



Education and personalized genomics: deciphering the public's genetic health report

Where do members of the public turn to understand what genetic tests mean in terms of their own health? Now that genome-wide association studies and complete genome sequencing are widely available, the importance of education in personalized genomics cannot be overstated. Although some media have introduced the concept of genetic testing to better understand health and disease, the public's understanding of the scope and impact of genetic variation has not kept up with the pace of the science or technology. Unfortunately, the likely sources to which the public turn to for guidance – their physician and the media – are often no better prepared. We examine several venues for information, including print and online guides for both lay and health-oriented audiences, and summarize selected resources in multiple formats. We also note on the roadblocks to progress and discuss ways to remove them, as urgent action is needed to connect people with their genomes in a meaningful way.

KEYWORDS: DTC test ■ education ■ genetics ■ genomic medicine ■ genomics ■ journalism ■ personalized genomics ■ science

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Two developments in the past decade have combined to produce direct-to-consumer (DTC) genetic and genomic testing on a scale that outstrips predictions made by academic human geneticists only a few years ago. First, the Human Genome Project, the International HapMap Project and others spurred the development of both genetic maps of human variation and technologies to test this variation on a genome-wide scale. In 1998, students could earn a PhD degree for describing a handful of novel SNPs in a gene; in 2008, students could assess 300,000 or more SNPs in thousands of individuals to reach the same degree. In a very short time it has become technically and economically feasible to offer not just single-marker DNA tests but scans for hundreds of thousands of genomic variants.

Second, the rise of the internet and social media has allowed nascent companies to reach the lay public in multiple different ways. The average consumer may first have heard about DNA testing in a courtroom drama or a forensic science television show, rather than in a context of health and disease. Alternatively, they may have seen various billboards advertising paternity testing: 'Who's the daddy?' in Baltimore (MD, USA), 'Paternity testing' in Tennessee (USA), or 'Are you the father?' in Toronto (ON, Canada), to name a few. The profile of DNA testing received an additional boost in 2005, when talk-show host Oprah Winfrey and other celebrities went through DNA ancestry tests.

The results, documented in multiple media outlets, revealed an unpredicted and complex genomic composition for each individual tested.

It is only in the last 2 or 3 years that DNA testing for health- or disease-related purposes, rather than for ancestry or forensic uses, has been widely discussed. Members of the lay public are now much more likely to encounter any form of genetic testing in healthcare settings such as during a pregnancy or cancer diagnosis. But the availability of genome-scan tests and their use in genome-wide association studies (GWAS) predicting a percentage risk for complex diseases has launched a new industry of DTC genetic testing. The 'Retail DNA Test', specifically involving a genome scan for hundreds of thousands of SNPs as offered by the company 23andMe (CA, USA), was even chosen by *Time* magazine as the '2008 Invention of the Year'. Along with the meteoric rise in throughput and the complexity of genetic tests, DNA testing companies have recently branched out with their marketing efforts to reach mainstream audiences, with a particular focus on web-based approaches. The web-service Google Ads (Google, CA, USA) even allows companies to target their advertisements to specific news stories – for example, the *New York Times* website's 1 September 2009 story by Jane Brody entitled: 'Buyer beware of home DNA tests' was, when we accessed it, followed by three advertisements for DNA testing (two for ancestry determination and one offering 'testing for more than 200 genetic disorders'). As

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we will detail, companies have set up their own Facebook [101] and Twitter [102] accounts to reach individuals anywhere with an internet connection; the latest group of genome-scan firms also include extensive web-based genome navigators as part of their services. Like many other authors on this topic, we note there is a difference in the quality of information provided between companies at the cutting edge of science (examples include 23andMe, deCODE and Navigenics) and other companies whose level of service is much less laudable, but in this perspective we consider the resources available to individuals seeking information on any test of DNA.

Estimates of public enthusiasm with regard to DTC genetic or genome-scan testing vary widely depending on how the question is asked – from the 91% responses in favor of genetic testing measured in a 2008 survey conducted by a market research firm [1] to 0% in a smaller Australian study specifically asking about DTC ‘genetic testing marketed through the internet’ for depression [2]. A larger study found that only 22% of US consumers responding to a survey were aware of ‘personal genomic tests based on GWAS variants’ and only 0.3% had actually used such a test; 42% of health professionals said they were aware of similar tests [3]. The rise in DTC genetic or genomic tests caught many human geneticists off guard, since they were used to working with a framework of well-educated genetic counselors to explain the complicated and sensitive issues of a scientific process to patients who had been referred for genetic testing [4]. Instead, many new users of DTC tests are asymptomatic but are interested in the risk factors conveyed by combinations of genetic variants. These users may modify their lifestyle or seek further unnecessary medical procedures in response to data that have not been replicated or information that has not been accurately explained to them [103]. Given the enthusiasm of the public, in this brave new world there is a dire need for education geared towards the public on genetic or genomic testing. In this perspective, we will focus on the available resources and the critical deficits in education available to three main groups – the lay public receiving genetic tests, health professionals being asked to interpret these tests and the media communicating genetic information to both parties.

