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# Culture, law, ethics, and social implications: Is society ready for advanced genomic medicine?

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## EDITORIAL

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In recent years, rapid and continuing advances in our knowledge and interpretation of information on the human genome have transformed our understanding of human health, medical diagnostics, and therapy. Accessing information on genetic material is now quicker, cheaper, and more widespread than at any other time in our history. As innovations and discoveries involving human genetic material continue, there will be an ongoing necessity to carefully integrate advanced genomic medicine (AGM), including personalised genetic diagnostics and intervention, into the wider community. AGM offers the potential of personalised genomic profiling that enables the diagnosis of disease and targeted treatment, yet despite the advantages of such technology, there are issues with accurate diagnosis and interpretation of data. Even experts struggle with the complex nature of the data that is currently gathered.[1,2]

In parallel to AGM, the general public are already sending saliva samples to explore their genetic makeup using the Internet through companies and websites like 23andMe (*https://www.23andme.com),* usually without clinician involvement. They are also sharing their health experiences through websites such as PatientsLikeMe (*www.patientslikeme.com*). Medical professionals who are already coming to terms with the availability of an increasing number of genetic-based screening tests may now be confronted with an engaged, informed public who are both requesting and presenting medical professionals with a multitude of data about their genetic profile; similarly, an increasing number of screening tests and diagnostic procedures involving genetics are being used in clinical settings. As the science and technology behind AGM continues to grow, so too will the urgency to ensure the successful transfer of this technology from bench top to bedside—from scientific discovery to practical application. The development and application of new techniques and ideas will need to be carefully addressed by considering a whole-of-system approach as scientists, the medical fraternity, and the public come to terms with this new technology.[3] Both the medical profession and the public have high expectations of AGM; however, many potential barriers are already visible, including cultural, ethical, educational, legal, and social implications, and if these are not addressed successfully, such issues could potentially limit the growth of this exciting field.

## Community integration

AGM has tremendous potential to provide the public with health benefits. What is unknown, however, is the public's receptiveness or resistance to the use of AGM in the community. How will we know whether communities are ready or likely to welcome or reject the introduction of AGM? Presently, there appear to be few answers as to whether the expansion of AGM would be acceptable by the public, and in what form. Each community likely has its own beliefs, values and wishes in relation to genetic material and the use of information that arises from their own genes. Lack of sensitivity or ignorance of these beliefs, values, and wishes may confound any future use of the technology; proponents of widespread use of AGM may find that without exploration and identification of potential barriers the successful use of such technology is hindered or even prevented. To ensure the smooth delivery of AGM, it will be necessary to facilitate both vertical and horizontal integration of AGM services, with horizontal integration providing standardised levels of care and service delivery across each group, while vertical integration will involve providing consistency of practise throughout the passage of delivery regardless of the type or nature of AGM involved.

The ethnic diversity of each community will also likely deliver perspectives on AGM that will need to be independently assessed and addressed. Indigenous peoples, immigrant communities and various other sub-groups (e.g. such groups defined by age or generation) within society may all have different views on how AGM applies to their culture or life experiences. Ignoring the worldviews, beliefs, and values of any individual group may well prove problematic and affect the wholesale delivery and uptake of AGM to the wider community.

Healthcare professionals must not remain a “silent contributor” to such conversations. At present, it is unknown how AGM is viewed by the medical community: their knowledge about AGM, and even willingness to use it, is at this time relatively unexplored. Given medical professionals will be at the forefront of AGM use in both hospitals and the community, an understanding of the profession’s readiness to provide such a platform is a key component of the future use of this technology.

How do the opinions and attitudes of consumer groups and patients in relation to AGM differ, in relation to diagnosis, intervention, prevention, or support? For instance, what does the term “informed consent” mean to individuals and groups? The opinions and attitudes of consumer or user groups are integral to the successful delivery of health services in this new age of medicine. By understanding how society views and relates to AGM, effective frameworks can be designed and implemented that will lead to the efficient delivery of AGM in health care.

**Already, insurance companies have been known to modify the acceptance of policies or claims based on genetic tests; it is unclear how the provision of larger amounts of genetic information will be handled, and for what purposes. “Genetic tourists” now travel overseas to have screening tests prior to contacting insurance agencies because of the potentially negative consequences of some genetic test results.**

Until communities are consulted—including both medical and consumer communities—the integration of AGM in society will certainly be uninformed, potentially problematic, and possibly inefficient. Issues of access (public vs. private) and cost will need to be addressed to provide services where they are required and accepted; such matters require the exploration and identification of both community and culturally appropriate values or wishes, and are essential in providing a platform for AGM in the community. If such issues are not addressed, the use of AGM could increase the healthcare gap between the rich and the poor, thereby widening any pre-existing healthcare inequities.

