Genomics: going global

GLOBAL ALLIANCE FOR GENOMICS AND HEALTH

ANALYSIS: EXCLUSIVE

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To present a truly international perspective on the Global Alliance for Genomics and Health, International Innovation has interviewed three of its leaders located in Europe, North America and Asia. Here, the aims, challenges and advancements affecting the field of genomic medicine and data sharing worldwide are revealed.

**PETER GOODHAND**

Executive Director, based in North America

What inspired you to become involved with the Global Alliance?

Having worked in the private, public and philanthropic sectors – and with experience in helping move research into products, practice and policy – I wanted to do something new that brought the three branches together to help accelerate health research into better human health. Based in North America, I am Executive Director, which entails using my expertise in pulling people and different sectors together to create alignment.

Can you elaborate on the Alliance’s core goals?

The essence of the Global Alliance is to accelerate progress in medicine and healthcare by enabling the responsible sharing of genomic data. Many existing initiatives are associated with a distinct disease state such as cancer or rare diseases, a specific region or country, or a particular sector. However, we believe that in order to learn from genomic data, it is imperative to do so across disease states, countries and sectors. We look at this network as an ecosystem and focus on bringing together a large community and finding ways to enable that community to collaborate.

Are there specific challenges commonly encountered with building international collaborations for advancing research, advocacy and technology in the field of genomic medicine?

There is a well-established history of data sharing on genomic research – for example, the Human Genome Project, 1,000 Genome Project and, more recently, the International Cancer Genome Consortium. As we start to get closer to clinical knowledge and applications specifically for medicine, there are many other factors that come into play, including issues regarding health systems, national restrictions on data storage and protection of privacy. We need to explore different approaches and use federated data and metadata to ensure we are respecting boundaries and protecting privacy while simultaneously learning from these data. Although this is not impossible, the more you enrich these data with clinical information, the bigger the challenge becomes. The Alliance is looking at ways to address these barriers and create solutions for them – there are over 25 task teams working on specific aspects of this.

Can you provide exciting examples of any recent innovations or research discoveries in genomics and health that the Global Alliance has helped facilitate?

There are three projects we are currently focusing on. The Beacon Project – which aims to test the willingness of international sites to share genetic data in the simplest of all technical contexts – is a great demonstration of data sharing. We have launched over 15 Beacons on data sets that allow people to ask a basic question in an open manner.

The second project we are supporting, which has a more research and clinical application, is the Matchmaker Exchange. This project is looking at how rare disease databases match phenotype and genotype and is subsequently finding better ways to link them together so that rather than having to individually search multiple databases, you would be able to search one database and have access to many.

The Alliance’s third project is the BRCA Challenge, which is bringing together many clinical experts from around the world that are knowledgeable in the BRCA1 and BRCA2 genes, which are linked to breast and other cancers. This project aims to find more effective ways of building larger and higher quality sets of data so that people can create a more definitive understanding of breast cancer.

Broadly speaking, how is the study of genomics improving human health outcomes?

Genomics is contributing to the identification of specific diagnoses of illnesses such as rare diseases, and is therefore helping to end the diagnostic odyssey that individuals affected by rare diseases have gone through for many years. Furthermore, in diseases like cancer we are seeing more drug targeting and better prediction of drug response, and there is also a tremendous opportunity to design effective precision (personalised) medicine from genomic information.
Can you introduce yourself and your reason for joining the Global Alliance?

I am a retired medical geneticist and have spent my career researching inherited diseases and seeing people and families with such conditions in the clinic. I was Head of the Department of Medical Genetics at the University of Amsterdam, at Guys Hospital in the University of London, and then at the University of Cambridge, UK. I was also interested in science policy and funding, and was a Council Member of the UK Medical Research Council, and a Governor of the Wellcome Trust. I have strongly supported the principle of early sharing of scientific data because it makes for better research, more efficient use of scarce scientific resources and is ethically sound in giving best returns to those who volunteer their bodies, tissues and information for scientific research. My background and beliefs made me an immediate supporter of the Global Alliance.

How important is it for the Alliance to continue harnessing worldwide collaborations?

The international aspect to the mission is critical, because health problems and their genomic connotations are international. Furthermore, open collaboration requires building trust between scientists and others such as science funders and advocacy groups, so that the use of data will be beneficial and the shared data will not be abused. Such trust is more likely to emerge if the effort crosses national boundaries.

In what ways is genomics leading to developments in precision medicine?

Medical practice has always lumped people with similar symptoms into groups such as diabetes, cancer or hypertension, but we now know that these conditions are often heterogeneous at a cellular or biochemical level – although two people may both, for example, have high blood pressure, the causes of this may be quite diverse. These different causes may need different treatments. Furthermore, people often react somewhat differently to the same drug, because we all vary from one another in details of our chemistry just as we vary in the shapes of our faces.

Genomics helps to differentiate these subtleties, breaking people and their diseases down into much smaller groups, so that they are more likely to get a treatment that will work for them and their particular illness. Using genetic information to guide treatment for the best result is developing in many different areas of medicine: from certain types of lung cancer, to the discovery of new treatments for a small proportion of cases of previously untreatable conditions like cystic fibrosis and muscular dystrophy.

