

Inclusion of Diverse Populations in Genomics Research and Health Services: Genomix workshop report

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Abstract

Clinical Genetic Services and Genomic research are rapidly developing but, historically, those with the greatest need are the least to benefit from these advances. This encompasses low-income communities, including those from ethnic minority and indigenous backgrounds. The “Genomix” workshop at the European Society of Human Genetics (ESHG) 2016 conference offered the opportunity to consider possible solutions for these disparities from the experiences of researchers and genetic healthcare practitioners working with underserved communities in the USA, UK and Australia. Evident from the workshop and corresponding literature is that a multifaceted approach to engaging communities is essential. This needs to be complemented by redesigning healthcare systems that improves access and raises awareness of the needs of these communities. At a more strategic level, institutions involved in funding research, commissioning and redesigning genetic health services also need to be adequately represented by underserved populations with intrinsic mechanisms to disseminate good practice and monitor participation. Further, as genomic medicine is mainstreamed, educational programmes developed for clinicians should incorporate approaches to alleviate disparities in accessing genetic services and improving study participation.

Keywords

Disparities, Genetic services, Ethnic minorities, Indigenous populations, Genomic research

Introduction

As genomics research enters its second decade after the completion of the International Human Genome Project, the research conducted still is not fully reflective of the diversity found in global populations. Underserved populations include both populations in low and middle-income countries and low-income communities in high-income countries. Although some efforts have been made to address disparities, the former continues to receive little attention even though there are many compelling reasons to do so e.g. achieving a greater understanding of genomic variation and reducing existing health inequalities (Christianson *et al.*, 2013; Nippert, 2013).

The latter includes minority and indigenous populations who have historically had poor access to genetic services. This has been seen both in general genetic services (Roberts, Cullen and Bunday, 1996; Khan *et al.*, 2010) as well as in specialist cancer services (Allford *et al.*, 2014). The poor access of African-American women to BRCA1 genetic testing is a particularly relevant exemplar (Armstrong *et al.*, 2005; Levy *et al.*, 2011). Further, the provision of genetic screening programmes may also be inadequately developed in these communities e.g. a lack of pre and post-test counselling in neonatal sickle cell screening (Hussein *et al.*, 2015).

This article will focus on underserved populations in high-income countries in the context of the USA, UK and Australia and explore why recognising these disparities matter, indicate some reasons for their existence and based on a workshop at the European Society of Human Genetics (ESHG) conference in 2016, consider possible solutions for addressing them.

Why does this matter?

Tackling inequalities in genomics research and access to genomic medicine are important to enhance the scientific rigour of research conducted in genomics, improve the utility and applicability of their findings to underserved communities and to understand the impact of access on health outcomes. Some diseases are more common in certain populations. This could be autosomal recessive diseases due to founder mutations, population carrier rate or consanguinity while other conditions may be more common but without a clear inheritance e.g. prostate cancer in men of African descent (Metcalfe *et al.*, 2008; Williams and Powell, 2009; Chornokur *et al.*, 2011; Farrell *et al.*, 2013; Martin, Starks and Ambs, 2013; Powell and Bollig-Fischer, 2013). The disease burden among migrant populations too are constantly evolving and the impact of Westernisation and globalisation means that there are new healthcare challenges that could be addressed through studying genomic variation (Garduño-Díaz and Khokhar, 2012; Dubé *et al.*, 2015; Barlas *et al.*, 2016).

However, there is a caveat: discussions with African American, Latino and White communities in the USA suggest that, although gene-environment interactions contribute to group differences in health outcomes, social conditions trigger group-level genetic differences and, in particular, contribute to poorer health outcomes among African Americans (Isler *et al.*, 2013). A review by Via *et al.* also suggested that researchers have to be mindful of the correlation between genetic ancestry and socioeconomic and environmental factors that could underlie differences seen in the disparities between different ethnic groups (Via, Ziv and Burchard, 2009).