Public

Faced with the advertising and new awareness of genomic tests heralding the advent of ‘personalized medicine’, where would a member of the

lay public begin his or her search for information on the background and validity of these tests? Some may start at their local library or bookstore, searching for books on genome testing or personalized medicine. The book selection has the potential to greatly impact their view on testing – those choosing literature such as Craig Venter’s book, *A Life Decoded: My Genome, My Life* [5], may come away enthusiastic about the possibilities of the information provided by their own genome with regard to their health. Others might discover books such as *The Soul Genome* by Paul von Ward [6], which actually describes theories of reincarnation, and take away an entirely different interpretation of genome testing. In our admittedly subjective experience in genetics education of over 15 years, the most useful educational books specifically for genomic information (thus focusing on very recent efforts) include [7–9].

We also look forward to the January 2010 book releases specifically focussed on personalized genomics from Huntington Willard and Susanne Haga (Duke Institute for Genome Sciences and Public Policy, NC, USA) [10] and from Francis Collins (NIH) [11].

Of course, many people will turn to the internet for information – a Google search for ‘genetic testing’ returned more than 1.5 million hits (conducted on 18 August 2009). Genomics is no different to any other subject – the internet contains the entire spectrum, from very useful and accurate sites, to sites that are well advertised but misleading at best and downright dangerous at worst. Basic resources such as WebMD [104] are not always useful either – a search for the term ‘genomic’ yields a first link to a relevant news story from the National Cancer Institute (MD, USA) on prostate cancer susceptibility and genomic variation, but the second link is to a ‘health tools’ slideshow on head lice. In **Box 1**, we have selected an alphabetical list of – in our experience of education and public communication activities in genetics – valuable and well-curated web resources for public, physicians and the media. Using these websites as an initial reference, individuals can develop a foundational understanding of genetics, genetic tests and risk assessment. We would like to mention a few in more detail.

- Genetic Alliance [105] – their ‘Trust It or Trash It?’ quality assessment tool is especially useful, which is designed to help the public judge the quality of educational information related to genetics and genetic tests [106].

- Genetics and Public Policy Center [107] – the Center has conducted a number of studies on public opinion and genetic testing, representing some of the best data we have in the area.
- Scitable [108] – this collection of reviews covers many topics in genetics and has separate spaces for faculty and students (Chris Gunter [HudsonAlpha Institute for Biotechnology, AL, USA] was consulted regarding aspects of the site throughout its creation).
- PHG Foundation [109] – website for the UK's Foundation for Genomics and Population Health; reporters have told us they appreciate the news coverage of scientific articles that have just been published on this site.

Obviously, we must also mention the three main DTC genome-scan test companies at present – 23andme (CA, USA), Navigenics (CA, USA), and deCODEme (Reykjavík, Iceland). (In June 2009, another company called Pathway Genomics entered this arena as well.) These companies have clearly put much effort and creativity into their presentations of genetic/genomic information to the public. Generally speaking, the deCODEme and Navigenics services concentrate more on the relationship between genetic variants and disease, while the 23andMe site facilitates a more of an exploration of social networking and focuses on the cachet of knowing more information about one's self. We have experience with two of the sites and find that with deCODEme, the ancestry information is presented in a clearer way, but the background information is not as useful for our desired applications in educational settings. For 23andMe, we found that the division of raw data into two groups was valuable – clinical reports and research reports ('research that has not gained enough scientific consensus to be included in clinical reports'); the research reports are rated with 1–4 stars. This helps to convey some of the uncertainties behind the GWAS results on which many of the claims about risk are based.

In 2009, another source became available that is often used by the public – the so-called 'social media', platforms that allow individuals to share media and content with a wide audience and to interact instantly. These include wikis, blogs and microblogs, such as the service Twitter [102], where one posts one's thoughts with a limit of 140 characters at a time, as well as networking sites like Facebook [101] and MySpace [110]. There are more than 100 Facebook Groups associated with genetic

Box 1. Alphabetical list of selected websites useful for genetics and genomics information.