How is the era of ‘big data’ affecting the field?

Technical advances mean that sequencing the whole genome of a patient is becoming practically feasible; however, this generates a huge amount of data – there are over 3 billion DNA bases in one genome. It is becoming clear that the complexity of interactions between people’s genomes and aspects of their health means it will be necessary to study hundreds of thousands, or even eventually millions, of individuals. These huge amounts of data are hard to analyse, but could be enormously rewarding.

What opportunities have advances in technology provided for genomic medicine?

Without modern developments in IT, we would not be able to even think of capturing, storing and analysing such large quantities of data, let alone sharing them with colleagues to speed up making sense of it all.

MARTIN BOBROW
Vice-Chair of Steering Committee, based in Europe

WORKING TOGETHER

The Global Alliance has formed four working groups to facilitate the sharing of genomic data for advancing human health and medicine

Clinical Working Group

Strives to develop effective and scalable ways to share clinical and genomic data. The group looks across all disease states and all parts of the globe to find existing or emerging initiatives with which the Global Alliance can connect.

Data Working Group

Concerned with the effective storage, representation and analysis of genomic data. Part of its work includes collaborating with academic and industry leaders to enable interoperability.

Regulatory and Ethics Working Group

Focuses on ethical, legal and social implications of the Global Alliance. This includes coordinating international policies and standards necessary for the formation of efficient and comprehensive data sharing projects.

Security Working Group

Examines how the Alliance can ensure the protection of data and people’s privacy by investigating data security, addressing user access control and audit functions, and setting appropriate standards.
KAZUTO KATO
Member of Steering Committee and co-Chair of Regulatory and Ethics Working Group, based in Asia

How has your previous experience prepared you for your role within the Alliance?

I trained in the field of developmental biology and spent four years as a postdoctoral fellow in Sir John Gurdon’s Laboratory at the University of Cambridge, UK. I have worked on ethical, legal and social issues as a member of large-scale international genomics projects such as the International HapMap Project and International Cancer Genome Consortium. I am working for the Alliance as an expert on ethics and regulation, while also providing perspectives from non-Western regions, most importantly, Asia.

Does the Global Alliance face any region-specific challenges?

Researchers in Western countries are accustomed to working collaboratively on new challenges by having an ambitious ideal. The notion of facilitating the sharing of genomic and health-related data at the global scale is one of the best examples of this. Contrastingly, my fellow researchers in Japan are often cautious about starting a project that has inevitable hurdles. While researchers in Japan are often adventurous as individuals—as can be demonstrated in the case of Dr Shinya Yamanaka, who won a Nobel Prize for his discovery of induced pluripotent stem cells—developing a large ambitious project is often difficult, and people tend to avoid risks. Therefore, one current challenge is convincing Asian researchers about the value of working together with colleagues from around the world on data sharing. Another challenge is that geographical and linguistic barriers often hinder effective collaboration.

Can you outline the aims and activities of the Ethical, Legal and Social Implications (ELSI) Research Unit that you are currently directing?

Our unit is established within a large genomics project, Genome Science, funded by the Japanese Government’s Ministry of Education, Culture, Sports, Science and Technology (MEXT). The research unit is held within the Department of Biomedical Ethics and Public Policy, Graduate School of Medicine, Osaka University, Japan, where I have been a professor since 2012. Genome Science’s aim is to support genomics researchers in Japan by conducting large-scale genomic sequencing and bioinformatics analysis – at present, approximately 40 researchers across Japan are involved in the project. ELSI’s role is to solve ethical problems by closely working with medical genomics researchers and analysing ethical issues more deeply and carefully. Our unit also conducts investigations into the ELSI of genomics and regularly publishes articles in the area – we also recently began to collaborate with researchers at the Centre for Health, Law and Emerging Technologies at the University of Oxford, UK.

What are the ethical issues relating to genomics and stem cell research?

There are many ethical concerns in genomics research. One issue is finding an effective mechanism to share genomic and clinical data from patients while protecting it. We also need to ensure that our data-sharing activities are consistent with the regulations of relevant countries and regions. Another matter is identifying ways to carry out the process of informed consent so patients and citizens who participate in research fully understand that their data are being shared. The Global Alliance has a responsibility to explain the purpose and procedures of data sharing in an easily accessible language. Last but not least, we need to make sure that the benefit of data sharing is not just given to those in rich countries, but also to those in developing countries.

How is the Alliance seeking to address these problems?

We started the Global Alliance’s Regulatory and Ethics Working Group (REWG), which has 13 executive members and more than 50 experts in ethics, law and data protection from all around the world. REWG’s first product was the Framework for Responsible Sharing of Genomic and Health-Related Data. This Framework is founded on Article 27 of the 1948 Universal Declaration of Human Rights and guarantees the right of every individual in the world “to share in scientific advancements and its benefits,” including to freely engage in responsible scientific enquiry while ensuring “the protection of the moral and material interests resulting from any scientific production of which the person is the author”.

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