

Translating translational medicine into global health equity: What is needed?



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ABSTRACT

While genomics, and other omics, research is rapidly advancing in the US and Europe, progress has been slower in less resourced countries. The imbalance has given rise to concern about whether the benefits of these advances, namely new and better tests, treatments, risk identification, and prevention strategies, will be shared and available to those living in less resourced reaches of the globe. In effort to give voice to researchers, an informal survey about barriers to advancing translational medicine was administered to attendees of the 11th Asia Pacific Conference on Human Genetics, 2015, Hanoi. The overall goal of the survey was to identify unmet needs and rank their importance. Most attendees completed the survey. Not surprisingly funding is indicated as a major need. Respondents reported that lack of bioinformatics and computational tools, trained data scientists and access to datasets is creating a significant lag behind better resourced regions. Results are intended to inform efforts to create a regional consensus statement of need. Such a regional statement could help funding organizations and policy makers seeking to promote global genomics benefit sharing.

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Paradigm shifts are occurring at all stages along the path to translational medicine, complicating the process while also creating exciting opportunities. Translational genomics is no longer simply a nebulous bridge from bench research to bedside care with research only conducted in a top down fashion involving studies that are done in secrecy, with little reporting back to research participants. Patients are contributing more to, as well as demanding more from, their clinical encounters and genetic/genomic data is more easily flowing between greater and less resourced institutions. The opportunity to develop high impact solutions that are focused on greater relevance to human health is also greater than ever before. At the same time, there is no consensus as to what are truly 'best practices' for handling issues of data management/integration, ethics, patient empowerment roles, provider knowledge, etc. A shared understanding of how best to proceed is key to establishing viable translational and precision medicine.

Genomics, and other omics research is rapidly advancing in the US and Europe but such progress has been slower in less resourced countries (Tekola-Ayele and Rotimi, 2015). The imbalance has given rise to concern about whether the benefits of these advances, new and better tests, treatments, risk identification, and prevention strategies will be shared and available to those living in less resourced reaches of the globe (Dickenson, 2004; Chadwick and Wilso, 2004). An interest in identifying challenges and opportunities for improvement in low resourced countries is not new. Several international groupings have undertaken to understand needs and impediments for implementing translational medicine. Last year the U.S. National Human Genome

Research Institute and the U.S. National Academy of Medicine convened 90 leaders of genomic medicine from the U.S. and 25 other countries spanning 5 continents to identify regional capabilities and the current state of implementation and opportunities for collaboration, in efforts to identify specific differences between the 'haves and have nots'. An informal poll of attendees revealed that the majority of these countries have specialized clinical genomic capabilities (cancer treatment, rare disease diagnosis and microbial pathogen identification) but lack capabilities for newborn genomic sequencing, or RNA, metabolomics or proteomic profiling. Of note, is that poll results were similar to those in a 2012 survey (Manolio et al., 2013).

Ascertaining global challenges and opportunities for improvements typically involves polling experts who can afford to travel to such meetings. Non-expert researchers and clinicians are less often polled. The opportunity to do just this presented itself last fall at the 11th Asia Pacific Conference on Human Genetics (APCHG 2015) last fall in Hanoi, Vietnam, which was themed Genetics and Genomics: The Path to Translational Medicine. The meeting, which occurs every other year, offered the chance to learn about research and clinical impediments as well as needs for achieving successful strategies from researchers and clinicians in small as well as larger institutions in Vietnam and surrounding countries.

In effort to give voice to researcher needs, as well as clinician needs, a short informal survey about barriers to advancing translational medicine was administered to attendees. The overall goal of the survey was to identify regional unmet needs and rank their importance. The purpose was to use results to begin developing a regional consensus statement that could help funding organizations and policy makers seeking to promote global genomics benefit sharing.

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Attendees were asked the following five questions:

1. What do you view as the major regional/local impediments to advancing translational medicine? (open question)
2. What are the top 3 needs? Choose only 3 of the following and rank your choice 1st, 2nd and 3rd: Access to data, Funding, Trained data scientists, patient engagement, seamless integration of genomic and clinical data, or knowledgeable clinicians.
3. What are the 3 greatest research needs? Choose only 3 of the following and rank your choices 1st, 2nd and 3rd: access to datasets, bioinformatics/computational tools, data storage and management, access to clinical data, need for standardized data, ethical issues (consent and privacy), funding, data scientists
4. What are the 3 greatest clinical delivery needs? Choose only 3 of the following and rank your choices 1st, 2nd and 3rd: genetic resources, clinical decision support, provider education, ability to identify and refer patients, or patient engagement.
5. What is important to emphasize in a consensus statement? (open question)

60 attendees completed the survey. Responses were as follows.

In response to question 1, the vast majority of respondents reported that resource limitations and lack of government support was the biggest barrier, followed by lack of knowledge and training, poor collaboration, lack of acceptance of genomic health as a priority and data sharing. Normative beliefs, lack of interest, patient engagement, regulation and language barriers were also reported by a few. In regard to question 2, the greatest need reported was funding (28), followed by trained data scientists and access to data, with knowledgeable clinicians, a close third (Fig. 1). While not surprising that funding would be identified as the greatest need, it is perhaps noteworthy that the need for trained data scientists and data access was deemed the next greatest need. Beyond reflecting that the need for data is universal, this result suggests a specific gap that with the help of experts in the West and well-funded training programs might well be readily filled.