- <http://learn.genetics.utah.edu/>
- www.23andme.com
- www.ashg.org/press/
- www.cdc.gov/genomics/resources/index.htm
- www.decode.me
- www.dnapolicy.org
- www.dolandc.org
- www.geneticalliance.org
- A vision for the future of genomics: education and community engagement: www.genome.gov/27530874
- www.nature.com/scitable
- www.navigenics.com
- www.ncbi.nlm.nih.gov/sites/GeneTests/?db=GeneTests
- www.nchpeg.org
- www.phgfoundation.org/

A more extensive list, particularly for physicians, can be found in [31].

testing, ranging from genealogy to disease support groups [101]; anyone with a Facebook account can join such a group and post comments that would reach other group members. In the same vein, there are more than 15,000 Twitter accounts with 'DNA', 'genetic', 'genomic', or 'genome' as part of their name or self-description (searched for using [111]). Given the propagation of DNA imagery into popular culture, it is not surprising that a number of these accounts are not even linked to science, but a number of Twitter accounts are in fact used to disseminate research findings, pitch genetic products or educate followers regarding personalized medicine.

On Twitter, anyone can set up an account and 'follow' other individuals whose comments they find interesting; in turn, once you start to post your own 140-character thoughts or 'tweets', you may be 'followed' by people interested in your comments. In our experience (Chris Gunter manages accounts for both herself and our workplace), with some judicious filtering applied, Twitter is a valuable tool for keeping up with developments in personal genomics across the web. In Box 2, we have compiled a list of Twitter accounts that include companies in DTC testing, consumer advocate groups, scientists working in the field, science journalists who are often writing in the field, physicians and members of the public who have become enthusiasts on the topic. These represent many, but of course not all, of the users commenting on personal genomics and genetics. The alphabetical list captures those we have subjectively found to be the most useful, insightful and consistently active over the past year.

Box 2. Alphabetical list of selected Twitter accounts offering regular personal genomics commentary or information.

- @23andMe
- @AccessDNA
- @bcriskdoc
- @blaine_5
- @bmahersciwriter
- @dgmacarthur
- @DNABloggers
- @DNAPolicy
- @Duncande
- @EdwardWinstead
- @Epigenetique
- @geneticalliance
- @geneticist
- @GeneticsSociety
- @genomepop
- @GenomeResearch
- @GenomeWeb_News
- @genomicslawyer
- @genomics_xprize
- @girlscientist
- @helixhealthct
- @hudsonalpha
- @humangenomeorg
- @j_perkel
- @knome
- @markgfh
- @moorejeh
- @myGeneticist
- @NatureRevGenet
- @Navigenics
- @PathwayGenomics
- @patsycat21
- @phylogenomics
- @ResearchAmerica
- @YourFaveGene

Data from [102].

Twitter users, such as those in **Box 2**, regularly provide information on the latest developments and debate amongst themselves as to the merits or flaws in genomic testing. Importantly, for those seeking primary information, Twitter users often use their comments to post a weblink to research articles or blog entries that give more details on the data behind stories. One such post from @genomicslawyer on 18 September 2009 read:

“What happens if a DTC genomics company goes belly up? <http://bit.ly/JPfTz>.”

This post linked to a blog entry with the same title [112] that was sparked by the observation of financial difficulties affecting several companies.

For this blog entry, two lawyers examined the privacy policies in detail from a number of DTC genomics companies. They conclude:

“...the prediction here is that if your DTC genomic company of choice goes belly up, there is a good chance that its assets, including its database of genomic information, will be up for sale.”

Clearly, this information is useful to members of the public whose concerns about privacy and security of their genomic data will dictate their choice of testing service.

Physicians

Multiple studies such as those from the Genetics and Public Policy Center (Washington DC, USA) [103] have demonstrated that members of the public favor genetic testing and trust their doctors to have the results, while overwhelmingly not trusting their employers or health insurers with the same information. Although we in no way wish to minimize the role of all health professionals in providing genetic or genomic information to their patients, here we will focus on the educational resources available to physicians specifically. As in many areas, the demands on the physicians' time and knowledge with respect to integrating genetics into their practice are increasing at a rate that outpaces their educational background. The ability to recognize individuals who are candidates for genetic testing, identifying the appropriate test and providing both pre- and post-test counseling are becoming an increasingly important part of many specialties. However, for many physicians, their lack of genetic knowledge serves as a roadblock for these skills [12,13]. While clinicians grapple with the validity and implication of ordering a genetic test for a single disorder, DTC genetic testing now offers patients the opportunity to receive an avalanche of information regarding the risks for a cornucopia of genetic disorders, most of which may never have crossed the patient's mind.