Nevertheless, a lack of access to clinical genetics by these communities or a lack of awareness of these issues among healthcare professionals may have an impact on genetic testing and research and is likely to skew our understanding of human disease and variation. This, in turn, could make interpretation of results more difficult (Carlson *et al.*, 2013; Manrai *et al.*, 2016; Petrovski and

Goldstein, 2016) and lead to poorer health outcomes in these communities (Modell *et al.*, 2000; Susswein *et al.*, 2008). This is evident in pharmacogenomics research where efficacy and side effects of certain drugs may be affected by genomic variation and ethnicity (Kaneko *et al.*, 1999; Desta *et al.*, 2004; Cohen *et al.*, 2006; Hung *et al.*, 2006). Moreover, in cancer research, disparities in cancer morbidity and mortality that adversely impacts underserved groups may in part be ascribable to a failure to include diverse populations in biospecimen banks (Lawson *et al.*, 2015).

However, all future research with underserved, particularly minority and indigenous communities, will have to overcome mistrust from historical malpractices (Gamble, 1997; Corbie-Smith *et al.*, 1999; Fairchild and Bayer, 1999; Scharff *et al.*, 2010; Boyer *et al.*, 2011; Sheppard *et al.*, 2013; George, Duran and Norris, 2014). This includes the Havasupai Diabetes Project where blood samples consented for an investigation into the high incidence of diabetes amongst the Havasupai tribe were used to investigate genetic causes of schizophrenia, inbreeding and population migration theories without the tribe's consent (Mello and Wolf, 2010; Pacheco *et al.*, 2013). This mistrust has propagated within these communities and is ever present today. A survey of African American premedical students suggested that these students have several concerns about genetic testing- including discrimination, privacy and eugenics. Surprisingly these concerns were increased, not lessened, by genetics education (Laskey *et al.*, 2003).

The next section highlights solutions from a ESHG workshop to improve access of underserved groups to genetic services and research opportunity, particularly focusing on minority and indigenous communities.

Possible Solutions

Community engagement

Critical to improving underserved communities' access to genomic services and participation in genomic research is early **community engagement**. This involves working with communities to identify their needs and concerns. Strategies to address these could involve town hall meetings that are popular among African American communities in the USA (Ansell *et al.*, 2009; Fouad *et al.*, 2010; Schoenfeld and Francis, 2016) or more defined focus groups (Streicher *et al.*, 2011; Walker *et al.*, 2014). The work of the National Centre for Indigenous Genomics (NCIG) at the Australian National University (ANU) with Aboriginal communities is an excellent example of the latter where focus groups were used to recognise disparities, address past poor research practice and explore themes concerning culture, kinship and genes, and the language of inheritance (Callaway, 2016).

Focus studies conducted among African American and Latina/Latinos in the USA also identified communication strategies as a key tool to reduce barriers. This includes reducing language barriers, increasing dispersion of information via a variety of channels and engaging representatives from the communities of interest (Schulz, Caldwell and Foster, 2003). As such, multifaceted channels of communication facilitated by community leaders, faith leaders and patient champions could also be used to engage with these underserved communities and build trust (Yancey, Ortega and Kumanyika, 2006). The most successful of these strategies seem to be directed by the communities themselves e.g. videos produced by the Aboriginal communities with the NCIG and written material produced for the British Pakistani community in East Lancashire, UK. These efforts can also be reinforced by the use of role models or celebrities that can have a major impact on healthcare behaviour (Evans *et al.*, 2014).

Another example of community engagement is the outreach work in Leicester, UK. For many years the Leicester Genetics Education Centre of Excellence in Teaching and Learning (GENIE) has developed a reputation in community education outreach, from teaching DNA fingerprinting and promoting mental health awareness in schools to explaining how the remains of Richard III were identified through mitochondrial inheritance techniques (King *et al.*, 2014). This led to the inception of the 'Supporting Families with Cancer' project with Macmillan Cancer Support which resulted in additional outreach events, stakeholder projects, primary care triage projects and developing the world's first YouTube channel for clinical genetics (Lakhani *et al.*, 2013; Jones *et al.*, 2016). The clear message from these projects was that patients felt that supporting general practitioners in the community make effective referrals into specialist services was a key principle to accessing good care.

Redesign health care systems

Beyond community engagement, it is important to offer tangible improvements to health care systems. This includes strategies to communicate with patients from linguistically diverse backgrounds. This is demonstrated in Victoria, Australia where nearly a quarter of its population speak 260 languages other than English at home. Victoria's population is diverse and rapidly changing with 46.8% born overseas or having at least one parent born overseas and where the overseas-born population continues to steadily rise (Australian Bureau of Statistics, 2011).

