**Request for Information (RFI):**
**Input on Clinical Whole Genome Sequencing for Low and Middle Income Communities**

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**Notice Number: IGH-RFI-001**

**Release Date: 3/1/2022**
**Response Date: 4/1/2022**
**Contact Email: info@ihopegenetichealth.org**
**Issued By: Genetic Alliance**

## iHope Genetic Health

### Background

iHope™ Genetic Health (iGH) will eliminate the barriers to genomic insights for tens of thousands of individuals across the globe each year by addressing access to high-quality clinical whole genome sequencing (cWGS) as well as education and support. Leveraging contributions of genomic sequencing technology from Illumina and donations from philanthropists and technology and logistics partners, iGH will provide awards of reagents and, where necessary, sequencers and software to enable genomic testing across the globe. iGH will provide cWGS and patient support services for undiagnosed individuals in low and middle income countries[1] (LMICs) and low-to-moderate income[2] (LMI) households across the globe and in the United States (US) respectively. Over time iGH will develop in-country, sustainable solutions for communities with little to no access to genomic care.

[1] The World Bank Group defines low and middle income (LMIC) countries as low-income economies ($1,005 or less GNI per capita) or as lower-middle-income economies ($1,006 to $3,955 GNI per capita).
[2] Low-to-Moderate Income (LMI) means any census tract (or equivalent geographic area defined by the Bureau of the Census) in which at least 50% of households have an income less than 60 percent of the Area Median Gross Income (AMGI), or which has a poverty rate of at least 25%.

iGH is unique in several ways. We will:

- Support laboratories to provide cWGS for undiagnosed individuals in low and middle income countries (LMICs) and low-to-moderate income households (LMI)
- Facilitate patient support services
- Facilitate access to therapies
- Match patients to support when available
- Facilitate patient control of their data
- Invite participants to research studies
- Ensure no cost access to Institutional Review Board and participant approved research data for any academic or nonprofit researcher, ensure low-cost access to biopharmaceutical companies
- Disseminate insights, findings, and results quickly
- Build capacity in-country, in areas and regions that need support

Expected outcomes:

- Sequence tens of thousands of undiagnosed individuals
- Create the world's largest patient-owned genomic data resource
- Eliminate barriers to genomic testing to open up tremendous opportunities to accelerate the delivery of precision care, generate new insights and drive health equality globally
- Open new pathways to develop therapies with more precompetitive systems and sharing
- Create more robust and facile patient networks

**Information Requested**

iGH is requesting information from stakeholders to formulate a Request for Proposals (RFP) from laboratories and their associated clinical sites, to serve the needs of undiagnosed patients.

We request information from all stakeholders, especially:

- Patients and families
- Genomic laboratories
- Hospitals and/or clinics that care for patients with genetic disorders

As well as:

- Advocacy organizations
- Individual clinicians
- Healthcare administrators
- Genetic disease researchers
- Governmental agencies
- Policy makers

**Genetic Alliance seeks comments on any or all of, but not limited to, the following topics:**

Patients and families:

- What barriers to accessing genetic testing have you faced? (You may wish to consider testing availability, cost, and clinician awareness as part of your answer.)
- What support is desired after the return of cWGS results? What is currently available?
- Do you wish to have access to your genetic data? What education would you need to feel comfortable managing access to this data if requested by researchers?

Genomic laboratories:

- What tests do you currently offer? If you offer clinical whole genome sequencing, what is your test definition? If you are not currently offering clinical whole genome sequencing, what is your principal barrier to doing so?
- What are your current testing dynamics – including number of cases per year, average turnaround time from sample accession to report delivery and number of variants delivered per case? What support would be needed to expand your annual volume and reduce your turnaround time?
- How is your test interpretation handled? Do you need additional software tools and/or support?
- How is primary data management handled? iGH will enable patients to have access to their own data – what support would you need to transfer both phenotypic and genotypic information to a patient-permissioned data platform?
- iGH is predicated on partnerships between laboratories and clinical sites. Do you have established relationships which will enable mid to high-volume testing of LMIC and LMI households? What is the minimum volume you could guarantee through such relationships?
- How can participating laboratories standardize their sequencing and reporting to ensure uniform high quality and comparability of results?

