GENETIC ALLIANCE

ANNUAL REPORT 2019

CONTENTS

PONDERINGS

As you will see in this annual report, Genetic Alliance remains committed to giving people and communities the tools they need to transform the research enterprise. Critical to us is the ability of people to share their lived experience in a way that is easy to do and useful to researchers. To that end, this was a very big year for Genetic Alliance. We undertook a major collaboration with LunaDNA. For a long time we have sought a partner who understood the importance of people at the center, not in name only, but a system that codifies the individual as the driver, the decider, the central focus. With Luna, a public benefit corporation, we are well on our way to achieving this.

We are vigorously engaged with PCORnet, now for five years. During this time, we have developed many engagement tools, both for giving researchers concrete ways to integrate individuals and communities into research from the inception of an idea through to changes in clinical practice, and tools to evaluate how well they are doing.

We are excited to hone our focus in 2020 and build a deep and robust Bootcamp for registries and biobanks, and up the game for all of the disease advocacy organizations with whom we work. As our engagement with LunaPBC expands, we hope to disrupt the current biomedical research paradigm to accelerate solutions for those who suffer.

Sincerely,

Sharon Terry

PRESIDENT AND CEO

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Managing Director, Tucker Capital Corporation
Catalytic Change Agent

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ENGAGEMENT

Genetic Alliance is committed to a vision whereby nothing happens for or to a group of stakeholders without meaningful engagement. This is the starting place for any activity or project. Therefore, because we have worked for 33 years as grassroots community leaders from the ground up with diverse stakeholders, we have excellent expertise in engaging communities and organizations. We provide that expertise in and through our projects, and offer as a core service to other entities.

National Patient-Centered Outcomes Research Network

Over the course of 2019, Genetic Alliance contributed to strategy, processes, policies, and engagement planning for the Network.

5 Years

KEY PART OF PCORNET'S COORDINATING CENTER

In its role as the Coordinating Center, Genetic Alliance supports collaborative engagement activities in PCORnet by convening the PCORnet Engagement Committee, a multidisciplinary group charged with advocating for, embedding, and facilitating people-centered research across the Network.

Genetic Alliance brought expertise from the perspective of the community—from the ground up—and elevated learning; co-production of tools; monitoring and assessment of participant-driven research; and established methods to make it easier for people in the healthcare system to access and share their data.

In collaboration with this Committee, the Coordinating Center developed a suite of tools to enable communities to better define, plan, implement, and measure engagement activities.

Additionally, Genetic Alliance conducted a robust engagement assessment to analyze engagement strategies and activities being utilized across PCORnet. Through this project, the team **surveyed and interviewed all 35 PCORnet networks** to identify best practices, challenges, and lessons learned, as well as a compilation of products and tools.

Patient Industry Engagement Project

In September 2019, Genetic Alliance engaged key representatives from advocacy groups and industry to increase PCORnet's knowledge and capacity about patient advocacy organizations and industry partnerships as an accelerator for engagement in every aspect of research. In November 2019, Genetic Alliance will plan and convene a one-day workshop for PCORnet.

PROJECT OBJECTIVES

- Understand the basic characteristics of these partnerships models
- Determine resources and talent needed to deploy similar models in PCORnet
- Define which models should be emulated and scaled
- Capture available support and interest in scaling these models

The National Genetics Education and Family Support Center

Companion project to the Regional Genetics Networks (RGNs) is led by three organizations working together: Genetic Alliance, Family Voices, and Parent to Parent USA.

Supported RGN efforts to reach communities in the U.S. Territories – including Puerto Rico and The U.S. Virgin Islands

40 NEW RESOURCES

Completed a refresh of the Advocacy ATLAS adding new and updated resources to the online toolkit

2 SCHOLARSHIPS

Provided scholarships for family representatives from the RGNs to attend the first Cultural Agility training at AMCHP

The National Genetics Education and Family Support Center (Family Center) continued to provide information on genetic services and support as well as provided training for families.

