GENETIC ALLIANCE

ANNUAL REPORT 2020

IN THIS REPORT

3 Ponderings

4 Council and Staff

6 Promise for Engaging Everyone Responsibly

11 BioBank

12 Institutional Review Board

13 Disease InfoSearch

16 Policy Work

17 Expecting Health

PONDERINGS

Genetic Alliance, like many nonprofits and other businesses, was very challenged this year. At the end of the prior fiscal year, we determined that we needed a shift from trying to work within traditional healthcare systems to more grassroots, cutting edge activities. Then along came the pandemic. In some ways we were well positioned since we downsized a bit in early 2020. We are also grateful for the support of the Paycheck Protection Program.

Despite the financial struggles, FY2020 was a phenomenal year for us. In it we solidified a number of partnerships, including our biggest one with LunaPBC. Working with this public benefit corporation has allowed us to realize some very big and core dreams. We were able to move all of our technology for our now 17 year old registry system to Luna. We are grateful for the ability this gives us to enable dozens of advocacy organizations to work toward their research goals. And we look forward to 2021 as a year of immense growth in that regard. We will launch our bootcamp so that even more advocacy organizations and community groups can take charge of their path to better health. New organizations joined our Genetic Alliance BioBank and Disease InfoSearch, creating an excellent suite of resources for newly diagnosed and researchers.

Our maternal and child health program, Expecting Health blossomed this year despite the fiscal challenges.

We participated in a number of important federal policy efforts, and were most active in working to educate the congress on the Ending the Diagnostic Odyssey Act. We have great hope for that in the new congress.

Sincerely,
Sharon F. Terry, CEO

COUNCIL

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UNDERSTANDING RESEARCH NEEDS

Genetic Alliance supports disease advocacy groups in their quest to alleviate the suffering in the lives of their members, primarily by building research capacity for these organizations.

We recognize that recruitment, retention, engagement, data collection, and community building are all activities that are often best done in advocacy organizations. Collection of real-world evidence and patient reported outcomes are more robust when done by these communities, so it is integral that they have the necessary resources readily available to them.

In order to support these integral efforts, Genetic Alliance offers many services and programs ranging from the Genetic Alliance BioBank and Genetic Alliance IRB (an established IRB with over 125 approvals and hundreds of amendments and reviews) to the Genetic Alliance Promise for Engaging Everyone Responsibly (PEER) which currently has 26 members with a number of groups in the process of joining.

PEER

Promise for Engaging Everyone Responsibly (PEER) is Genetic Alliance's signature research program and platform. Since its establishment in 1995, PEER has evolved steadily to meet the needs of various communities through consistent policy and technology advancements. The inherent values that guide its development – community engagement, data collection across conditions, participant control of data, and a design that asks and answers the questions most critical to those participants – remains front and center today. The program allows communities to set up registries and/or studies to accelerate research on their condition. It is cross-condition, creating an interoperable database so that phenotypes, pathways, and other commonalities between conditions can be studied. People control their data and always have a string on it.

DELIVERING ON A PROMISE FOR ENGAGING EVERYONE RESPONSIBLY

Genetic Alliance Promise. Communities have a responsibility to support one another. Our success and failures are intrinsically linked together, we lift each other up to the benefit of us all.

- PEER is built by the community for the community.
- The true power within the community lies with the members.
- We empower our members to control their data, and act in accordance with their wishes.
- Every individual who provides data for research has a right to the results of that research. Built into our research processes are methods for responsibly sharing insights and research results back to the members of our communities.
- Research we support is for the benefit of individuals and communities. We will endeavor to only ask questions that have the potential to improve the lives of a group or humanity at large.

TECHNOLOGY

Over the course of 2020, we moved to rely on technology in a partnership with LunaPBC. We exceed the requirements of the host of privacy regulations and guidelines such as GDPR and CCPA. We have a library of validated instruments, a survey builder for your customized questions, connection to 700,000 EHR portals in the USA, and the capacity to connect with and store genetic and genomic information. We have also had our own IRB since 2003, so application and approval is easy. The system includes an analysis environment including Tableau, Jupyter notebooks, R and so on.

LunaDNA operates at the intersection of high tech and social impact, including a vocal stance on promoting people from subjects of research to partners in discovery, to drive a new generation of science. The technology and related services capture, normalize, and make research-ready multi-modal data including self-reported outcomes, real-world data, electronic health records, and DNA files. In contrast to institutional and geographic institutions that silo data, LunaDNA establishes a data-sharing relationship directly with individuals, which is geographically borderless and interoperable across digital communities. This enables organizations to study concepts broadly and delivers value to all participants. Public Benefit.

Corporation, LunaPBC manages LunaDNA. Our person-centric, privacy-by-design data stewardship model has received awards from notable institutions including the World Economic Forum and Fast Company's 2020 Most Innovative List.