In 2007, the American Society of Human Genetics (ASHG) released its recommendations on DTC genetic testing [14]. For physicians, the society stated:

“To ensure that providers are aware that genetic tests are being provided DTC and that some of these tests may lack analytic or clinical validity, professional organizations should educate their members regarding the types of

genetic tests offered DTC, so that providers can counsel their patients about the potential value and limitations of DTC testing.”

One of the largest groups in this area, the National Coalition of Health Professional Education in Genetics (NCHPEG [MD, USA]), states that ‘All health professionals should understand’, among a list of core competencies, ‘one’s professional role in the referral to or provision of genetics services, and in follow-up for those services’ [113]. Similarly, the Centers for Disease Control (CDC) and Prevention (GA, USA) also offer a comprehensive list of genomic competencies for all health professionals [114].

A study of primary care physicians in north western USA [12] identified their desire for access to up-to-date information regarding available genetic tests, guidelines to identify appropriate candidates for testing and specialized skills related to risk communication. Importantly, while primary care physicians were willing to consider integrating genetic testing into their practice if doing so would improve the care of their patients, many of those surveyed did not feel that most genetic tests alter patient management. While this view may have been valid in the past, as the applications of genetics in medical practice continue to emerge, genetic testing will play a central role for the practicing physician. In 2007, the US FDA responded to published studies on variation found in two genes, *CYP2C9* and *VKORC1*, and included pharmacogenetic information in the product labeling of the drug warfarin [115]. For hepatitis C, a very recent study demonstrated that variation in the human gene *IL28B* predicts a twofold change in response to the nearly year-long drug regimen given to patients; these results can be translated into clinical practice almost immediately, particularly when helping to convince patients who are reluctant to endure this regimen that their genetic data indicate that it will be effective [15]. The use of single-gene tests, or of those of a pattern of a few genes, are reaching more doctors, particularly as they demonstrate prognostic value in cancer or chronic diseases.

As mentioned above, a US study reported that 42% of health professionals said they were aware of ‘personal genomic tests based on GWAS variants’ [3], which is similar to a recent study from Japan showing that only 38% of general physicians were aware of DTC genetic testing. In Japan, those physicians that were aware of testing had heard about it in the same manner as the public – through media sources or the internet [16]. More

and more physicians will be presented with the results of genetic tests, particularly genome-wide scans. They must therefore be able to interpret risks, a moving target as new studies are released, as well as explain that an ‘elevation’ in risk for a specific variant may only be 1.2-fold, which is not very high. The physician may be asked to translate the risk to family members, accounting for genetic and environmental factors that remain incompletely understood even by scientists on the cutting edge of this research. At the same time, they are under pressure to comply with government regulations (some still in flux) on the privacy of these data and on nondisclosure to third parties [17]. This gulf between the expectations placed upon physicians and the training available to them leads to warnings like the conclusion of a 2008 *JAMA* review:

“Many gaps in knowledge about organization, clinician and patient needs must be filled to translate basic and clinical science advances in genomics of common chronic diseases into practice” [18].

It is important to realize that the genetic training received by many physicians occurred during their undergraduate training, which may have been several decades ago when genetics was viewed as a minor subspecialty. Even today, coverage of genetics varies widely across medical school curricula. A recent study [19] demonstrated that less than a half of the medical schools surveyed reported offering a stand-alone course in genetics, with 20% of medical schools devoting fewer than 20 contact hours to the subject. Barely a tenth of the respondents noted that practical training in genetics was a principal course objective. Unfortunately, these findings are reinforced by studies demonstrating that genetic-based information and skills learned early in medical school are poorly retained [20], and that the experiences of medical residents with genetics in medical school were almost entirely related to rare disorders and thus may unfortunately be of limited clinical relevance to many of their patients [21].

There has been a growing call for genetic education in medical schools to focus on common aspects of genetics and their applications to clinical practice [22,23]. In our own experience (Neil Lamb [HudsonAlpha Institute for Biotechnology] served as the director of the Medical Genetics course for the Emory University School of Medicine [GA, USA] from 1999–2006), this transition involves shifting

