditions

40k

unique visits in first

7

months



50

years of newborn screening

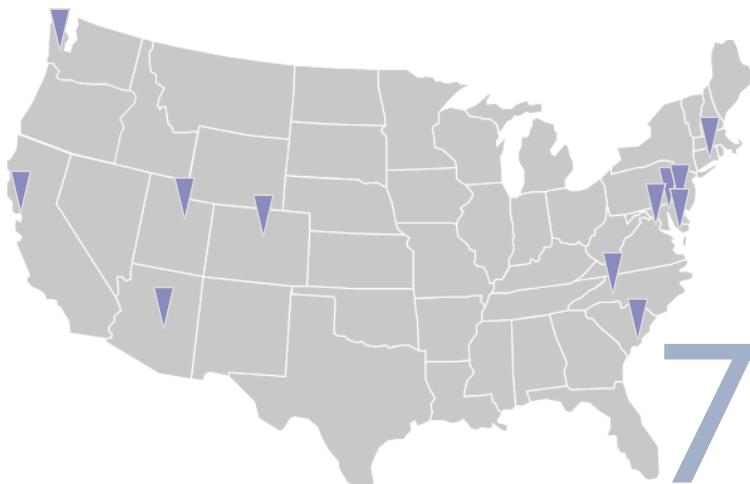
Newborn  
Screening

4k

gold pins distributed for Be Bold, Wear Gold campaign

2

years of awareness-building and education through Baby's First Test



15k members in the Genetic Alliance Rare Disease & Genetic Condition Support Community



21 articles in 9 journals



# Advocacy: Magnifying the Consumer Voice

Since its founding, Genetic Alliance has been the voice of advocacy in genetics. We support both individuals who advocate on behalf of themselves or others, as well as thousands of disease advocacy organizations that support communities of individuals with common need. We empower individuals and families to turn obstacles into opportunities, take control of their own health decisions and information, and make their voices heard.

In 2013, Genetic Alliance continued to expand upon existing advocacy resources and launched three exciting new tools for the community.

## Advocacy ATLAS

It is often challenging for individuals with genetic conditions and their families to locate and access the resources, services, and support they need. To make it easier, Genetic Alliance partnered with Parent to Parent USA and Family Voices. Together, we compiled existing advocacy and leadership tools from our networks of parents, parent advocacy groups, disease-specific advocacy organizations, disability groups, and others. We engaged families throughout the process and found that there was a particular need for tools on leadership and advocacy. To meet this need, in September 2013, we soft-launched the Advocacy ATLAS ([www.geneticalliance.org/advocacy-atlas](http://www.geneticalliance.org/advocacy-atlas)), which features 200 quality tools to empower individuals and families to advocate in ten topic areas. Outreach and promotion of the ATLAS will take place in 2014 and beyond, along with analysis of its utilization and impact.



**Mast Cell Disease Symptom Help**  
By Trish45 - New reply 3:31 pm  
Discussion in How my disease has changed my life - 17 replies

Hello, I'm new here. I have mast cell disease. I'm wondering if anyone can help me understand some of my symptoms. I think I'm most concerned about the numbness and if this will be temporary or permanent ...

**MCAD pgd2 and aspirin**  
By floating\_cloud - New reply 3:23 pm  
Discussion in How my disease has changed my life - 9 replies

Hello, has anyone got experience in using aspirin for blocking the pgd2? Dr Castells speaks of this working well for some people. I am thinking of starting out on the low dose of 81 mg while on hospital ...

**Mac Tel Support**  
By kimmt954 - New reply 3:23 pm  
Discussion in My disease has no treatment - 3912 replies - Photos |||

This is a continuation of the discussion called "Idiopathic Juxtafoveal Telangiectasis Type 2" started on July 15 2012, under the NORD Rare Disease Support Community, in the My Disease Has No Treatment ...

**How to track down a CSF leak?**  
By morglein - New reply 3:22 pm  
Journal - 6 replies

Background: I have hEDS/MCAS and live in Southern Oregon, where specialist doctors are few and far between in this area. I would like to find a doctor who can help me figure out if I'm suffering a CSF ...

**Severe pain, but don't know what's causing it..?**  
By Behcet's Girl - New reply 3:20 pm  
Journal - 28 replies

For the past few days I've had severe pain in my hips/thighs. It's so bad that I can't walk without wanting to cry. I've put ice on it, tried a heating pad, taken pain medicine, etc. Nothing is working ...