WEBINARS

Hosted an educational webinar on November 8th, which highlighted successful examples of partnerships between NYMAC RGN, Midwest RGN and their respective regional family leaders. (58 attendees and 9 recording views).

Hosted an educational webinar on August 27th to highlight advocacy resources such as DiseaseInfoSearch.org and Advocacy ATLAS. (62 attendees and 75 recording views).

PRESENTATIONS

Attended and exhibited at conferences such as the Family Voices annual meeting, Association of Maternal and Child Health Programs annual meeting, Global Genes RARE Patient Advocacy Summit, and American College of Medical Genetics and Genomics annual meeting.

Presented at the Southeast Regional Genetics Network and Heartland Regional Genetics Network Annual Meetings, participated in panels (at the SERN meeting and at the Heartland meeting – both discussions were focused on the role of consumer advocacy in genetics).

The Family Center showed measurable success in reaching families through robust online resources.

Disease InfoSearch

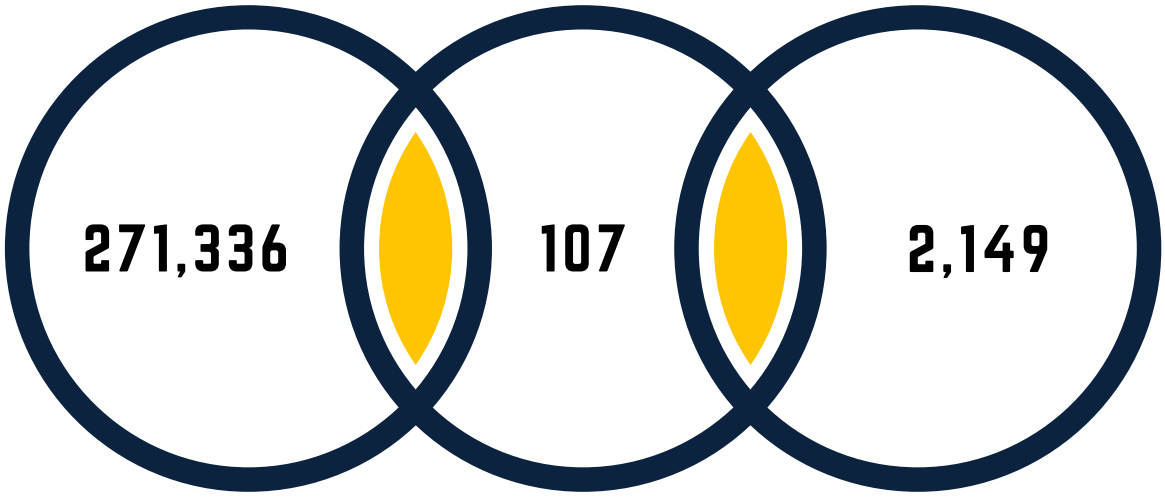
Online source on over 10,000 conditions aims to help families avoid a broad internet search containing high literacy, poorly contextualized information about the condition.

Genes In Life

GenesinLife.org is a place to learn about all the ways genetics is a part of life, with a specific focus on genetics services, including genetic testing and working with genetics professionals.

Advocacy ATLAS

A web-based toolkit specifically designed to provide individuals with special healthcare needs and their families tools and strategies to advocate for whatever they may need.



273,592 UNIQUE PAGES VIEWS

- 306,530 Total Pageviews Across Platforms

RESEARCH DONE DIFFERENTLY

The culture of health research is evolving. In the past, academic and industry investigators were its primary stewards, but today communities of people can, and do, drive their own research. We represent a network of more than 10,000 health organizations whose primary partners are individuals, families, and communities. Together we respond to people's needs through the transformation of health systems. Collaboration and partnership are integral to our model, and we work with our partners to pilot and test a variety of new methods, tools, and technologySolutions.