the curriculum from a series of basic science lectures focused on genetic research in rare diseases to a collection of case-based vignettes introducing genetics from a variety of clinical specialties and its application to diagnosis and treatment. Like other medical schools, internet-based genetic databases and role-playing were introduced [24,25], providing medical students with the opportunities to apply the knowledge they have learned. These efforts were met by students with appreciation and a rising sense of the importance of genetics in their future practice. Most recently, similar to the efforts of medical schools across North America [26], the School of Medicine at Emory University underwent a comprehensive restructuring of the entire undergraduate medical curriculum, moving towards a more integrated model. Among other modifications, an intensive short course in genetics was placed at the beginning of the first year to lay the groundwork for the role of genetics in health and disease. Additional genetic concepts and applications were incorporated across the remainder of the undergraduate medical experience. Unfortunately, the integration of genetic concepts across the medical curriculum is rare, and only a few comprehensive programs (such as those at the University of California San Francisco [CA, USA] and Duke University [NC, USA]) exist. In addition, integrating genetics into medical education must extend beyond the classroom experience and into the clinic; otherwise, the concepts lack application to the everyday practice of medicine. Such system-wide curriculum revision is difficult to enact, especially because it requires the redistribution of content emphasis and teaching assignments. These changes often disrupt long-standing roles and responsibilities among the teaching faculty. Strong support from the school leadership is critical to obtaining faculty buy-in [27 AND NEIL E LAMB, HUDSONALPHA INSTITUTE FOR BIOTECHNOLOGY, AL, USA. PERS. EXPERIENCE]. Without this guidance, curricular reform can devolve into a battle for turf as various departments resist what they perceive as a loss of influence or importance within the curriculum.

Once physicians are actively practicing medicine, where can they turn for updates on genomic testing and its integration into the clinic? One of the largest providers of continuing medical education (CME) for health professionals, MedscapeCME (NY, USA), has only one CME activity explicitly focused on genomics and personalized medicine for both physicians and nurses [116]. This activity was available for

a period of 1 year, but as of this writing it is no longer available for credit, although it can still be read on the MedscapeCME site. Similarly, the American Medical Association did not, as of August 2009, have any CME activities relating to genetics on their website, but they posted a link through to the NCHPEG site instead. Through December 2011, the March of Dimes will offer CME credit for three modules on 'Genetics and Your Practice', but genomics does not appear to feature in these tutorials. The CDC Office of Genomics and Public Health maintains a list of genomics resources that may prove helpful [117], but, like most of these activities, requires motivation and filtering on the part of those seeking the information.

We are excited to note that in August 2009, Dr Francis Collins was appointed as the Director of the NIH. Given his tireless efforts on behalf of the Human Genome Project and the National Human Genome Research Institute (NHGRI [MD, USA]), we and many others hope that genetics and genomics education will be a focus of the NIH throughout his term.

Media

It is self-evident that the media play a major role in the public's understanding of personalized genomics. Many talented science journalists have invested time and energy into keeping up with the fast-moving developments and maintaining as much scientific accuracy as possible in their stories. They are under unrelenting pressures, not least of which are the financial difficulties relating to print and online media. They are also at the mercy of administrative whims, such as the closing of the entire science reporting department at the news network CNN (GA, USA) in December 2008. The impact of these decisions runs counter to the growing complexity of science and environmental issues that require thorough and considered media coverage and explanation. Although many science journalists do not have a formal background in science, masters degree programs in science journalism specifically have become more common in the last decade. A recent publication regarding the outcome of an interdisciplinary meeting on the topic, primarily sponsored by Canadian organizations, suggested that we specifically need to develop more training in science policy for journalists, allowing for the discussion of more complex issues facing us all [28].

A central problem in communicating science lies in the almost opposite approaches of scientists and journalists in conveying their work.

Scientists need to realize that while they are often trained to read an entire article and to look for the data to support the claims made, newspaper or web article readers are more likely to stop reading or to 'drop off' as a story proceeds from the headline to delving into the actual data. Therefore, journalists are trained to write in a completely different style from that of scientists, packing the attention-grabbing details into the very beginning of the story rather than setting the stage through background information and the explanation of methods. Similarly, journalists do not have the space to cite previous references (too often, stories do not even include the primary reference for the actual paper being discussed). Combining this with the fact that headline writers may not be the authors of the actual story means that news articles often begin with an enticing headline, even if that heading is not fully supported by the body of the article. So why is this a problem? Many people now exclusively get their news from internet sources and only see the headline on a Yahoo or Google reader. One explicit example is a 2 September 2008 piece from the website *Science Daily*, where the headline read, 'Infidelity gene? Genetic link to relationships found' [118]. The research study being reported on tested for an association between one polymorphism in an arginine vasopressor receptor gene that was demonstrated to be involved in pair bonding in voles, and possibly in human male relationship behavior. Despite that headline, it is only in the third paragraph that we learn 'the effect of this genetic variation is relatively modest, and it cannot be used to predict with any real accuracy how someone will behave in a future relationship'. But on this story's website, at the time we accessed it in August 2009, there was an advertisement for a genetic testing company right next to the text (along with, to be fair, advertisements for baldness cures and autism support).