COMMUNITY SUPPORT

PEER is a membership-based program, developed and run by Genetic Alliance, that provides support, guidance, and the required infrastructure needed to successfully create and launch a registry. Numerous opportunities for training, collaborating, and collective problem-solving allows organizations to benefit not only from the experiences of their fellow PEER communities, but also from the 25+ years of registry experience the Genetic Alliance team has to offer.

JOIN THE FIGHT TO END COVID-19

In March, Genetic Alliance through Disease InfoSearch and LunaDNA formed the COVID-19 registry to capture the experiences and insights contained within everybody's experience with COVID-19. The goal is to collect information which can be analyzed and studied in order to further understanding of the factors and circumstances surrounding COVID-19. In order to achieve this goal, all individuals were invited to participate, regardless of their COVID-19 status, to contribute data ranging from their self-reported answers to survey questions presented online to connecting and uploading their EHR or DNA sequences

COMMUNITY DEVELOPMENT THROUGH THE PROMISE FOR ENGAGING EVERYONE RESPONSIBLY (PEER)

On June 19th, Genetic Alliance and LunaDNA released an invitation to welcome five Black communities, without cost, to the LunaPEER community. A team of leaders in the fields of healthcare, science, and technology from the Black community chose organizations serving Black communities interested in social good to utilize our technology to benefit a Black community. Organizations will be given tools to develop and implement their own LunaPEER community data collection, engage with their communities, and power participant-centric research around health, social good, and/or quality of life, either through the creation of a registry or the release of surveys. In each registry:

- Individuals register for an account
- Individuals manage their data access preferences
- Surveys, which can include questions that are specific to the organization and questions that are generalized across the platform, serve to involve and engage participants
- Advanced data sharing including the option to connect electronic health records and upload DNA files is enabled
- Individuals can choose to share information on one or more conditions or topics, recognizing that many people experience many disparities, challenges, or have an interest in sharing information on multiple topics.

BY THE NUMBERS
PEER

40
OFFICE HOURS

Office hours were hosted to provide guidance, to understand and appropriately use the platform, assist PEER Sponsors in establishing registries and studies to collect data for research purposes, engage communities and recruit individuals to join studies/registries.

5
WEBINARS

The webinars on topics to help communities establish and maintain registries. Good planning is essential for the success of a registry. The decisions made during the planning phase have an impact on registry design, operations, and data analyses, as well as on the quality of the registry data.

26
COMMUNITIES

On the platform conduct studies to drive research for their condition.

BIOBANK

Genetic Alliance BioBank was founded in 2003 as a cooperative venture which aims 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. Individual organizations collect, store, and disseminate blood, saliva, buccal cells, DNA, and tissue of all types. An online LIMS system allows the organizations to carefully manage their inventory and monitor the accessioning of samples, submit work orders for the extraction of DNA or other preparation, and submit requisitions for the distribution of samples.

BY THE NUMBERS

7 COMMUNITIES STORING MORE THAN **80,000** SAMPLES

INSTITUTIONAL REVIEW BOARD

Genetic Alliance IRB (IORG0003358) was founded in 2003 and consists of leading experts in human research protections. Genetic Alliance has Federal Wide Assurance # FWA00017292. IRB Applications are accepted via an unique online webform which guides applicants through the process of describing their study in manner which is ready to be received by the Genetic Alliance IRB. The Genetic Alliance IRB has experience reviewing protocols for large-scale and multi-institutional studies with specific expertise in the areas of privacy review, registry management, survey-based studies, genomics and genetic testing, and return of results.

BY THE NUMBERS

58

Open protocols

16

Initial reviews conduscted

10

Protocols closed due to completion

DISEASE INFOSEARCH

Disease InfoSearch is an index of more than 10,000 diseases and their subtypes for individuals, families, health professionals, and researchers to easily find information. It was built with the purpose of making the dissemination of information easier and more accessible to all people. It is the mission of the Genetic Alliance to transform health via the engagement of individuals, families, and communities. Through the database, individuals are able to learn more about diseases that may affect them, their families, their communities, etc. They are also able to connect with others who may face similar challenges. By providing individuals with the information they need to know about a disease, Disease InfoSearch empowers them with knowledge and a vital, and oft-needed, support system.

People find support organization pages, mostly through natural searches (i.e., Google) and health professionals. Additionally, DIS is linked to each disease in PubMed, which is an extensive resource for researchers. Every paper on the condition indexed in PubMed has a LinkOut to Disease InfoSearch.

Researchers find DIS useful because it catalogues many essential attributes a research will consider in choosing a focus, for both the disease and the advocacy group. Lastly, pharma companies and other industry members come to us weekly, looking for research opportunities.