Platform for Engaging Everyone Responsibly (PEER)

Genetic Alliance's signature research tool – the Platform for Engaging Everyone Responsibly (PEER) – enables individuals, families, and communities to drive research priorities by creating customizable, dynamic, and accessible community research portals. Participants in PEER contribute their patient-reported outcomes and other health-related data for research using the platform's award-winning tools for data sharing. These tools place control in the hands of participants, empowering them to broadly share their data with a variety of researchers while still addressing individual concerns about privacy and access.

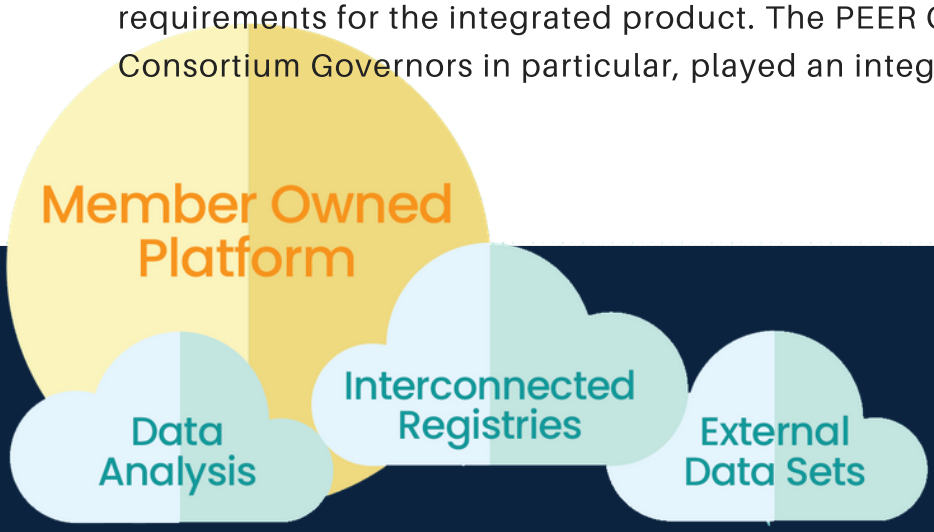
25 communities

50,000 individual users

On January 22, 2019, Genetic Alliance and LunaPBC announced a novel partnership to integrate PEER with the LunaDNA platform.

The integration combines the community-facing aspects of PEER with LunaDNA's superior tools for data collection and analysis, and their mechanism for giving people control, and ultimately shares in the company for their participation. This results in an enhanced platform for community-based research that recognizes people as true participants. The partnership empowers Genetic Alliance and LunaDNA to create seamless solutions to support individuals, disease foundations, advocacy organizations, and other community organizations -geographic, affinity, religious- while also powering disease research at scale. Shared values across both organizations ensure the ongoing focus of honoring a person's preferences and rights for data transparency, privacy, and control while accelerating science and creating shared value.

Over the course of 2019 the two organizations began the process of merging PEER with LunaDNA, working closely with PEER communities to define the requirements for the integrated product. The PEER Consortium, and the PEER Consortium Governors in particular, played an integral role in this process.



LunaDNA is a community-owned platform for health research that recognizes an individual's health data as currency with which to acquire shares of ownership in parent company LunaPBC. LunaDNA members contribute a variety of health information, including survey response, genetic information, and as of the summer of 2019, electronic health records. Researchers from nonprofits, for-profits, disease organizations, and research communities can then request access to LunaDNA to conduct research studies with participant-consented data. The data stays in the system and is accessible via a "research sandbox", ensuring that members' wishes regarding use of their data remain connected to the data over the course of its lifespan.

Community Engaged Network for All (CENA)

In March 2019, Genetic Alliance concluded its CENA project (Community Engaged Network for All) and transferred its network and gained knowledge into the Program for Engaging Everyone Responsibly (PEER). CENA was a collaborative composed of Genetic Alliance, University of California San Francisco, University of California Davis, and Private Access; and disease advocacy organizations chosen for this specific project, representatives of which are collectively referred to as the Steering Committee.

CENA people-driven communities participate in and lead research activities with varying roles and intensities across all phases of the research process.

The disease advocacy organizations (DAOs) were selected through a competitive process based on their readiness to participate in the PCORI PPRN vision.