Another good example of hotly debated media coverage is the CNN story from 5 August 2009, 'In China, DNA tests on kids ID genetic gifts, careers' [119]. The story covers a camp in China where, for the equivalent of US\$880, 'about 30 children aged 3–12 years old and their parents are participating in a new program that uses DNA testing to identify genetic gifts and predict the future'. While that idea is controversial enough, the story does not tell the reader which specific tests are being administered or what traits are being measured. Instead, readers learn that 'the test is being offered to children in China to help discover their natural talents'.

Readers also learn of rumors suggesting that Chinese basketball star, Yao Ming, may have been bred from two basketball star parents on purpose. But the readers do not learn anything about the reliability of genetic testing for height – at present, over 50 different regions of the genome have been implicated and together they contribute 5% or less of the genetic variance in height. In fact, a recent study demonstrated that the 'Victorian method' of measuring the height of the two parents and roughly averaging them was able to predict 40% of the genetic variance [29], an order of magnitude that is better than state-of-the-art genomics! However, this has not stopped genome-scan companies from including 'height alleles' in their DNA arrays.

In 2007, Celeste Condit of the University of Georgia (GA, USA) offered a very useful perspective titled: 'How geneticists can help reporters get their story right' [30]. She points out that 'the unwillingness of journalists to try to teach the lay public the word allele' contributes to the public's confusion regarding genetic variation results being reported so frequently these days. Her studies demonstrate that the phrase 'version of a gene' is more understandable to community advisory boards and focus groups. Understanding the concept of alleles is crucial to understanding the ideas of risk prediction based on GWAS for complex traits or diseases. This gap between scientific terminology and public understanding featured prominently in 2008–2009 as a number of media outlets paid for reporters to have genome scans (or the companies in question donated the scans). A spate of articles appeared in which journalists or scientists wrote about trying to make sense of their genome-scan results, and the risk they were told they had for developing certain diseases based on their allele combinations. The reports reflect more tension between scientists and journalists – scientists generally focus more on the aggregate genomic information provided by GWAS, while journalists are trained to humanize topics by using one individual's experience as the focus of the story. One such story in the UK newspaper, *The Guardian*, [120] chronicled the DTC test experience of journalist James Randerson, who paid GBP£825 to the company Genetic Health (London, UK) for testing in only 42 genes, and a personalized consultation regarding the results. When he presented the results to "five leading experts...[t]hey describe the predictions and advice from Genetic Health as 'poor', 'flawed', 'misleading' and 'baloney'". This may be amusing for geneticists to read, but leaves members of the public only more confused.

They might reasonably ask – how can the media report weekly on the discovery of a new gene for some condition or trait, and yet five experts call a consumer genetic test ‘misleading’ and ‘baloney’? Are genomic results useful in medicine or aren’t they? And how is the average consumer supposed to know what to believe?

We and many others suggest that training of the media to report on scientific results is absolutely crucial in preparing both the public and physicians for incorporating genomics into personalized medicine. As we discussed above, the advent of social media can help – one example, a website called ‘Help a reporter out’ [121], allows journalists to ask queries regarding registered sources. More geneticists could sign up through this site to provide scientifically sound advice to members of the media; the service also asks for potential sources through Facebook and Twitter (@helpareporter). The ASHG lists a number of resources for the media to use [122], although admittedly, some of these are simply links back to the American Medical Association or to older presentations and need to be updated to include more information on genomics specifically. The ASHG also offers a list of ‘expert speakers’, primarily from the society management and elected officials of the society, who will speak to the media. Journalists can also find some resources on genetics or genomics writing via their societies, including the National Association of Science Writers (with local chapters, such as the one in Washington DC, USA, being very active); the Council of Science Editors; or the World Conference of Science Journalists, which in 2009 was covered extensively in real-time on Twitter and was summarized via podcast [123].

