Focusing attention on ancestral diversity within genomics research: a potential means for promoting equity in the provision of genomics based healthcare services in developing countries

Nirmala D. Sirisena¹ · Vajira H. W. Dissanayake¹

Received: 20 February 2017 / Accepted: 8 June 2017 / Published online: 11 July 2017
© Springer-Verlag GmbH Germany 2017

Abstract Although we are well into the second decade after the completion of the International Human Genome Project, genomic research has failed to fully represent the diverse ancestry of global populations. The resultant healthcare challenges faced by populations underrepresented in genomic research needs to be tackled by the global scientific community. In this paper, we address several major factors which have contributed to the existing health disparity and put forward a combination of scientific and political interventions needed to bring about a change that will ensure all global populations benefit equally from the advances made in genomic medicine and research.

Keywords Ancestry · Diversity · Ethnic · Genomics · Research · Healthcare

Introduction

The successful completion of the International Human Genome Project was a significant breakthrough in genomic research that transformed our understanding of how individual variance and combinations of genetic risk factors can

modulate an individual's predisposition to disease as well as response to treatment (Stapleton et al. 2013). Although we are well into the second decade after its completion, the compendium of genomic research has failed to fully reflect the ancestral diversity found in the global populations. As our understanding of the complex interaction between genes, environment, and disease advances, genomic information will play a more significant role in healthcare in the future. Genomics and other 'omics' disciplines are valuable tools that can be effectively applied to many fields including food, drug, and health development (Helmy et al. 2016). However, despite these advances, the current lack of ancestral diversity within genomic research creates the risk of aggravating existing health inequities in ancestrally underrepresented populations. This poses a scientific and health equity challenge which needs to be addressed by the global scientific community. Thus, a combination of clinical, genetic, statistical, ethical, and political interventions is needed to bring about a change in order to ensure that genomic medicine benefits all populations equally. Appropriate policies will need to be implemented to ensure that the benefits of genomic research are effectively applied to health improvement in ancestrally underrepresented populations, especially in the developing world.

The appropriate assessments of race and ethnicity are crucial to health services research. Decisions regarding allocation of resources by international and national agencies and public and private organizations aimed at developing interventions to improve health outcomes are often based on data relating to racial and ethnic classifications (Ford and Kelly 2005). Even though much emphasis has been made on the impact of genomics on individual healthcare ("personalized medicine"), much less attention has been placed on its impact on public health at the population level, especially in the developing world. The major emphasis so far has been on institutionalized research geared towards genomics "discovery," rather than on

This article is part of the Topical Collection on *Inclusion of Diverse Populations In Genomics Research and Health Services: A Scientific and Health Equity Imperative*

✉ Nirmala D. Sirisena
nirmala@anat.cmb.ac.lk

Vajira H. W. Dissanayake
vajira@anat.cmb.ac.lk

¹ Human Genetics Unit, Faculty of Medicine, University of Colombo, Kynsey Road, Colombo 8, Sri Lanka

how such discoveries could be integrated into practice to evaluate its health impact among the global population. The fact that analyzing the vast spectrum of genomic and phenotypic variations in diverse human populations will ultimately be beneficial in developing effective interventions for improved personalized care needs to be emphasized (Pang 2013).

Situation analysis of the factors contributing to the existing disparity

A major drawback sets in when diverse populations are not adequately represented in genomic sequencing studies. It limits the ability to differentiate normal from pathologic variants, reduces the generalizability of the results, and may ultimately advance science for some but not for all, thus exacerbating health disparities for minority populations (Cohn et al. 2015). Accurate interpretation of whole genome and whole exome sequencing data relies heavily on establishing the incidence of variants in populations overall and associating them with disease development or therapeutic responses. Existing data sets may fail to comprehensively ascertain the pathogenic and benign variants that are present in diverse populations, which will in turn affect variant selection algorithms and pathogenicity assessments. In fact, databases of clinically relevant variants are known to have entries with misattributed pathogenicity and this limitation needs to be considered before they can be relied upon for screening (Adams et al. 2016). To overcome these deficiencies, a broad sampling of different ancestral populations is necessary since sequencing data from diverse populations could be used to evaluate novel variants and re-evaluate the significance of known variants.