The organizations serve a range of conditions from rare to common, the organizations' members are geographically diverse, and cover a wide spectrum of demographics and socioeconomic status.

During the course of the five years, the DAOs participating in CENA included Alive and Kickn, Alström Syndrome International, The Association for X and Y Chromosome Variations, Asthma and Allergy Foundation of America, Celiac Disease Foundation, Dyskeratosis Congenita Outreach, Inflammatory Breast Cancer Research Foundation, Hepatitis Foundation International, Joubert Syndrome Foundation, LymeDisease.org, MLD Foundation, National Gaucher Foundation , National Psoriasis Foundation, Pancreatic Cancer Action Network, PXE International, and The Fibrolamellar Registry.

NOTABLE ACHIEVEMENTS

Three core areas were enhanced through the work of CENA over these five years:

1. Shifting the culture of research from a traditional model to a patient-centered and participant-driven approach
2. Capacity to connect communities to each other and the research process
3. Expertise in developing or influencing the tools and approaches used to conduct research

With support from CENA, each of the organizations addressed research questions that emerged from their community.

Using tools shared across the network, these organizations prioritized research questions and built or improved upon systems to answer these questions in a timely manner. Organizations in CENA network benefited from an award-winning registry platform called Platform for Engaging Everyone Responsibly (PEER). Each community was enabled to launch an online registry for the condition(s) they represent and support. Genetic Alliance provided this cross-condition platform for engaging communities to safely share their information online, as well as community building and collaboration between condition communities.

PEER puts people at the center, managing their own data access and privacy settings. It allows for extremely cost-effective data capture from participants, including validated PRO and phenotype capture, in a real-world evidence manner. PEER employs an interface that is gamified to maximize retention and it is flexible to allow for continual fine-tuning or addition of questions, including changes based on input from advocacy organizations and academic research partners. Though the platform is deployed from servers managed by Genetic Alliance, each community customizes and branded it to enable the community to retain its own trusted brand.

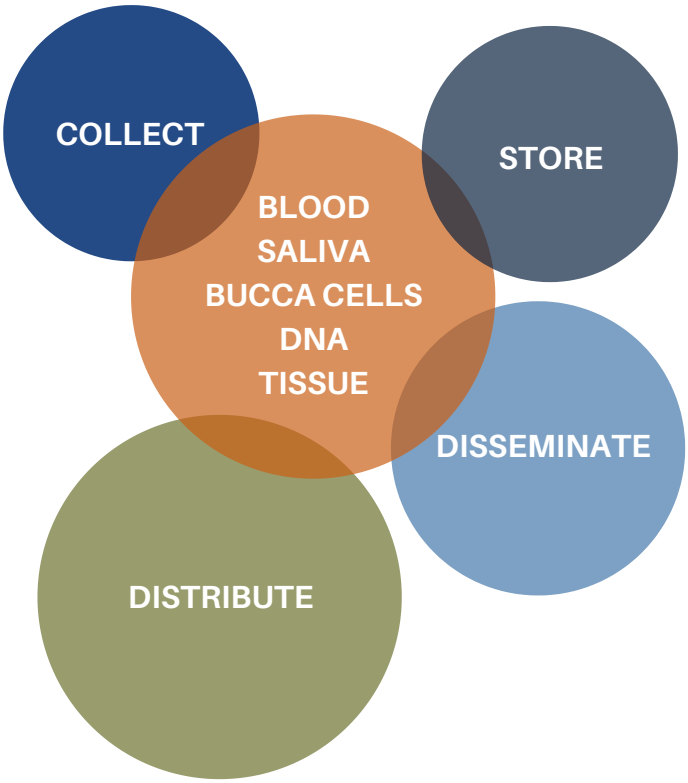
PROJECT OUTCOME

The advocacy-led communities were supported and grew through inter-community collaboration and the sharing of best practices. The CENA PPRN was a microcosm of the entire PCORnet data network since it included multiple advocacy organizations with dramatically varying disease symptomology, progression, body system involvement, frequency, available therapies, population demographics, and community engagement, all in one interoperable data network. Several disease advocacy organizations included associated conditions and therefore CENA represented a broad swath of diseases and issues.