**the middle of the night and day gang**  
xxxxxxxxxxxxxxxxxxxx

**Searching for validation**  
By iubyloa - New reply 3:00 pm  
Journal - 17322 replies - Photos |||

Day or night there's someone here To lend advice or just lend an ear. Friendships have grown from this sacred place Even though I've never seen your face ~ Wenz A gentle word and kind advice You are sure ...

## Inspire Community

We partnered with Inspire to create the Genetic Alliance Rare Disease & Genetic Conditions Support Community. This online forum connects patients, families, friends, and caregivers and provides another avenue for Genetic Alliance to engage people with genetic diseases. We share a weekly advocacy tip with the Community to supplement approximately 1,000 posts by members each month.

## Disease InfoSearch (DIS)

Throughout its history, Genetic Alliance has aggregated information on support and disease advocacy organizations. What began as a spiral-bound book is now a searchable, one-stop shop for information on genetic conditions.

Genetic Alliance formally launched DiseaseInfoSearch.org in March 2013 as an online database of almost 10,000 conditions and their related support and advocacy networks. The site is a credible resource for families and healthcare

providers hoping to learn more about the signs and symptoms of conditions, how to access support resources, and potential research opportunities. With the help of our medical and research partners, we devised and tested algorithms that pull information on each condition in DIS from public databases - including OMIM, PubMed, Genetics Home Reference, Gene Reviews, Genetic Testing Registry, and ClinicalTrials.gov - to complement information provided and vetted by the disease advocacy organizations.

**DISEASE INFOSEARCH**  
Powered by Genetic Alliance

Home    About    List an Organization    Learn More    Contact

Search
Advanced Search

[Close LiveSearch](#)

- [Cutis laxa, neonatal, with marfanoid phenotype](#)
- [Marfan syndrome](#)
- [Marfan Syndrome type 2](#)
- [Marfan-Like syndrome](#)
- [Marfan-like syndrome, Boileau type](#)
- [Marfanoid hypermobility](#)
- [Marfanoid mental retardation syndrome autosomal](#)
- [Microcephaly glomerulonephritis Marfanoid habitus](#)
- [Pectus excavatum](#)

**Introducing Disease Info**

Are you looking for disease information? Simply type in the name of the disease and get information from a database of over 10,000 conditions, support groups and foundations.

Sign up for Reg4All



Answer questions about your health. Let resources you select come to you. ... all while protecting your privacy.

# BioTrust

Translational science and the resulting drug discovery and development are seriously challenged. To achieve a revolution in health, a trust environment is necessary. In 2013, the BioTrust enterprise examined policies and created products to enable and increase the participation of individuals, families and communities in research. The BioTrust Ethics Team, together with the Genetic Alliance Institutional Review Board, provides oversight. BioTrust is a living laboratory for accelerating amazing theories and applying lessons learned.

## Genetic Alliance Registry and BioBank



Genetic Alliance Registry and BioBank (GARB) was the first patient-powered research network and is the only lay-run, cross-disease biobank and registry. It serves five disease advocacy organizations and holds collections of biological samples and clinical information for six conditions. To enhance its capabilities even more, in 2013, GARB became a member of the REDCap Consortium, which gives advocacy organizations a phenomenal platform on which to build registries.

The REDCap application allows users to build and manage online surveys and databases quickly and securely. As of this writing it is being used for more than 125,000 projects by over 165,000 users.



## Platform for Engaging Everyone Responsibly

To engage potential research participants and ease them into the research process, Genetic Alliance partnered with Private Access to build the Platform for Engaging Everyone Responsibly (PEER). In the past year, PEER received several first-place awards totaling more than \$1.4 million and is now the basis for a number of portals serving individuals and communities across the US and worldwide.