A recent paper on hypertrophic cardiomyopathy (HCM) highlights the importance of interpreting genetic test results against diverse control populations so that genetic variations that are benign and common in one racial or ethnic group do not get misclassified as being pathogenic (Manrai et al. 2016). Writing in *The New England Journal of Medicine*, Harvard University investigators reported that some variants originally identified as causing HCM by other researchers have since been determined benign. Using publicly accessible exome data, they identified that variants which had previously been considered causal in HCM are actually overrepresented in the general population. They identified shortcomings in the research methodology of the original study, especially the lack of inclusion of African-American patients in control cohorts, as being responsible for the initial faulty pathogenicity calls. This case highlights the importance of including more genetically diverse populations in study and control groups in order to provide more accurate and comprehensive information in disease association studies. It also reflects the need for systematic re-evaluations of prior findings. The misclassification of

benign variants as pathogenic shows the need for sequencing the genomes of diverse populations, both in the healthy controls and the tested patient population. These results also expand on current guidelines, which recommend the use of ancestry-matched controls for variant interpretation. As additional populations of different ancestry backgrounds are sequenced, one could expect variant reclassifications to increase, particularly for ancestry groups that have historically been less well studied (Manrai et al. 2016). In the case of novel variants, inclusion of sequencing data from diverse ancestry groups will help to clarify their pathogenicity, as well as to reassess the clinical significance of known variants.

The establishment of genetic research and genomic services in developing countries is oftentimes hindered because the medical profession in such countries is unable to provide those services due to lack of adequately trained and qualified medical, scientific, and bioinformatics personnel and lack of investment by governments on appropriate infrastructures, especially laboratory infrastructure to enable relevant genetic diagnostics and research to be done locally (Sirisena et al. 2016). Despite the rapid adoption of information technology and improved global collaborations and data sharing, the implementation of genomic medicine and research in most developing countries is happening at an alarmingly slow pace (Dissanayake and Barash 2016). Genomic data-driven medicine and research can only be a reality in settings where infrastructure and manpower for genome sequencing and clinical bioinformatics are in place.

Limited research infrastructures and poorly developed research and ethics governance mechanisms have posed as challenges for both researchers and ethics committees, especially in the developing world. Continued needs assessments and strategic action plans can help ensure that low-resourced countries are successful in research and clinical integration, and thus able to realize the benefits of genomic advances. In the current globalized world, international collaboration is the way forward. Creating an online regional network of experts and resources that is connected to specific local areas of needs could help set the stage to bring about the much needed change. A special issue on “Genomic successes in the developing world” published in the *Applied and Translational Genomics* journal in 2016 serves to demonstrate that the need, desire, and capacity to implement genomic medicine in low- and middle-income countries (LMIC) exist (Dissanayake and Barash 2016).

In addition, the paper “Translating translational medicine into global equity: what is needed?” provides insights into what is needed to achieve widespread equitable implementation of genomic medicine, based on a survey of unmet needs at the Asia-Pacific Society of Human Genetics meeting held in 2015 in Hanoi (Isaacson Barash 2016). The lack of bioinformatics and computational tools, trained data scientists and access to datasets,

and lack of funding was stated as some of the reasons responsible for the disparity. It was observed that in some cases, research was not being conducted in places where many members of minority populations lived. Areas identified through the survey which need further attention include the following: the need to convince government authorities that genomics is important and that funding a genomics infrastructure is imperative, the need for greater funding to train a genomics workforce, the need for researchers to collaborate, the need for different labs to share their internal data, 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 (Isaacson Barash 2016).

Frameworks to build capacity within the healthcare profession for the use of genomics to address the health needs of the public for disease prediction, prevention, and treatment have been addressed previously by a couple of international network initiatives and projects such as the “CAPABILITY” consortium and the multidisciplinary international network “Genetic testing in emerging economies” (GenTEE) (Nippert 2013). These collaborative model projects have addressed major challenges faced currently by LMIC such as how to ensure the successful translation of genetic/genomic laboratory and academic research into quality-assured pathways and how to develop a service delivery infrastructure, including health workforce training, and quality guidelines and procedures that lead to equitable and affordable access to high-quality genetic/genomic testing services. These model projects provide methods for rational, epidemiological-assisted conceptual health needs assessment (HNA) approaches to identify priorities for capacity building and provide health policy makers with data for informed decision making on how to plan, introduce, and develop medical genetic services to improve both individual and population health (<https://ec.europa.eu/jrc/en/publication/eur-scientific-and-technical-research-reports/genetic-testing-emerging-economies-gentee>).