Through UCSF's Mosaic platform, developed specially for CENA, groups piloted new methods of facilitating collaboration between researchers and participants by supporting a broadly accessible online environment where communities and researchers have equal voice in the development of research hypotheses. We believe significant benefits will continue to result from refining research hypotheses related to individual conditions in an open, transparent manner between researchers and participants, and between condition communities.

Genetic Alliance BioBank

The BioBank was founded in 2003 as a cooperative venture to decrease costs by increasing the "buying power" of a collective of communities. Individual communities, mostly disease advocacy communities, manage their own biobank under the Genetic Alliance's agreement with biobanks, cell banks, and other specialty repositories.



An online digital portal allows the organization to carefully manage collection kits sent to their members, bulk orders of such kits for conferences, accessioning of samples, extraction of DNA or other preparation, inventory, and distribution of samples. At this time, communities of 7 organizations are storing **more than 30,000 samples in the Genetic Alliance BioBank.**

Genetic Alliance Institutional Review Board (IRB)

Genetic Alliance IRB (IORG0003358) was founded in 2003 and is comprised of leading experts in human research protections. Genetic Alliance has Federal Wide Assurance # FWA00017292.

11
Initial Reviews Conducted

34
Continuing Reviews

12
Protocols Closed

Genetic Alliance IRB is one of 476 institutions nationwide that have joined SMART IRB, a platform designed to ease common challenges and burdens associated with initiating multi-site research. SMART IRB will help Genetic Alliance advocacy researchers involved in multi-site, NIH- funded studies streamline human subjects research, while ensuring robust protections for study participants.

The Genetic Alliance IRB is also serving as the reviewing IRB for a large whole genome sequencing (WGS) study. This is a landmark study for the Genetic Alliance IRB, which provided an opportunity to develop best practices and standards for reviewing WGS studies to ensure participant centricity and security when conducting research with whole genomes.

Disease InfoSearch (DIS)

Disease InfoSearch began as a printed directory and was added to the Genetic Alliance website in 2005 to make it a more accessible tool for mass sharing of disease support group information. In 2013 it became a standalone website with more extensive information about diseases. Over time, DIS expanded to include greater crowd sourced disease descriptions, listings of support organizations, and other resources such as links to websites, journal articles, ways to participate, news and events, and clinical trials.