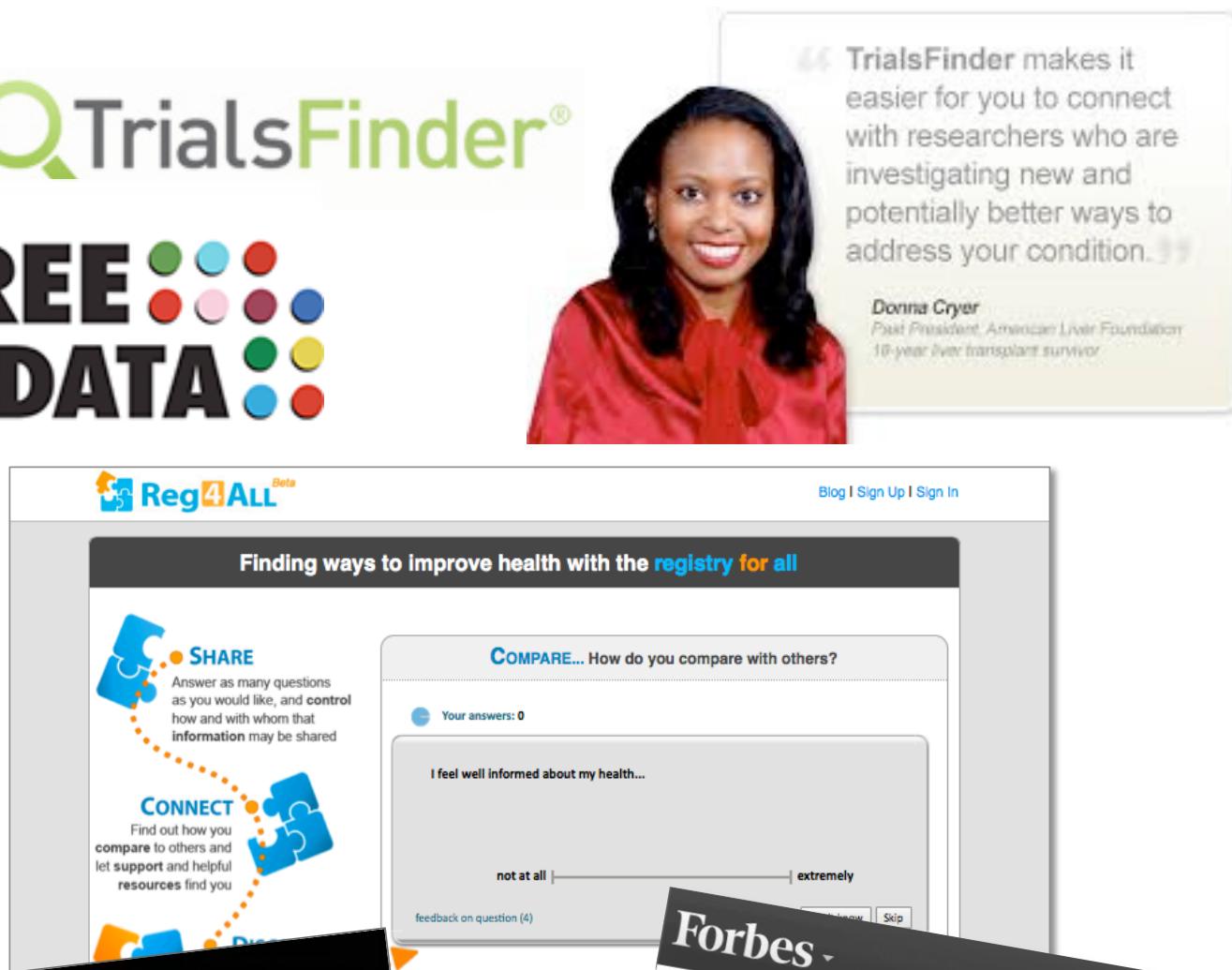
PEER enables individuals to determine their data sharing, privacy, and access preferences, and to change them over time. Individuals who desire in-depth participation can self-report common data elements that span diseases

and then continue to disease-specific data elements. The architecture of PEER also allows individuals to request that their electronic health data be added to the system under the HITECH Act.

Individual researchers can query the database and find information that has been properly consented for their access and use. By providing affected individuals with the education and means to connect directly with the research community, PEER will facilitate patient engagement in the research process, benefiting condition-specific as well as cross-disease clinical research endeavors.

PEER is the technology behind TrialsFinder, Reg4ALL, Free the Data, and seven sites for the PDUFA Patient-Focused Drug Development initiative.

An example of a portal, which is customizable for any organization, can be found at: [www.wepsicklecell.org/tell-the-fda](http://www.wepsicklecell.org/tell-the-fda)



# Genes in Life

Genetic Alliance puts genetic and genomic information into context for individuals and families. From family health history to whole genome sequencing, our combination of accessible, culturally competent print and web resources, multimedia, and health information technologies empowers people to make healthy decisions.