The Melbourne Genomics Health Alliance was a whole system approach to genomics driven by ten leading healthcare and research organisations in Victoria who came together to advance their joint interest in bringing genomics into healthcare. In 2013, the 7 founding members funded a collaborative and shared Demonstration project where patients with five diverse genetic conditions received genomic sequencing in parallel with standard care (Gaff *et al.*, 2017). The major aim of the project was to understand patient experiences of having genomic sequencing and their preferences for this type of testing in the future- using surveys as the mode of data collection. Patients were recruited based on their condition; therefore, challenges were not related to testing access but around gaining feedback from these participants on their motivations, concerns, preferences and understanding of genomic testing.

It was evident from this project that the experiences and preferences of Culturally and Linguistically Diverse (CALD) individuals will be important in determining the systemic changes that are required to overcome the challenges of embedding genomics into routine clinical practice. Preliminary data from the project demonstrated that 11% identified as English as a Second Language (ESL) with at least 18 separate first languages being spoken with little overlap. Although translation of the surveys into other languages was not deemed to be suitable due to the diversity of languages spoken, other strategies were implemented over the course of the project to increase survey participation from CALD individuals. These included exploring whether the survey could be completed with a family member, a trusted healthcare professional who can translate or over the telephone with a telephone interpreter. Melbourne Genomics now offers genomic sequencing to patients with 11 different conditions. Further consultations are being conducted with Indigenous and CALD groups to determine if there are cultural sensitivities that require consideration when developing structures and policies regarding the management of genomic data. The Demonstration project clearly highlights the benefits of a whole system approach to genomics and particularly improving access to translators and language/culturally sensitive materials (Hussain-Gambles, 2003) in engaging traditionally underserved communities.

Improving access using genetic outreach workers, simplifying referral processes and multi-agency partnerships will also complement such efforts (Khan, Kerr and Kingston, 2016). The work done in East Lancashire, UK is a good example of the latter where a systems approach to improving engagement with the British Pakistani community was undertaken by an innovative collaboration between primary care organisations (PCOs), commissioning groups, public health and the regional genetic services.

Approximately 30% of the population in East Lancashire is Muslim and of South Asian heritage. It has one of the highest child mortality rates in the country (Public Health England, 2016) with 41% of the deaths in under 18-year-olds resulting from a chromosomal, congenital or genetic disorder. With up to 75% of British Pakistanis in the region being in a consanguineous marriage and with no indications of a decline in this practice, the need to engage with this community was identified.

However, traditionally, uptake among these families have been poor as the utility of this service is not recognised (Khan *et al.*, 2010). As such, a multi-stranded approach that included investments at the community level, alongside genetic service enhancement and training of healthcare professionals was employed that has proven to be more effective than single stranded approaches. A key to the success of this approach has been the provision of enhanced genetics advice which included no language barriers, an understanding of, and sensitivity towards, the cultural context in which decisions are being made including gender, religion and cultural complexities. Healthcare inequalities have been reduced in East Lancashire through this collaborative approach. Families are presenting for information/support who hadn't previously attended clinics.

Strategic level changes

Ensuring that underserved populations are involved in genetic and genomic projects currently relies on pockets of good practice by interested parties. To ensure that this is consolidated, sustained and rolled out to other groups will require fundamental institutional level changes (Popejoy and Fullerton, 2016) such as the changes delivered by the Athena SWAN Charter for women in science. Under-representation of women in academia is an internationally recognised disparity that has persisted through time. This is particularly evident in academic medicine where just 28% of clinical academics in the UK are women. This disparity exacerbates with seniority as although women account for 42% of lecturers, only 18% are professors (Medical Schools Council, 2015).

Amidst this background of gender-inequality, the Athena SWAN Charter gender equality award scheme is one initiative that has made some inroads to tackle the prevailing disparity. The Charter established in 2005 by the Equality Challenge Unit (ECU) was instituted to encourage and recognise efforts to advance the careers of women in science, technology, engineering, maths and medicine (STEMM) (Caffrey *et al.*, 2016). Since 2011, biomedical research units, biomedical research centres and patient safety translational research centres that apply for National Institute for Health Research (NIHR) approved grants are required to reach a certain level of compliance with a series of outcome measures for improving the number of, and opportunities for, female academics in their respective institutions. The Athena SWAN has had a positive impact in advancing gender equality and although there is limited evidence at present to attribute the Athena SWAN to the observed increase in women in academic medicine (Gregory-Smith, 2015), it has raised awareness of gender inequality in the workplace and has brought about important structural and cultural changes to address gender inequality. A key to the success of this initiative may have been linking the Athena SWAN to government research funding (Ovseiko *et al.*, 2017).