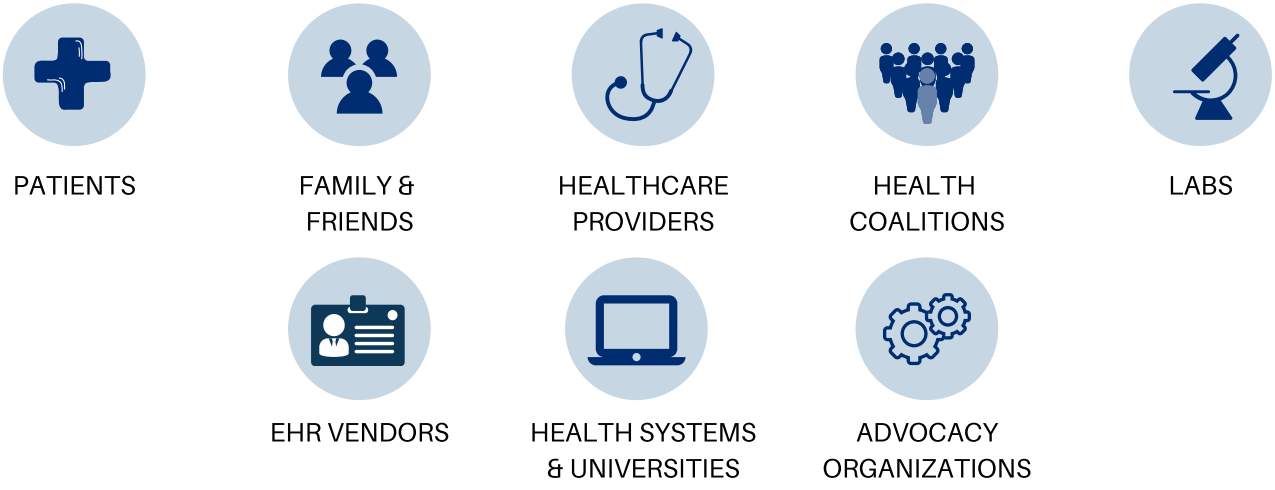
12,000
Active Users per Month

10,000
Diseases & Conditions

800
Support Organizations

DIS PARTNERS

Subject matter experts from disease advocacy organizations populate the information directly, ensuring that it is as credible, complete, and current as possible. In addition, DIS has a web service connection with National Center for Biotechnology Information databases, as well as the Genetics and Rare Disease Information Center, Online Mendelian Inheritance in Man (OMIM), and the European Union resource Orphanet, to permit greater integration and "one- stop-shopping". Researchers find DIS useful because it catalogues many essential attributes a researcher considers in choosing a focus, for both the disease and the advocacy group.



- Access high-quality, easy-to-digest information and resources from communities and curated databases
- Find support groups, clinical trial information, publications and research articles
- Pull relevant data fields through a secure API

International Rare Disease Research Consortium (IRDiRC)

Genetic Alliance was among the founders of the International Rare Disease Research Consortium (IRDiRC) almost 10 years ago. CEO Sharon Terry founded and led the Patient Advocacy Constituent Committee of IRDIRC as chair. She also serves, as she always has, on the Operating (formerly Executive) Committee.

In 2019, workgroups were created to advance new goals:

- All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.
- 1,000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options.
- Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients.

The group of funders, industry, researchers and individuals with a lived experience of disease are working relentlessly to figure out how to achieve these new audacious goals.

EXPECTING HEALTH

Expecting Health shares science-based and policy-informed information that reflects the lived experiences of individuals and their families. With a focus on pregnancy and newborn health, we utilize principles of community engagement and user driven design to reach diverse audiences. We do this through the power of relationships; convening the top experts; working with key leaders in health; and engaging with families at the center of the conversation.

NBS Family Education Program

The new Newborn Screening Education Program launched with a focus to gather information on how families learn critical health information during pregnancy and laid the foundation to build an educational tool for those interested in learning the basics of newborn screening.

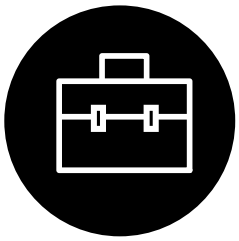
PROGRAM PURPOSE

EDUCATION

Increase the number of parents and families trained and educated on the newborn screening system annually

AWARENESS

Increase the number of partnerships to increase awareness of newborn screening and to facilitate the dissemination of materials to the target audiences



ADVOCACY

Increase the number of families/parents trained that report increased knowledge, skill, ability and self-efficacy to serve as leaders on newborn screening systems-level teams

OUTREACH

Increase the outreach to medically underserved populations and increase by 10 percent the number of medically underserved individuals that are aware of newborn screening and have access to information on the newborn screening system

FAMILY NEEDS ASSESSMENT SURVEY

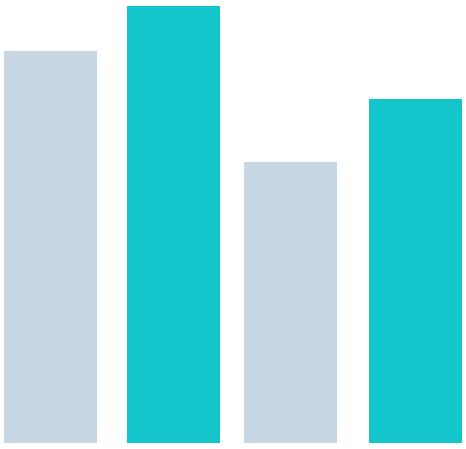
In 2019, one of the widest reaching NBS surveys was conducted. This survey was also one of the few newborn screening educational efforts to intentionally gather perspectives from medically underserved areas.