The national healthcare agenda of most low-resourced countries is driven by technical assistance given by international organizations like the World Health Organization (WHO) and the Commonwealth Secretariat. The WHO has been rather slow to fully embrace genomic medicine and its Human Genomics in Global Health Initiative (<http://www.who.int/genomics/about/en/>) has been poorly staffed and funded. The global agenda of the WHO is shaped by resolutions of the World Health Assembly (WHA). Actions based on such resolutions receive funding and are implemented through regional organizations of the WHO. In the case of genomic

medicine, there is no such WHA resolution. The only resolution on genomics is the Fifty-seventh WHA Resolution [WHA57.13, May 2004 (www.who.int/gb/ebwha/pdf_files/WHA57/A57_R13-en.pdf)] on Genomics and world health. In the Southeast Asian region of the WHO, where we are located, we do not know of any efforts by the WHO regional office to give effect to this resolution. Furthermore, a consultation on identifying regional priorities in the area of human genetics resulted in a report followed by a questionnaire-based survey of the situation analysis in member countries without any further follow-up action (http://www.who.int/genomics/publications/en/Identifying_Regional_Priorities_in_the_Area_of_Human_Genetics.pdf/SEA-RES-121/September 2003). The Sixty-third WHA Resolution [WHA63.17, Birth Defects (May 2010)] which called on member countries to help redress the limited focus on preventing and managing birth defects however has resulted in a series of workshops at the regional level and meaningful action in those countries. This resolution identified the need for the development of effective public health interventions to prevent birth defects including provision of appropriate community genetic services within the primary healthcare that can be integrated into maternal, reproductive, and child health services in LMIC (http://apps.who.int/gb/ebwha/pdf_files/WHA63-REC1/WHA63_REC1-en.pdf). This underscores the fact that even at the WHO level, there is a lack of capacity for meaningful engagement with genomics. In the case of the Commonwealth Secretariat, through its Health and Education Unit, it strives to empower national governments in developing countries to examine areas for improvement in the health sector using its public health policy tool kit (Dissanayake and Barash 2016). There is ample evidence to show that there are widespread geographic disparities in the distribution and implementation of genomic services (Moonesinghe et al. 2009; Rotimi 2012; Rotimi et al. 2013; Tan et al. 2016; Spratt et al. 2016). Therefore, there is a need to 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 LMIC. With the help of the WHO, more equitable implementation of genomic medicine could be achieved. Openings are now beginning to rise for LMIC to work with the Commonwealth Secretariat to develop a companion to the tool kit that would enable governments to examine and determine what they have to do to implement genetics and genomics in their respective countries. Similarly, opportunities are springing up to work with the WHO, first to create awareness through side meetings at their regional and global conferences, and then ultimately to work towards a WHA resolution on genetics and genomics.

Creating a way forward for increasing ancestral diversity within genomic research and more equitable implementation of genomic medicine

The ability to become proficient and implement genomic medicine offers opportunities for developing nations to advance knowledge as well as improve health risk identification, diagnoses, treatment, and prevention. In the area of genomic research, the interventions described below will help to ensure that, in the long run, ancestrally underrepresented populations are not left behind as the genomic revolution sweeps across the globe (Pang 2013). Policies will need to be put in place, for example, which will address the following:

- Increase research funding for capacity building and support education and training of researchers in genomic sciences
- Foster equitable international collaborations
- Establish ethical regulatory bodies to develop guidelines to cater for the ethical, legal and social aspects of genomics research
- Reduce inequities in research efforts and ensure ethical collaborations and equitable data access and sharing, data release, and publication
- Increase capacity of ethics review committees to review and monitor genomic research
- Build public trust and confidence in genomic research, genetic data sharing, and contributing samples and data to biobanks through community-based capacity building