## GenesInLife.org

Launched in August 2013, GenesInLife.org provides accurate, accessible information on genetics and health for individuals and families as well as healthcare professionals. Information is packaged in five main topic areas, each supplemented by additional print and web resources from both Genetic Alliance and our network.

1. *Genetics 101* walks you through the basics of genetics and inheritance.
2. *Genes and Your Health* answers common questions on the effects genes can have on your health and stresses the importance of incorporating family health history into your healthcare routine.
3. *After Diagnosis* offers a series of “next steps” and resources for individuals with a genetic condition, from information on insurance to transitioning from pediatric to adult-based healthcare.
4. *Testing and Services* provides explanations of the different types of genetic tests and screens as well as the healthcare providers that might prescribe these services.
5. *Research* includes information on the process of clinical trials and human subjects research along with resources for getting involved.

Please help us improve GenesInLife.org with this [short survey](#). Follow us on:

WELCOME GENETICS 101 GENES & YOUR HEALTH AFTER DIAGNOSIS TESTING & SERVICES RESEARCH

## How do my genes affect my health?

Your health depends on a combination of your genes, choices, and environment.

[see why](#)

[search](#)

### Your Genetic Testing Questions Answered

There are many types of genetic tests that can help you and your doctor make decisions about your health and care.

[DIFFERENCE BETWEEN SCREENING AND TESTING](#)

[WILL MY INSURANCE COVER THE COST?](#)

**Genes in Life Blog**

**Welcome**  
July 18, 2013 - 9:16am

Welcome to the Genes in Life blog! Every two months we will focus on a particular area of health and genomics. We'll highlight current events, exciting discoveries, and more.

[READ MORE](#)

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**On Twitter**

RT @GABioBank Make the exchange between researchers and #IRBs easier. Treat each other like... - [View](#)

## Consumer Engagement and Empowerment

As the National Genetics Education and Consumer Network (NGECN), over the past year Genetic Alliance has focused on identifying and addressing the needs of individuals with genetic conditions and their families.



As part of this far-reaching task, we assessed consumer engagement in the seven Regional Genetics Collaboratives (RCs) by conducting a survey as well as one-on-one discussions about needs, gaps, and ways of encouraging consumer participation

within the RCs. We then advised the RCs on creating messages that reach and engage different types of audiences.

We also disseminated the *Survey on Access to Care for Individuals with Genetic Conditions*, which explored insurance coverage, continuity of care, care coordination, transition from pediatric to adult healthcare, discussions on family health history, and support/referral to support. These six priority areas were chosen because of their national importance and potential to impact the quality of care a patient receives. The data collected is the first of its kind, summarizing responses on access to care and support from individuals reporting one or more genetic condition(s) across the life course.

We collected 1895 responses from people living in the US who have been told by a provider that they [or their child] have a genetic condition. Select findings (for individuals with genetic conditions ages 0-92) are as follows:

- 43.3% had unmet needs for genetic counseling
- The top barriers that prevented individuals from getting the genetic counseling they needed were:
  - Cost (18.9%)
  - Insurance coverage issues (17.1%)
  - Did not know where to go for treatment (9.3%)
  - Not available in area/transportation problems (8.6%)
- 41.8% never or rarely get the social and emotional support they need from a healthcare provider or support/advocacy group



## Powerful Patient Data

Genetic Alliance and Intermountain Healthcare co-hosted a summit on genomics and family health history in health information technology May 30-31, 2013, in Salt Lake City, Utah. The goal of the summit was to ensure that patient information, specifically family health history (FHH) and genomic data, is incorporated, shared, and used within the electronic medical record (EMR) to provide better patient care. At the summit, participants brainstormed concrete actions to meet this goal, heard from luminaries in the field, and viewed demonstrations of the latest technologies.

