



## Unsung heroes: Genomic successes in the developing world



Recounting progress in genomics typically involves detailing a series of milestones from the inception of the Human Genome Project to the present. Eric Lander did just that beautifully in his plenary talk opening the recent International Congress on Human Genetics 2016 in Kyoto. He traced the path from genetics to disease biology as it progressed from identifying disease genes to sequencing genomes, to mapping disease genes, to cataloging common and rare genetic variations to interpreting their clinical significance, to understanding how the genome folds into 3D maps, all of which bespeaks remarkable achievements, and innovation, in science and patient care. Not surprisingly, such regaling of success rarely if ever includes achievements by less resourced countries, because resource limitations for the most part bar contributions of the same magnitude. The challenges faced by researchers in these parts of the world are more readily known than the successes, however large or small. It is for this reason that we bring to your attention remarkable achievements occurring in such areas of the world.

In this special issue of the Journal several groups from developing countries from across the globe report their work and perspectives. In 'Pharmacogenomics for infectious diseases in sub-Saharan Africa successes & opportunities', Chaudhry and colleagues from South Africa, while addressing both the potential of pharmacogenomics as well as its failure to live up to initial expectations, highlight the fact that international consortia working on pharmacogenomic implementation guidelines have failed to come up with guidelines for the major infectious disorders affecting the developing world such as HIV/AIDS, TB, and malaria.

In the globalized world that we live in, international collaboration is the key to success. Limited research infrastructures as well as, poorly developed research and ethics governance mechanisms pose many challenges for both researchers and ethics committees. Roach and colleagues, in their paper 'Addressing ethical challenges in the genetics sub study of the national eye survey of Trinidad and Tobago' highlight some of these challenges while demonstrating their novel approaches to developing a culturally appropriate, multifaceted strategy to address these challenges.

In today's world the buzzword is genomic medicine. Genomic medicine is dependent on successful implementation of genome sequencing and clinical bioinformatics. At the recent International Congress of Human Genetics 2016 several plenary speakers declared, "Doctors are good, but exomes are better". Genomic data driven medicine can only be a reality in settings where infrastructure and manpower for genome sequencing and clinical bioinformatics is in place. Helmy and colleagues from Egypt and the UAE provide their perspectives on the challenges faced by the developing world in making genomic medicine a reality due to limited infrastructure for genome sequencing and bioinformatics, and suggest solutions to overcome them, in their article 'Limited resources of genomic tools in developing countries: challenges and solutions'.

Despite significant resource limitations, there are oases of excellence in genomic research in the developing world. These three papers illustrate the breadth of research and depth of success; specifically in forensic genomics, clinical informatics and agrigenomics/bioinformatics. Mohamed and Salama present evidence of the affection of tau normal pattern and pathological aggregates of tau in case of brain hypoxia and suggest therefore that tau protein may be a biomarker for asphyxia in 'Tau protein as a biomarker for asphyxia: A possible forensic tool.' Mulder and colleagues from both the Sickle Cell Disease (SCD) community and H3ABioNet report on their recent SCD Ontology workshop that produced the first comprehensive SCD ontology, in 'Proceedings of a sickle cell disease ontology workshop- towards the first comprehensive ontology for sickle cell disease'. The ontology permits improved data sharing, meta-analyses and further development and curation of databases and clinical informatics. As such it can serve as a model for other disease communities. Tospovirus is severe plant pathogen that damages food crops worldwide. In 'SeeHaBITaT: a server on bioinformatics applications for tospoviruses and other species', Habeeb, presents a novel server that enables both control measures and the capacity for other computational research where none had existed.

The challenge to take research from bench to bedside is always formidable and all the more so in resourced constrained countries. In their paper 'Implementation of genomic medicine in Sri Lanka: initial experience and challenges', Sirisena and colleagues describe how they used clinical exome sequencing and gene panel testing to diagnose three complex cases leading to improved patient care. Such experiences are for most part isolated and therefore there is a need for more widespread equitable implementation of genomic medicine. The final paper by Isaacson Barash 'Translating translational medicine into global equity: what is needed?' provides insights about what is needed to achieve widespread equitable implementation of genomic medicine, based on a survey of unmet needs at last fall's Asia Pacific Society of Human Genetics meeting 2015 in Hanoi.

To be sure, the successes reported here are significant. And yet, it is still necessary to ask why Genomic Medicine is lagging behind in the less resourced parts of the world. Most developing countries around the world today have large expatriate populations living in the West or affluent countries in the Middle East, working as migrant workers. Often such trained individuals return to their native countries to bring new knowledge and skills to fill the void. Further, increasingly, there is pressure for services available in the West to be made available in developing countries. And yet despite the rapid adoption of IT and increases in global collaborations and data sharing, genomic medicine is not keeping pace outside the West.

The national health agenda of most developing countries is shaped by technical assistance given by the World Health Organization (WHO). Sadly, the WHO has been slow to embrace Genomic Medicine and the Human Genetics Program at the WHO has been poorly staffed and poorly funded. The global agenda of the WHO is shaped by

resolutions of the World Health Assembly (WHA). Action based on such resolutions received funding, and are implemented through the regional organizations of the WHO. In case of Genomic Medicine, there is no such WHA resolution. The only resolution that comes close to genetics and genomics is the resolution calling on Member States to help redress the limited focus to date on preventing and managing birth defects, especially in low- and middle-income countries. This resolution, made at the 63rd WHA in May 2010, called on Member States to prevent birth defects wherever possible, to implement screening programs, and to provide ongoing support and care to children with birth defects and their families ([http://apps.who.int/gb/ebwha/pdf\\_files/WHA63/A63\\_R17en.pdf](http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_R17en.pdf)) On the ground in most countries this translated into efforts aimed at strengthening birth defects surveillance (*World Health Organization Regional Office for Southeast Asia, 2013*). We urge the various international groups that are working on Genomics and Health to now focus their attention on lobbying the WHO for a global plan of action for implementation of genomic medicine with special focus on low- and middle-income countries. This special issue serves to demonstrate that the need, desire and capacity to implement genomic medicine in low- and middle-income countries are there. With the help of the WHO, equitable implementation of genomic medicine is more likely.

In conclusion, this special issue captures several translational genetics and genomics success stories from the developing world. The ultimate beneficiaries of this work have to be the people at large. That

would happen only in the context of a healthcare workforce that is trained to use genetic and genomic information in their professional practice. That has its own challenges which were highlighted in a previous issue of this journal by *de Abrew et al. (2014)* Imparting the necessary knowledge, skills and experience on the healthcare workforce to implement genomic medicine is one of the biggest challenges facing the world today and we plan to deal with this fully in another special issue of the journal in the future.

## References

- de Abrew, A., Dissanayake, V.H.W., Korf, B.R., 2014. Challenges in global genomics education. *Applied & Translational Genomics* 128-129.
- World Health Organization Regional Office for Southeast Asia, 2013a. Prevention and control of birth defects in South-East Asia region. Strategic Framework (2013-2017). World Health Organization, p. 48 ([http://www.searo.who.int/entity/child\\_adolescent/documents/sea\\_cah\\_12/en/](http://www.searo.who.int/entity/child_adolescent/documents/sea_cah_12/en/)).

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