Increase funds for genomic research and capacity building through establishment of centralized sequencing facilities (centers of excellence) and creating effective educational training programs

By realizing the importance of genomic research and its applications on health, drugs, and food security, governmental policies should prioritize research funding for genomics. In this regard, education of policy makers on the scientific, technical, and ethical facets of genetics/genomics in healthcare is needed (Nippert 2013; Penchaszadeh 2015). States must commit to developing well-funded and efficient systems for the evaluation and regulation of the application of genetic/genomic technologies in healthcare. Such systems should be based on criteria for the determination of their clinical validity and utility, cost, and comparing their efficacy with that of alternative technologies and interventions (Penchaszadeh 2015). In addition, academic and research institutes should take action to overcome the challenges confronting the development and utilization of genomic technology through request for increased allocation of research funds from national budgets.

Establishment of centers of excellence could be a very promising solution for providing high-quality sequencing services for several laboratories and research groups. These centers can be set up at the national or regional level, thus promoting technologies and their dissemination into resource-limited settings to achieve equity. The ultimate beneficiaries of such work would have to be the population at large. That would happen only in the context of a healthcare workforce that is adequately trained to use genetic and genomic information in their professional practice (de Abrew et al. 2014). One solution would be to create a resource network to connect regional experts and related resources to researchers and data scientists in need (Nippert 2013). Online educational/ training resources, as well as the opportunity to connect with experts for specific research advice, knowledge transfer, collaborative problem-solving, and capacity building, could well serve both researchers in training as well as translational researchers. Such networks can further benefit researchers 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. Examples include the Pan-Asia Pacific Genome Initiative, the “CAPABILITY” consortium and the GenTEE network, the Global Organization for Bioinformatics Learning, Education and Training, the Asia-Pacific Bioinformatics Network, and various Centers for Global Health around the world (Nippert 2013; Manolio et al. 2015; Isaacson Barash 2016).

Furthermore, in order to minimize the infrastructure requirements, modern genome sequencing technologies such as MinION by Oxford Nanopore Technologies (Helmy et al. 2016) can be utilized in resource-constrained settings. Such measures would minimize the requirements of adopting genome sequencing technology to an affordable device, preparation kit, stable internet connection, and a standard personal computer. Although this technology is still in its infancy, it nevertheless represents a promising solution for utilizing a wide range of genome sequencing applications with minimal laboratory and computational skills.

Education of health professionals to assess potential benefits and risks (including ethical issues) of the adequate utilization of genetic/genomic technologies in healthcare is essential. As genomic sequencing is increasingly used in research with the anticipation of informing clinical healthcare options, a new set of decisions and dilemmas face both participants and researchers. These include how healthcare providers interpret and communicate results and the ongoing need for the counseling and education of those receiving them (Cohn et al. 2015). Developing the researchers’ skills is one of the most important aspects in advancing genomic research in developing countries. This development should be geared towards both experimental and computational skills (de Abrew et al. 2014). Education and training in the basics of genomics

and bioinformatics could be introduced at the level of secondary education while more advanced training can be at the undergraduate and graduate levels (Cohn et al. 2015).

Fostering equitable international collaborations

Another key element is that of fostering international collaborative partnerships in genomics based on the principles of fair play and equity, so that researchers in low-resourced settings are treated with respect as equal partners (Parker and Kwiatkowski 2016). Such initiatives provide a framework for research that includes ongoing engagement to help researchers, clinicians, and communities better understand the process, utility, and applications of genomic science in clinical care. Collaboration between developed and developing nations in genomic research will undeniably result in a significant increase in capacity building for genomic research. Through such partnerships, the developed nations can also provide support for establishing the centers of excellence by providing funding, equipment, and training.