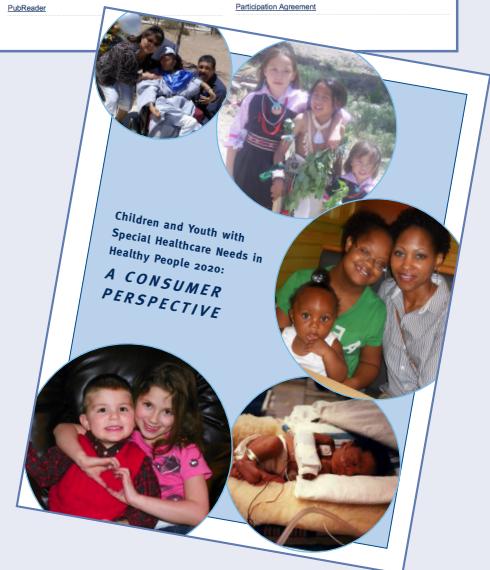
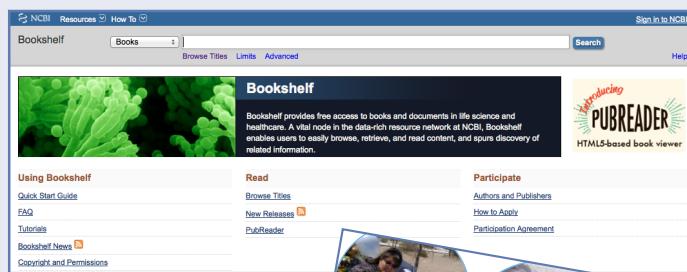
Outcomes of the conference included:

- A defined “use case” that illustrated effective collection, flow, and utilization of this patient data within our information technology ecosystem
- Increased awareness of current use of genetic/FHH data within EMRs
- Increased awareness of tools for collection of FHH
- Next steps regarding standards and public policy, including inclusion of FHH in Meaningful Use recommendations

## Developing and Disseminating Resources on Genetic Services

- Incorporated genetics education materials in the NCBI bookshelf
- Produced Spanish version of the *Guide to Genetic Counseling*
- Published *Children with Special Healthcare Needs in Healthy People 2020* monograph in conjunction with Family Voices

- Disseminated genetics education materials to over 1,000 genetics professionals, researchers, and consumers
- 10 articles published in Exceptional Parent Magazine



# Expecting Health

While newborn screening activities remain a key focus, we are increasing our presence throughout the perinatal period. As prenatal screening becomes more available to the public, Genetic Alliance is well positioned to provide the tools that will help families make the most informed decisions possible.

## National Newborn Screening Clearinghouse

On September 7, 2013 Genetic Alliance celebrated the second birthday of Baby's First Test ([www.babysfirsttest.org](http://www.babysfirsttest.org)), the nation's most comprehensive newborn screening web tool. Since its launch, the site has received nearly 205,000 visits, and the average number of monthly visits has tripled.



## Engaging Individuals and Families



The Consumer Task Force on Newborn Screening was created to engage stakeholders with an interest in newborn screening policies, activities, and current events. Members complete a one-year program with three components: training, project development, and project execution. This equips stakeholders with the skills and knowledge to make an impact in the maternal and child health community and expands the capacity of both the members of the Task Force and their respective organizations and communities. Members build long-lasting bonds with fellow parents and also establish themselves as experts in the arena of family advocacy.

In addition to their community and educational projects, the members of this year's Task Force participated in the 2013 Joint Newborn Screening and Genetic Testing Symposium and International Society on Neonatal Screening Meeting, the largest gathering of newborn screening professionals of the year. Task Force members highlighted the needs of families on the parent/patient panel, which many meeting participants said was the most important event of the entire four-day conference.

Left to Right: Amanda Rose Adams, Tomiko Brooks, Kelly Huber, Kay Kelly, Michelle Leeker, Sara Lockie, Molly Martzkie, Kary Padding, Renee Stapley, Colleen Zak

## Funding Innovation to Fill Gaps in Training and Education

Genetic Alliance gave out five Challenge Awards this cycle, with an emphasis on moving beyond traditional website engagement. To date, nearly 1 million dollars has been requested to fill the gaps in healthcare provider training and public education on newborn screening at the local, state, and national level. Recipients educated and engaged midwives, NICU nurses, and home health care workers; translated materials into Russian, Chinese, Spanish, French, and Arabic; and compared the utility of standard brochures and video interventions for new parents from lower socio-economic levels. The cornerstones of the selected projects were scalability, sustainability, and novel exploration of the stated problem.



## Be Bold, Wear Gold Campaign

September 2013 marked the 50th anniversary of newborn screening. To commemorate the occasion and celebrate newborn screening awareness month, Baby's First Test launched the Be Bold Wear GOLD campaign, which was designed to raise awareness and increase education about this invaluable public health program.

Throughout September, families, advocates, state health departments, legislators, and state laboratories wore gold pins and the color gold to express their conviction that all babies deserve a healthy start. Genetic Alliance created "Celebration Kits" with materials and information that states can use as they plan their local awareness programs.