BY THE NUMBERS
DISEASE INFOSEARCH

DISEASES & CONDITIONS

10,000+

237 new diseases and conditions added in 2020

SUPOORT ORGANIZATIONS

865

46 new disease support organizations added in 2020

122,830 Unique visits to the site

228,093 Disease and condition pages viewed

74,913 PubMed article accessed

PATIENT INDUSTRY ENGAGEMENT PROJECT

In November 2019 Genetic Alliance convened a one-day workshop for PCORnet with 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. The specific goals of the meeting were to describe models for these partnerships and provide some recommendations for PCORnet leadership to consider as it continues its work on sustainability.

Key recommendations for PCORnet were provided from stakeholder interviews with key opinion leaders from advocacy organizations and industry Genetic Alliance conducted in preparation for the workshop:

- Improve the description of products and services to industry and patient advocacy groups
- Improve PCORnet's marketing materials and collaterals. Develop a clear summary of the number of patients by demographics, diagnosis, currently in PCORnet, such as a "Table 1", that could be easily shared with industry partners. Develop a clear description of PCORnet's data assets.
- Develop a map of PCORnet in terms of centers of excellence by disease area that could be used by industry to engage with PCORnet investigators to conduct studies.
- Develop an internal and external marketing campaign to highlight internally to investigators what PCORnet is, how to engage with it. Externally, diffuse the message about PCORnet and its capabilities, including through the open-houses where industry would be invited to discussions with investigators and partnered patient organizations.
- Highlight the critical role PCORnet can play in collecting PROs through its relationship with patient advocacy groups already participating in PCORnet
- Highlight the critical role PCORnet can play in increasing diversity in trial recruitment
- An educational offering that would develop a number of legal templates in plain language for patient advocacy organizations to use when working with industry.
- This work will only be successful if there is an authentic relationship developed with patient partners and patient advocacy organizations.

POLICY WORK

EXPANDING ACCESS TO WHOLE GENOME SEQUENCING

Nearly 80% of all rare diseases have a genetic cause, and half of rare disease cases impact children. The average diagnostic odyssey can last from anywhere from five to seven years. You can imagine the anguish of parents watching their children suffer, watching them endure one test after another. The toll this takes on the family, emotionally and financially, is a great travesty in an age where comprehensive genetic screening is available and affordable. Whole Genome Sequencing will alleviate an enormous part of a huge burden these families carry.

In August 2019, a bipartisan group of U.S. senators introduced legislation that would help thousands of children with rare diseases get a diagnosis and proper treatment faster through whole genome sequencing. This bill has needed a great deal of shepherding, which Genetic Alliance has provided throughout the year. We have educated health legislative aides in more than 100 offices.

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

In 2020, we supported families to have more guidance, more support, better health.

266

individuals participated in online training courses

725

Individuals attended online learning sessions

3.8 MILLION

Individuals attended online learning sessions

710,688

Individuals participated in online training courses

COMMITMENT

We are health communicators committed to providing actionable, relatable science-based information. During this year of changing health information and uncertainty, we continued our work of educating and supporting new and expecting families. To alleviate any anxiety or confusion related to COVID-19, we created a variety of timely COVID-19 materials and connected with partner organizations to meet families where they are.

OUR COVID-19 RESOURCES

Website articles that covered topics from the prenatal to postpartum period

Webinar to support family needs during the pandemic

Infographics to share important COVID-19 information

Educational module about COVID-19 and newborn screeing

We are maternal health advocates committed to fighting for equitable healthcare and accessible health education. Rooted in social justice, our programs aim to provide access to important health information throughout the prenatal and early motherhood years.

We attended a local community health fair for low-income families and connected with over 50 moms to share the benefits of healthy eating, including eating seafood during pregnancy.

We developed a free, online newborn screening module so families have access to critical information about the test that every baby gets with 266 course sign-ups to date.

We celebrated Black History Month to raise awareness for the unique history, experiences, and contributions of Black women in America in the reproductive and maternal health space generating

18,042

social media impressions.

COLLABORATION

We are leaders at the intersection of family health and genetics. With our expertise in elevating the family experience in the rare disease and newborn screening communities, we collaborate with advocacy and professional organizations across the nation.

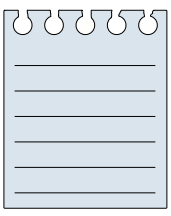
We lead the content development on **2 federally-funded programs** focused on newborn screening and family support.

We are community partners for **2 national initiatives** focused on improving health services and outcomes for women and children, specifically through reducing maternal mortality and improving telehealth services.

We leverage and support the work of organizations to ensure that new and expecting families are getting actionable, digestible information to feel empowered in their healthcare decisions

We have a growing partnership program with **12 partner organizations** to expand our reach to new and expecting families.

We **co-hosted a 5-part training series** with the EveryLife Foundation to educate and engage newborn screening stakeholders.



We **submitted public comments** to support updated guidelines on dietary needs of women and children.

Genetic Alliance

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