Establish ethical regulatory bodies to develop guidelines to cater for the ethical, legal, and social aspects of genomics research

In addition to standard ethical, legal, and societal issues associated with biomedical research in general, genomic research poses special challenges in important areas such as identifiability, informed consent, selection and role of participants, and reporting of results (Pang 2013). Little or no guidance is available for researchers and participants concerning consent for genomic research, especially in the developing world. This creates problems for participants, who may not have the confidence to give their consent, and also for researchers/clinicians who provide both clinical care and research to their patients. Current approaches to consent do not sufficiently take into account the researchers' or participants' interests and the specific issues that genomic research raises such as the thin boundary line between research and clinical care (Carrieri et al. 2016). Patient/participant data cannot be de-identified (e.g., pseudo-anonymized and anonymized) in the clinical context. It is therefore vital for researchers to ensure that their information governance is compliant with the best current practices and legal guidance. Research ethics committees must therefore be put in place to guide clinicians and researchers in conducting research within the boundaries of ethical regulatory frameworks.

Reduce inequities in research efforts and ensure ethical collaborations and equitable data access and sharing, data release, and publication

De-identified data sharing is essential for enabling and promoting genomic research in a way that will maximize the benefits to public health and society (Knoppers et al. 2011). One area in which issues of equity are of particular importance is data sharing. The sharing of genomic data across the international scientific community is an essential requirement for success in understanding of diseases affecting diverse populations. The challenge is to create a level playing field between data producers and data users that respects the legitimate interests of researchers who have generated samples and data (Parker and Kwiatkowski 2016). For the sustainability of genomic research, it is imperative that the research participants and frontline research workers should have good grounds for placing their trust in the people and institutions conducting genomic research. A recent study on attitudes to data sharing in low-resourced settings identified a great deal of public support for sharing of data but also identified trust, minimizing of harm, fairness, and reciprocity as key requirements for continued support (Parker and Kwiatkowski 2016). 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. Such initiatives can serve as a model for other disease communities as well (Mulder et al. 2016).

Re-evaluation of the patchy literature regarding disease variants depends on continued data sharing and standardization of reporting. As more and more variant annotations are shared and updated, clinical systems would be able to make this information available in near real time to physicians and genetic testing laboratories so that issues relating to misclassification of variants are minimized (Manrai et al. 2016). Additionally, strategies are required to effectively tackle the gap in access to new knowledge. Creating central academic libraries that have access to non-open access journals, establishing agreements between the universities and research centers in developing countries, and scientific publishers to grant access to scientific articles for free would help to fill this gap (Parker and Kwiatkowski 2016).

Increase capacity of ethics review committees to review and monitor genomic research

There is also the need for development of a greater understanding of genomics among members of research ethics or

scientific review committees for whom genomics may be a relatively new concept, and who raise concerns because of lack of familiarity (Parker and Kwiatkowski 2016). Ethics committees should address the return of research findings to participants/patients, for example, by introducing a policy on returning summary or aggregated research results to participants at the end of the study (Carrieri et al. 2016). In addition, the democratic and pluralistic debate on ethical and scientific application of genetics/genomics in healthcare, involving all stakeholders such as parent-patient organizations, health professionals, social movements, ethicists, policy makers, and industry should be encouraged.

Build public trust and confidence in genomic research, genetic data sharing, and contributing samples and data to biobanks through community-based capacity building

Community-based capacity building will be necessary for individuals and communities to understand and use genetic and genomic information to improve health outcomes. In public health, capacity building for research can be defined as equipping an existing community to understand and participate as partners in research (Cohn et al. 2015). Genomic biobanks underlie much of modern genomic research. These banks link genomic information with clinical data, health outcomes, and other information. They provide the keys to both basic research and facilitate translation into interventions which will ultimately lead to public health advances (Pang 2013). Achieving equitable minority representation in genomic biobanking is a challenge faced by researchers today. Some of the most commonly acknowledged controversies are those that relate to consent for the collection, storage, and use of (medical or genetic) information through biobanks. Understanding how the public perceives genetic and genomic research, what their concerns and expectations are, and their attitude towards using genetic information in health decisions is critically important for the planning and provision of genetic services and research. Several areas of public concern have been raised, including the potential misuse of genetic research to promote social discrimination, particularly in the prenatal period, and the storage and protection of genetic information, as well as the issue of access to this information by third parties (e.g., insurers, police). The above-mentioned challenges are in line with the growing literature on public attitudes towards genetic and biobank research and point to areas of concern that must be addressed in order to foster public trust and participation in research. As such, careful attention needs to be paid to achieving public trust and confidence in the conduct of genomic research and in the international collaborations essential to its success (Parker and Kwiatkowski 2016).