819 TOTAL PARTICIPANTS

258 PARTICIPANTS FROM MEDICALLY UNDESERVED AREAS

SURVEY RESULTS

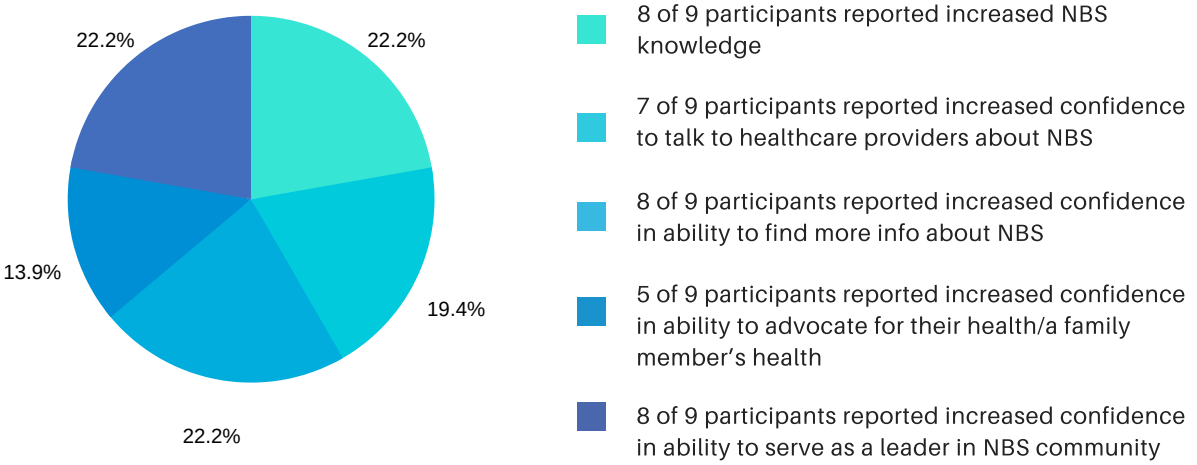
The survey looked at a subset of 500 participants recruited by RTI (our evaluation partner for this program).



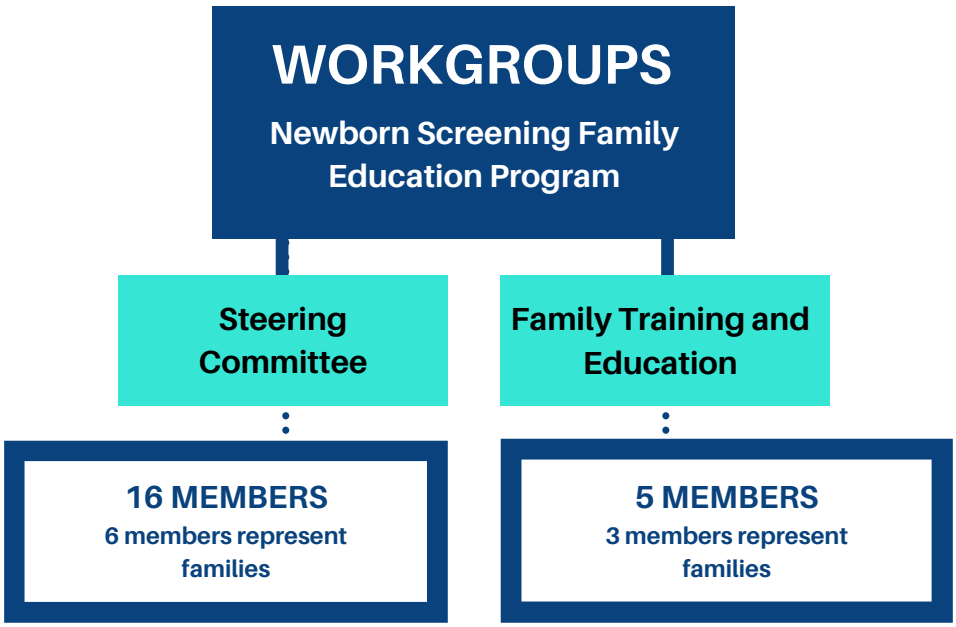
Only 54% of participants from medically underserved areas ***were aware of NBS*** compared to 67% of others. Only 49.5% of participants from medically underserved areas were ***first informed of NBS before birth*** (at the optimal time) compared to 61.4% of others.