## Regulation

As AGM becomes more widely used, undoubtedly more regulatory issues will come to light. Good practice standards, strong privacy measures, professional guidelines, and ethical guidance that take into account support from the community and medical professionals, and robust legislation that allows successful AGM health service delivery will need to be considered as technology develops. The impact of AGM on the community will most likely require healthcare delivery and systems to be managed and regulated, but there is no consensus yet on how this should be achieved. The legal fraternity will be called upon to assist in the development of laws relating to the use and implementation of new technology; ethics committees will be asked to comment on as yet unknown technologies and uses of information; and policy makers will need to balance the range of diverse and potentially competing interests within the regulatory framework. Cost effectiveness will need to be considered, as will issues around the potential horrors of eugenics that will become more possible as costs lower and the use of AGM technology becomes more widespread.

Deciding how researchers may approach patients, how they may retain and store information, and how such information may be shared will all become pressing questions as AGM becomes more widely used. Researchers will not require physical samples to make breakthroughs in medical science—the large data sets about individual patient’s genetic material that are held on computers are now able to be transmitted around the world at the push of a button. How this information could and should be shared are questions that require ongoing public consultation. Failure to do so may prove difficult to reconcile with the general public, with the example of the historical, unconsented use of Henrietta Lacks cancer cells—the first human immortal cell line, responsible for thousands of scientific experiments and publications—providing ample stimulus for such consultation to be initiated.[4]

As AGM continues to develop, so will the need to regulate and bring together patient, community, and medical requirements to cater for acceptable standards and best practices. This may include the participation of groups that are commercially interested in genetic information such as the health care, cosmetic, and pharmaceutical industries, and insurance agents. Already, insurance companies have been known to modify the acceptance of policies or claims based on genetic tests;[5] it is unclear how the provision of larger amounts of genetic information, and the access to this information, will be handled and for what purposes. In addition to accessing websites to obtain information about their genetic profile, "genetic tourists" are now travelling overseas to have screening tests prior to contacting insurance agencies because of the potentially negative consequences that genetic test results may bring. At present, there is little information that relates to individual communities in terms of regulatory gaps or enablers, consumer-informed standards, or guidelines for the implementation of AGM; the development of appropriate guidelines will need to involve all parties so that AGM can serve to benefit patients, their families and communities.

## Education

Presently, it is unclear how much knowledge the wider community has about AGM. This includes cultural, ethnic, and medical groups. Until such information is gathered, it will remain difficult to plan information campaigns or strategies to implement AGM effectively. Knowledge needs to be assessed on not only the type of technology that is available, but also other key issues such as how the information will alter as treatment outcomes, side effects, regulations, patient guidelines, storage and access to personal information, and the law.

In recent years there have been instances of adverse publicity from genetic discoveries that have likely generated suspicion and distrust in the general public. The claim concerning unconsented discovery of the "warrior gene" from genetic samples in New Zealand,[6] and the unconsented use of genetic information from the Havasupai Indians in America,[7] are just two instances of misuse of genetic information that have yielded adverse publicity that could hinder the development or introduction of AGM to the wider community. The adverse publicity that arose from unconsented use and publication of data arising from samples of genetic material may well have compromised the public trust in such technology, especially from ethnic groups who have not been consulted appropriately. In such instances, education refers not only to the level of knowledge of the consumer, but also to scientists and medical professionals who must understand how the development and use of genetic technologies are critically dependent on synergies and understanding between all AGM user groups.

## Conclusion

The use of AGM will continue to develop and progress as future scientific advances are made. Patient awareness and attitudes, including the identification of barriers for service delivery, will need to be explored to facilitate the effective introduction of AGM. Following such progress will come a variety of challenges that encompass various cultural, legal, ethical, and social issues. In order to successfully use AGM in society, and to allow efficient and respectful delivery of AGM for the benefit of all user groups, conversations that bring all the affected user groups into consideration need to happen now. The perspectives of all end users—patient, science, and medical groups—need to be sought immediately to enable and facilitate the smooth integration of AGM into society. Failure to do so will potentially hinder the significant healthcare advances that AGM offers the community.

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## PEER REVIEW

Externally peer reviewed.

## CONFLICTS OF INTEREST

JC is the deputy editor of the AMJ.