Essential aspects of capacity building and factors affecting decisions to participate in genomic biobank research were

identified by Cohn et al. He observed that community members sought information about genetics, genomics, and biobanking and were interested in understanding how their individual participation and the participation of their families and communities would improve health. They expressed an interest in education tailored to their specific health risks and if genomics could be used in a predictive way, to identify an increased risk for disease development. They further expressed an interest in learning more specifically about how genomics could be used to identify environment-related health risks and inform local policy.

DNA research takes place in the larger context of society. Thus, an early and deliberate attempt to address minority representation while biobanks are in the process of being developed provides individuals and communities to play a more collaborative role in genomic research. How the environment impacts community health may serve as an interesting starting point for future research. Working with communities on the areas they identify as important health risks is the foundation for sustainable community-based participatory research (Fluegge 2016).

Genomic discoveries and technologies promise numerous opportunities for improving health. A prerequisite for these potential health improvements are the healthcare consumers' understanding and acceptance of these new developments. Regular, transparent dialog between the researchers and the public could allow a greater understanding of the research process, as well as assist in the design of efficient and effective genetic health services (Etchegary et al. 2013).

Conclusions

It is imperative to understand the genomic diversity of minority ethnic groups through capacity building, training, and education and utilize this information for improving the efficiency of the healthcare delivery system provided for these diverse groups by engaging them in genomic research. Application of genetic/genomic research should not drive attention away from social determinants of health and disease such as living and working conditions, socioeconomic conditions, nutrition, and exposure to infectious and toxic agents. These factors influence most health differences at the population level and must be addressed to ensure the best possible level of health for all.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Ethical standards This article does not contain any studies with human or animal subjects performed by any of the authors.