As such, similar initiatives may also be used to facilitate adequate representation of underserved populations in genomic research and as patient representatives at all levels e.g. grant committees, ethics committees and research steering groups. Grant selection criteria, ethical approval and research progress reports should also include ongoing assessments of the representativeness of study populations. Moreover, patients from underserved communities and researchers with an interest in healthcare access equality need to actively participate in and apply lobbying pressures on commissioning groups to ensure that initiatives that address disparities are kept at the forefront of discussions on service design and research specification.

Further, clinicians serving underserved populations will need ongoing education on advances in genomic medicine and management approaches based on genetically determined variants (Feero and Green, 2011; Radice *et al.*, 2011). Although understanding which genomic variants are medically actionable beyond standard phenotypic information will require ongoing efforts to improve patient participation from diverse backgrounds, educational strategies to reduce inequalities in access as well as developing an understanding of cultural factors in minority communities should be pursued.

Research databases should also be sufficiently powered with participants from ethnic minority groups and other underserved communities to draw meaningful conclusions (Chow-White and Duster, 2011; Lawson *et al.*, 2015). This applies to studies such as the UK 100,000-genome project with its aims to develop a matched genomic variant and clinical phenotype anonymised data set in an agreed, standardised and unified format with longitudinal pulled through data sets to assist with national and international clinical research and commercially driven collaborations to improve our understanding of human variation and how this links to disease and influences healthcare outcomes given therapeutics interventions. This can also be enhanced through replication studies in other countries if these are widely accessible and in a readable and usable format. As society becomes increasingly diverse, this also includes embracing more objective measures of genetic makeup beyond traditional definitions of race and ethnicity (Mersha and Abebe, 2015).

Finally, the coding of ethnic group status in referrals to genetics services and participation in genomic projects needs to be accurate so that healthcare outcomes and access to new technologies can be prospectively compared for the studied populations. This data is currently often incomplete and needs to be more routinely and systematically captured. Further, the success of strategies that address disparities also needs to be documented using practical metrics, for example, increase in appropriate referrals, increase uptake in the extended family and, in the case of reproductive genetic risk, reduced infant morbidity and mortality.

Conclusion

Inequality of access to new types of medical technologies through socioeconomic or ethnicity barriers are likely to accentuate healthcare disparities. There are moral, scientific and historic reasons why including underserved groups in Genomic Medicine projects and research is advisable to truly understand human genomic variation and improve healthcare outcomes. This will require multi-faceted culturally sensitive, educational and outreach based approaches that simplify patient pathways and are underpinned by understanding the needs of and working with, local communities.

With the drive towards personalised medicine, it is imperative that existing inequalities in genomics research and disparities between communities are addressed to prevent exacerbating them. Targeted education and outreach could help to engage minority communities and break down the barriers that hinder access to genetic services. However, the current definitions of race and ethnicity

limits the ability to assess recruitment to genomic research and the time has come for medical research to embrace more objective measures of genetic makeup.

Recommendations

Community Engagement

- I. Use multifaceted channels of communication with the aid of patient champions, faith leaders and community leaders to engage with and educate underserved populations
- II. Enable general practitioners in the community to make effective referrals to specialist services

Redesign health care systems

- I. Improve access to translators and language/culturally sensitive material
- II. Use multi-stranded approaches in a multi-agency manner to engage with communities and advance joint interests

Strategic level changes

- I. Implement institutional changes in grant requests and service design and ensure there is adequate representation of underserved communities at all levels
- II. Provide ongoing education for clinicians serving underserved populations in advances in genomic medicine and strategies to tackle disparities in access and research participation among underserved communities
- III. Ensure databases are adequately powered by individuals from underserved communities
- IV. Complete ethnic minority coding in referrals and utilise practical metrics to document success of strategies

Compliance with Ethics Guidelines

Savio Mathew, Julian Barwell, Naz Khan, Ella Lynch, Michael Parker and Nadeem Qureshi declare that they have no conflict of interest. This narrative article does not describe primary research.

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