TRAINING

In 2019, nine family members received training on the newborn screening system. Four participants were from medically underserved areas.



SUPPORT



Baby's First Test

In 2019, Expecting Health continued to expand the impact of the Baby's First Test Program.

4
Presentations by staff

1
Workshop on technical assistance

8
New resources developed

"A Newborn Screening Education Best Practices Framework: Development and Adoption" article was published in the International Journal of Neonatal Screening.

"Parent Reflections on Receiving Out-of-Range Newborn Screening Results" webinar was attended by 108 newborn screening stakeholders.

1,351 resources were distributed through online engagement

628,769 total users accessed Baby's First Test site

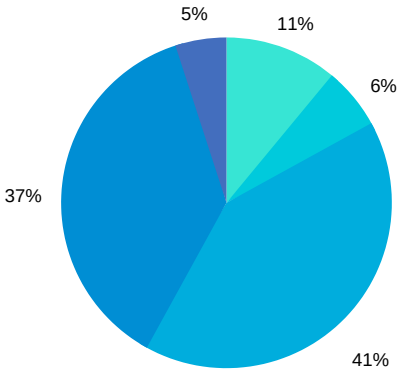
10.3 million people reached during NBS awareness month

Ask an Expert received **126 questions**

WHERE DO WE GARNER SUPPORT?

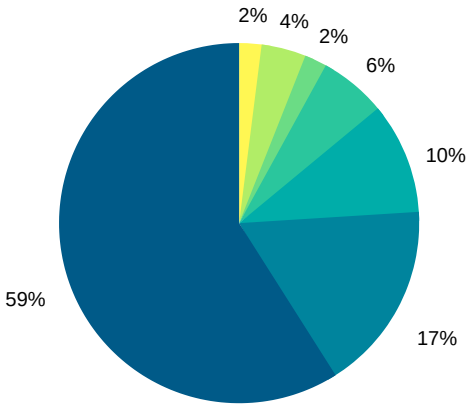
Genetic Alliance's work is supported by government grants and contracts, industry and corporate support, individual donations, and fees generated by services. We continually work to diversify our funding sources. A key part of this diversification plan is to secure strategic, fee-for-service partnerships that leverage our expertise, serve the needs of the field and community, and bring greater financial sustainability to our organization. For FY2019, a large portion of our funding came from PCORI through PCORnet, and additional funding came from the Health Resources and Services Administration, Maternal and Child Health Bureau, Genetic Services Branch.

GENETIC ALLIANCE CONDENSED FINANCIAL INFORMATION



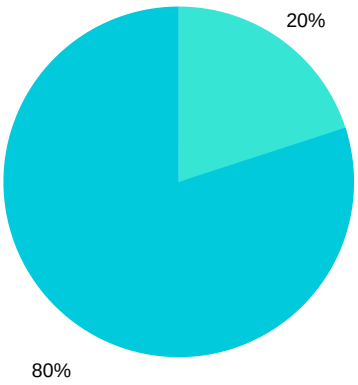
2019 Revenue

- PEER & BioBank
- Corporate Contracts
- Gov't Contracts & Grants
- Foundation Contracts
- Other



2019 Expense

- Office Expenses
- Professional Fees & Other
- Travel and Meetings
- Rent and Depreciation
- IT and Communications
- Partner Organizations and Consultants
- Salaries and Benefits



2019 Fund Spending

- General and Admin
- Programs

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