References

- Adams MC, Evans JP, Henderson GE, Berg JS (2016) The promise and peril of genomic screening in the general population. *Genet Med* 18: 593–599. doi:[10.1038/gim.2015.136](https://doi.org/10.1038/gim.2015.136)
- Carrieri D, Bewshea C, Walker G et al (2016) Ethical issues and best practice in clinically based genomic research: Exeter Stakeholders Meeting Report. *J Med Ethics*. doi:[10.1136/medethics-2016-103530](https://doi.org/10.1136/medethics-2016-103530)
- Cohn EG, Husamudeen M, Larson EL, Williams JK (2015) Increasing participation in genomic research and biobanking through community-based capacity building. *J Genet Couns* 24:491–502. doi:[10.1007/s10897-014-9768-6](https://doi.org/10.1007/s10897-014-9768-6)
- de Abrew A, Dissanayake VHW, Korf BR (2014) Challenges in global genomics education. *Applied & translational genomics* 3:128–129. doi:[10.1016/j.atg.2014.09.015](https://doi.org/10.1016/j.atg.2014.09.015)
- Dissanayake VHW, Barash CI (2016) Unsung heroes: genomic successes in the developing world. *Applied & translational genomics* 9:1–2. doi:[10.1016/j.atg.2016.05.002](https://doi.org/10.1016/j.atg.2016.05.002)
- Etchegary H, Green J, Dicks E et al (2013) Consulting the community: public expectations and attitudes about genetics research. *Eur J Hum Genet* 21:1338–1343. doi:[10.1038/ejhg.2013.64](https://doi.org/10.1038/ejhg.2013.64)
- Fluegge K (2016) The new frontier in health services research: a behavioural paradigm guided by genetics. *J Health Serv Res Policy*. doi:[10.1177/1355819616664374](https://doi.org/10.1177/1355819616664374)
- Ford ME, Kelly PA (2005) Conceptualizing and categorizing race and ethnicity in health services research. *Health Serv Res* 40:1658–1675. doi:[10.1111/j.1475-6773.2005.00449.x](https://doi.org/10.1111/j.1475-6773.2005.00449.x)
- Helmy M, Awad M, Mosa KA (2016) Limited resources of genome sequencing in developing countries: challenges and solutions. *Applied & translational genomics* 9:15–19. doi:[10.1016/j.atg.2016.03.003](https://doi.org/10.1016/j.atg.2016.03.003)
- Isaacson Barash C (2016) Translating translational medicine into global health equity: what is needed? *Applied & translational genomics* 9: 37–39. doi:[10.1016/j.atg.2016.03.004](https://doi.org/10.1016/j.atg.2016.03.004)
- Knoppers BM, Harris JR, Tassé AM et al (2011) Towards a data sharing code of conduct for international genomic research. *Genome Med* 3: 46. doi:[10.1186/gm262](https://doi.org/10.1186/gm262)
- Manolio TA, Abramowicz M, Al-Mulla F, et al (2015) Global implementation of genomic medicine: We are not alone. *Sci Transl Med* 7: 290ps13. doi: [10.1126/scitranslmed.aab0194](https://doi.org/10.1126/scitranslmed.aab0194)
- Manrai AK, Funke BH, Rehm HL et al (2016) Genetic misdiagnoses and the potential for health disparities. *N Engl J Med* 375:655–665. doi:[10.1056/NEJMsa1507092](https://doi.org/10.1056/NEJMsa1507092)
- Moonesinghe R, Jones W, Honoré PA et al (2009) Genomic medicine and racial/ethnic health disparities: promises, perils, and the challenges for health care and public health policy. *Ethn Dis* 19:473–478
- Mulder N, Nembaware V, Adekile A et al (2016) Proceedings of a Sickle Cell Disease Ontology workshop—towards the first comprehensive ontology for sickle cell disease. *Applied & translational genomics* 9: 23–29. doi:[10.1016/j.atg.2016.03.005](https://doi.org/10.1016/j.atg.2016.03.005)
- Nippert I (2013) “CAPABILITY” and “genetic testing in emerging economies” (GenTEE). *J Community Genet* 4:293–296. doi:[10.1007/s12687-013-0158-9](https://doi.org/10.1007/s12687-013-0158-9)
- Pang T (2013) Genomics for public health improvement: relevant international ethical and policy issues around genome-wide association studies and biobanks. *Public Health Genomics* 16:69–72. doi:[10.1159/000341500](https://doi.org/10.1159/000341500)
- Parker M, Kwiatkowski DP (2016) The ethics of sustainable genomic research in Africa. *Genome Biol* 17:44. doi:[10.1186/s13059-016-0914-3](https://doi.org/10.1186/s13059-016-0914-3)
- Penchaszadeh VB (2015) Ethical issues in genetics and public health in Latin America with a focus on Argentina. *J Community Genet* 6: 223–230. doi:[10.1007/s12687-015-0217-5](https://doi.org/10.1007/s12687-015-0217-5)
- Rotimi CN (2012) Health disparities in the genomic era: the case for diversifying ethnic representation. *Genome Med* 4:65. doi:[10.1186/gm366](https://doi.org/10.1186/gm366)
- Rotimi C, Shriner D, Adeyemo A (2013) Genome science and health disparities: a growing success story? *Genome Med* 5:61. doi:[10.1186/gm465](https://doi.org/10.1186/gm465)
- Sirisena ND, Neththikumara N, Wetthasinghe K, Dissanayake VHW (2016) Implementation of genomic medicine in Sri Lanka: initial experience and challenges. *Applied & translational genomics* 9: 33–36. doi:[10.1016/j.atg.2016.05.003](https://doi.org/10.1016/j.atg.2016.05.003)
- Spratt DE, Chan T, Waldron L et al (2016) Racial/ethnic disparities in genomic sequencing. *JAMA Oncology* 2:1070–1074. doi:[10.1001/jamaoncol.2016.1854](https://doi.org/10.1001/jamaoncol.2016.1854)
- Stapleton G, Schröder-Bäck P, Townend D (2013) Equity in public health: an epigenetic perspective. *Public Health Genomics* 16: 135–144. doi:[10.1159/000350703](https://doi.org/10.1159/000350703)
- Tan DSW, Mok TSK, Rebbeck TR (2016) Cancer genomics: diversity and disparity across ethnicity and geography. *J Clin Oncol* 34:91–101. doi:[10.1200/JCO.2015.62.0096](https://doi.org/10.1200/JCO.2015.62.0096)