## Beyond the Bloodspot

On the eve of newborn screening's 50th anniversary, we convened a meeting to discuss how to ensure the program's continued success. Over the last decade, advocacy organizations and individuals with children affected by rare conditions have become increasingly vocal about the need to expand research and the list of conditions on state newborn screening panels. We examined the changing landscape of screening and debated what lessons can be learned from both emerging technologies and established population-based screening programs. More than 100 people participated from across the country and committed to continuing the dialogue about how to shape a healthcare system that can support both public screening of newborns and advancing technologies.

## Financials

Genetic Alliance's work is supported by a blend of funding from government grants and contracts, industry and corporate support, individual donations, and fees generated by services and events. We continually work to diversify our funding sources. A key part of this diversification plan is to secure strategic, fee-for-service partnerships that leverage our expertise, serve the needs of the field and community and bring greater financial sustainability to the organization.



## Statement of Activities and Changes in Net Assets

as of September 30th of each year

	2013 Total	2012 Total
<b>Revenue and Other Support</b>		
Government Contracts and Grants	\$1,955,242	\$2,196,518
Biobank Program Revenue	289,753	156,438
Special Events	115,718	227,152
Contributions	22,218	149,340
Listserv Income	300	2,000
Interest Income	41	54
Other Revenue	15,027	60,594
 Total Revenue and Other Support	 2,392,580	 2,792,096
Net Assets Released from Restrictions	-	-
 Total Revenue and Other Support	 \$2,392,580	 \$2,792,096
 <b>Expenses</b>		
Program Services		
General Programs	\$2,639,234	\$2,683,107
Policy	-	1,516
Supporting Services		
General and Administrative	17,283	38,923
Fundraising	86,302	74,228
 Total Expenses	 \$2,742,819	 \$2,797,774
 Net Increase (Decrease) in Net Assets	 (350,239)	 (5,678)

## Operating Fund Sources

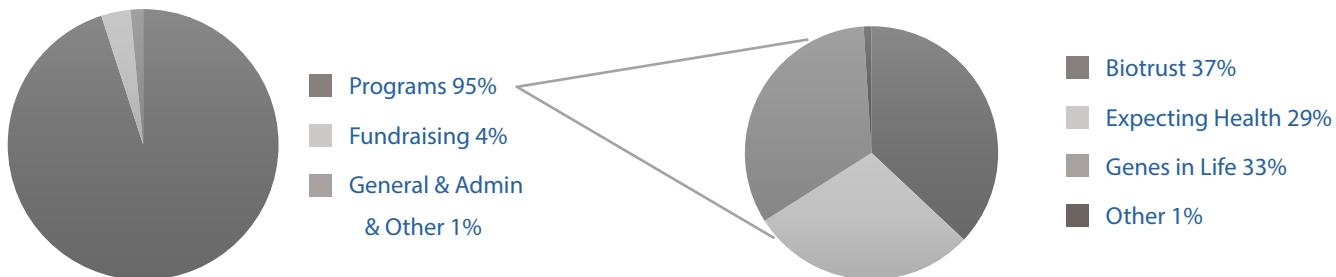


# Statement of Financial Position

as of September 30th of each year

Assets	2013	2012
Cash and Cash Equivalents	\$219,313	\$55,973
Accounts Receivable	157,816	84,765
Grants Receivable	220,601	586,763
Prepaid Expenses	28,760	31,130
Deposits	6,595	6,595
Furniture & Equipment	553,307	676,349
<b>Total Assets</b>	<b>\$1,186,392</b>	<b>\$1,441,575</b>
<b>Current Liabilities</b>		
Accounts Payable and Accrued Expense	\$207,802	\$105,144
<b>Long-term Liabilities</b>		
Deferred Rent	47,544	55,146
<b>Total Current Liabilities</b>	<b>255,346</b>	<b>160,290</b>
<b>Net Assets</b>		
Unrestricted	931,046	1,085,031
Temporarily Restricted	-	196,254
<b>Total Net Assets</b>	<b>931,046</b>	<b>1,281,285</b>
<b>Total Liabilities and Net Assets</b>	<b>\$1,186,392</b>	<b>\$1,441,575</b>

## Fund Spending





**Genetic Alliance**

WWW.GENETICALLIANCE.ORG

T: 202.966.5557

F: 202.966.8553

[info@geneticalliance.org](mailto:info@geneticalliance.org)

4301 Connecticut Ave., NW  
Suite 404  
Washington, DC 20008-2369