Hospitals and/or clinics:

- What are the primary barriers to providing your patients access to cWGS? What are the ideal solutions to enable this testing?
- How many patients per year do you care for with genetic disorders? Do you require support with appropriate patient identification?
- What care and support services are available to patients after return of genetic testing results? Do you need additional medical genetic or genetic counseling education/support at your site?
- Do you have existing experience with cWGS or similar tests? Do you have established relationships with cWGS testing providers?
- Do you have established relationships and/or experience working with pharmaceutical companies to provide therapies on a compassionate-use basis and/or engaging in clinical trials?
- How would you identify if patients appropriate for cWGS testing are from LMIC or LMI households?

**Additional stakeholder questions:**

Clinicians:

- What are the primary barriers to having your patients receive cWGS testing? (You may wish to consider availability, reimbursement, awareness and turnaround time as part of your answer.)
- For patients who would receive cWGS through iGH, what additional support services would be required for you/the clinicians and the patients?
- How many patients do you see annually that you feel could benefit from cWGS testing? What are their primary indications for testing?

Policy makers:

- What policies would you like to see implemented to support cWGS and patient support services?
- What policies must be established or improved to support cWGS for undiagnosed patients in need?
- What policy barriers must be overcome to routinely provide cWGS to patients in your region?
- What evidence is needed to assess the impact of cWGS on patient care?

Researchers:

- What data should be collected to research newly identified or as-yet-to-be-identified gene-disease associations and/or develop therapeutic biomarkers?
- What tools would help you analyze the genomic, clinical, and patient reported outcomes data? Note: US probands will also have electronic health records data associated with their account.
- How should impact on patient care be evaluated?

Please add anything else you would like us to know.

**RFI Response Guidelines**

- Responses must be provided as a Microsoft Word (.docx) or PDF attachment and be no more than 5 pages in length (using 12-point Arial-, Calibri-, or a Times New Roman-type font) and 1-inch margins. Only electronic responses will be accepted.
- Responses must be provided as attachments to an email and sent to info@ihopegenetichealth.org no later than 5 PM PT on Friday, April 1, 2022. (It is recommended that attachments with file sizes exceeding 25 MB be compressed to ensure message delivery.)
- If proprietary information is included in your response, please indicate that.
- Respondents may answer as many or as few questions as they wish. Respondents may also share other information.

- This RFI is for information and planning purposes only and shall not be construed as a solicitation, grant, or cooperative agreement or as an obligation.

*Special Note: If you are a family member in need of services, please register here to be contacted in the future. We will not be responding immediately, so if you do have an urgent medical issue, contact your healthcare provider in addition to registering with us. Do not email us with sensitive health information, we want to protect your privacy.*

**Information about the Issuing Organization**

iHope™ Genetic Health is a program of Genetic Alliance. Genetic Alliance is a 501 (c) (3) that engages individuals, families, and communities to transform health. For more than three decades, we have empowered advocacy organizations and other communities to collaborate with clinical, policy, service, research, and other health institutions at the international, national, state, and local levels. Genetic Alliance has earned the trust of diverse stakeholders. As such, our convening of, and activities with, multidisciplinary entities result in novel, relevant, and effective solutions. Genetic Alliance builds and liberates capacity in networks, thus enabling each entity to leverage the others' learnings, resources, and tools.

Our work has been strongly influenced and deeply embedded in activism - we have roots in radical advocacy, the early AIDs movement, and justice activism. We have a deep history in genetics, having been founded before the Human Genome Project was conceived. We have participated in and led hundreds of projects and programs in genetics and genomics, including as co-founders of the National Academies of Medicine Board on Health Science Policy and Roundtable on Genomics and Precision Health, Global Alliance for Genomics and Health, International Rare Disease Research Consortium, the Personalized Medicine Coalition, the Coalition for 21st Century Medicine, Obama's Precision Medicine Initiative, and the Cancer Moonshot. We led the coalition that enabled the Genetic Information Nondiscrimination Act to be enacted as US federal law. In all of this, we have always held dearly the health of all, particularly those in underserved and resource-limited communities, as our guiding light.