In response to question 3, attendees reported that the greatest unmet research need is funding (31), with bioinformatics/computational tools and access to clinical data tied for second greatest need, and access to datasets the third greatest need (Fig. 2). Access to clinical data, the need for standardized data, and ethical issues were designated as greater needs than data scientists. The low ranking of the need for data scientists here conflicts with its higher rank as an overall need in question 1. A number of different speculative explanations are possible for this discrepancy, such as the need to access and be able to work with data is greater than the need for more data scientists, or the respondents presumed that data scientists were subsumed in the need for data access or respondents simply didn't understand the question correctly.

In response to question 4, attendees reported that clinical decision support was the greatest unmet clinical delivery need, followed by provider education and access to genetic resources (Fig. 3). This result

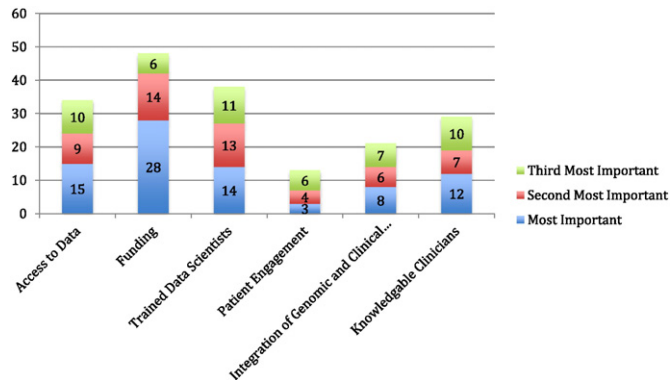


Fig. 1. What top three needs does your institution require to advance translational medicine?

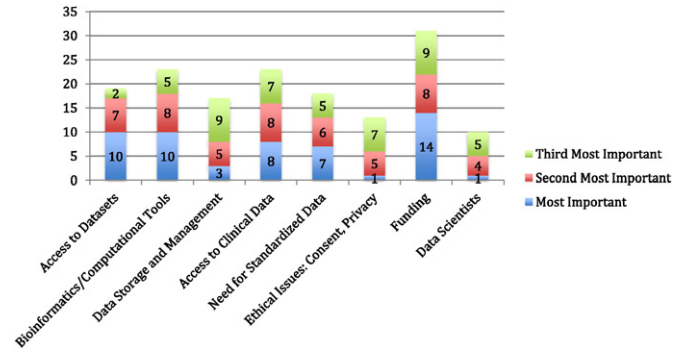


Fig. 2. What are the three greatest needs in the area of research?

suggests a defined need for knowing how to integrate new genetic/genomic knowledge into clinical delivery. While the question did not distinguish between informatics/machine based or actual person-to-person expert knowledge support, responses suggest that the benefits of genetic/genomic advances will not be optimally realized unless known knowledge about how to apply advancements in a clinical context is obtained. This suggests both a need for translational research as well as applied research designed to identify how best to integrate new clinical knowledge.

Only 24 of 60 respondents answered question 5. Five respondents indicated that a consensus statement should address important regional differences amongst member countries. Four reported that de-identified data should be shared. Three respondents reported that government support and funding needs should be specified in such a document. Two attendees reported that genomic education for clinicians and patients was important to include in a consensus statement on need. The remaining respondents reported a broad range of other vital factors; specifically, the need to convince government that genomics is important and that funding a genomics infrastructure is imperative, the need for funding to train a genomics workforce, the need for individual disciplines to collaborate, the need for global help with basic clinical research, the need for affordable genetic and genomic tests, the need to design laws and regulations to ensure the existence of public genomic health programs, and the need to train data scientists and the need to access both data and new knowledge.

One relatively easy and immediate solution would be to create a resource network within Asia Pacific to connect regional experts and related resources to researchers and data scientists in need. Online educational/training resources as well as the opportunity to connect with experts for specific research advice could well serve both researchers in training as well as translational researchers, and implementation initiatives through knowledge transfer, collaborative problem-solving and capacity building. Such a network can further benefit researchers

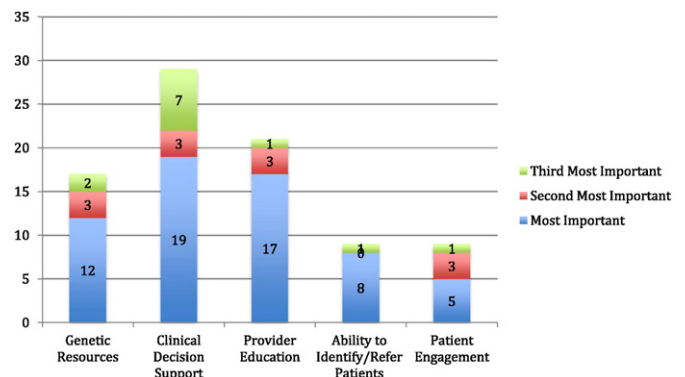


Fig. 3. What are the three greatest needs in the area of clinical delivery?

and clinicians in resource constrained settings by connecting to existing global initiatives designed to share approaches and lessons learned towards accelerating the implementation of genomic medicine worldwide, such as the Pan-Asia Pacific Genome Initiative, The Global Organization for Bioinformatics Learning, Education and Training, the Asia-Pacific Bioinformatics Network, and various Centers for Global Health around the world (Manolio et al., 2015; Ranganatha et al., 2012; Weitzel et al., 2016; Attwood, et al., 2015; Daar et al., 2007).

In conclusion, this preliminary informal survey indicates that a broad range of unmet infrastructure needs exist in the region, not the least of which is funding and capacity building. Further research is needed to better understand unmet needs and to engineer strategic solutions. Creating an online regional network of experts and resources that is connected to specific local areas of needs could help jumpstart progress. Continued needs assessments and strategic remedies can help ensuring that low resourced countries are successful in research and clinical integration and thus able to realize the benefits of genetic/genomic advances.

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