Conclusion & future perspective

Geneticists have to make their peace with the availability of DTC genetic testing, and work with other communities to provide as much information as possible to the public. Aesop’s fable, ‘The Lioness’, describes a debate among the animals as to which of them deserved the

most credit for producing the greatest number of children at birth. When the animals turned to the Lioness, she exclaimed with a laugh, ‘Why! I have only one, but he is the King of the beasts’. In other words, the value is in the worth, not in the number. So it is when we consider genome scans and sequencing – hundreds of thousands of genetic markers may be examined, but ultimately, it is those variants that influence disease risk in a clinically relevant manner that are of the most value to the public. The scientific challenge in identifying those key variants is matched by the challenge in communicating their relationship with disease (and a host of other ethical, legal and social issues that we have only been able to touch on here). In the future, it is likely that one’s personal genetic liability will be recognized as being as important as blood pressure, BMI and cholesterol. We will all be offered testing for genetic variation on a regular basis within 5–10 years. It is also likely that pharmacogenomic results will become more commonly incorporated into the standard dosing of drugs and will be applied in both diagnostic and prognostic tests for all manner of complex diseases. To reach this goal, well-reasoned educational efforts – free of jargon and easily understood – must be developed for three audiences: the public, the physicians that treat them and the writers of the media we all consume. As we have detailed, several strong first steps have been taken in this direction, but even greater efforts must be made in order for the conversations to move more fully into the public consciousness.

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The authors have no relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in the manuscript. This includes employment, consultancies, honoraria, stock ownership or options, expert testimony, grants or patents received or pending, or royalties.

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Executive summary

- Direct-to-consumer (DTC) genetic testing has emerged on a large scale in the last few years and is likely to be around for many more.
- Nonscientists who are interested in DTC genetic testing will turn to a number of sources for more information, including many forms of media and the advice of health professionals.
- Most physicians are not properly informed about genetic testing, particularly newer genome-scan studies and the difficult work of reforming medical school curricula must be undertaken to make this happen.
- Journalists have different aims for conveying information than scientists do, and thus, stories on genome-scan results and their meaning for health applications may leave the public confused.
- A number of resources do exist for education on genomic tests, including internet sites and social media, and these can serve as a base for developing user-friendly and understandable tutorials on personal genomics.

Bibliography

Papers of special note have been highlighted as:

▪ of interest

▪▪ of considerable interest

- 1 Ray T: Research shows public eager for genetic Dxs but confused how to interpret results. *Pharmacogenomics Reporter*, 13 August (2008).
- 2 Wilde A, Meiser B, Mitchell PB, Schofield PR: Public interest in predictive genetic testing, including direct-to-consumer testing, for susceptibility to major depression: preliminary findings. *Eur. J. Hum. Genet.* (2009) (Epub ahead of print).
- 3 Kolor K, Liu T, St Pierre J, Khoury M: Health care provider and consumer awareness, perceptions, and use of direct-to-consumer personal genomic tests, United States, 2008. *Genet. Med.* 11(8), 595 (2009).
- 4 Evans J, Green R: Direct to consumer genetic testing: avoiding a culture war. *Genet. Med.* 11(8), 568–569 (2009).
- 5 Venter JC: *A Life Decoded: My Genome, My Life*. Penguin Press Science, London, UK (2007).
- 6 von Ward P: *The Soul Genome: Science and Reincarnation*. Fenestra Books, AZ, USA (2008).
- 7 Yashon R, Cummings M: *Human Genetics and Society*. Brooks/Cole, KY, USA (2008).
- 8 Reilly P: *The Strongest Boy in the World: How Genetic Information is Reshaping Our Lives*. Cold Spring Harbor Laboratory Press, NY, USA (2006).
- 9 Gibson G: *It Takes a Genome: How a Clash Between Our Genes and Modern Life is Making US Sick*. FT Press, NJ, USA (2008).
- 10 Willard H, Haga S: *The Book of Genes and Genomes*. Springer, Berlin, Germany (In press).
- 11 Collins F: *The Language of Life: DNA and the Revolution in Personalized Medicine*. HarperCollins, NY, USA (In press).
- 12 Trinidad SB, Fryer-Edwards K, Crest A, Kyler P, Lloyd-Puryear MA, Burke W: Educational needs in genetic medicine: primary care perspectives. *Community Genet.* 11(3), 160–165 (2008).
- 13 Freedman AN, Wideroff L, Olson L *et al.*: US physicians' attitudes toward genetic testing for cancer susceptibility. *Am. J. Med. Genet.* 120A(1), 63–71 (2003).
- 14 Hudson K, Javitt, G, Burke W, Byers P; ASHG Social Issues Committee: ASHG statement on direct-to-consumer genetic testing in the United States. *Am. J. Hum. Genet.* 81, 635–637 (2007).
- **Important reference from the Society of Human Geneticists.**
- 15 Ge D, Fellay J, Thompson AJ *et al.*: Genetic variation in *IL28B* predicts hepatitis C treatment-induced viral clearance. *Nature* 461(7262), 399–401 (2009).
- 16 Ohata T, Tsuchiya A, Watanabe M, Sumida T, Takada F: Physicians' opinion for 'new' genetic testing in Japan. *J. Hum. Genet.* 54(4), 203–208 (2009).
- 17 Avar D, Knoppers B: Genomic medicine: considerations for health professionals and the public. *Genome Med.* 1(2), 25 (2009).
- **Thoughtful commentary on the many difficulties in genetics education for two of the groups we describe.**
- 18 Scheuner MT, Sieverding P, Shekelle PG: Delivery of genomic medicine for common chronic adult diseases: a systematic review. *JAMA* 299(11), 1320–1334 (2008).
- 19 Thurston VC, Wales PS, Bell MA, Torbeck L, Brokaw JJ: The current status of medical genetics instruction in US and Canadian medical schools. *Acad. Med.* 82(5), 441–445 (2007).
- 20 Greb AE, Brennan S, McParlane L, Page R, Bridge PD: Retention of medical genetics knowledge and skills by medical students. *Genet. Med.* 11(5), 365–370 (2009).
- 21 Telner D, Carroll J, Talbo Y: Genetics education in medical school: a qualitative study exploring educational experiences and needs. *Med. Teach.* 30(2), 192–198 (2008).
- 22 Association of American Medical Colleges: *Report IV – Contemporary Issues in Medicine: Genetics Education*. Association of American Medical Colleges, DC, USA (2004).
- 23 Salari K: The dawning era of personalized medicine exposes a gap in medical education. *PLoS Med.* 6(8), E1000138 (2009).
- 24 Waggoner DJ, Martin CL: Integration of internet-based genetic databases into the medical school pre-clinical and clinical curriculum. *Genet. Med.* 8(6), 379–382 (2006).
- 25 McIlvried DE, Prucka SK, Herbst M, Barger C, Robin NH: The use of role-play to enhance medical student understanding of genetic counseling. *Genet. Med.* 10(10), 739–744 (2008).
- 26 Irby DM, Wilkerson L: Educational innovations in academic medicine and environmental trends. *J. Gen. Internal Med.* 18(5), 370–376 (2003).
- 27 Muller JH, Jain S, Loeser H, Irby DM: Lessons learned about integrating a medical school curriculum: perceptions of students, faculty and curriculum leaders. *Med. Educ.* 42, 778–785 (2008).
- 28 Bubela T, Nisbet MC, Borchelt R *et al.*: Science communication reconsidered. *Nat. Biotechnol.* 27(6), 514–518 (2009).
- 29 Aulchenko YS, Struchalin MV, Belonogova NM *et al.*: Predicting human height by Victorian and genomic methods. *Eur. J. Hum. Genet.* 17(8), 1070–1075 (2009).
- 30 Condit C: How geneticists can help reporters to get their story right. *Nat. Rev. Genet.* 8(10), 815–820 (2007).
- **Great explanation for scientists on how they should prepare for and then interact with the media.**
- 31 Uhlmann WR, Guttmacher AE: Key internet genetics resources for the clinician. *JAMA* 299(11), 1356–1358 (2008).
- **Excellent reference for pertinent websites.**
- **Websites**
- 101 Facebook
www.facebook.com
- 102 Twitter
www.twitter.com
- 103 Genetics & Public Policy Center: *U.S. Public Opinion on Uses of Genetic Information and Genetic Discrimination* (2007)
http://www.dnapolicy.org/resources/GINAPublic_Opinion_Genetic_Information_Discrimination.pdf
- **One of the only large-scale studies on this topic.**
- 104 WebMD
www.webmd.com
- 105 Genetic Alliance
www.geneticalliance.org
- 106 Trust It or Trash It?
www.trustortrash.org
- **Very useful site for the lay public to evaluate other resources providing information about genetics (or any science really).**
- 107 Genetics and Public Policy Center
www.dnapolicy.org
- 108 Scitable
www.nature.com/scitable
- 109 PHG Foundation
www.phgfoundation.org/
- 110 MySpace
www.myspace.com
- 111 TwitterCounter
www.twittercounter.com
- 112 Vorhaus D and Moore L: What happens if a DTC genomics company goes belly up? *Genomics Law Report* (2009)
www.genomicslawreport.com/index.php/2009/09/18/what-happens-if-a-dtc-genomics-company-goes-belly-up/

- 113 National Coalition for Health Professional Education in Genetics: *Core Competencies in Genetics for Health Professionals (3rd Edition)* (2007)
www.nchpeg.org/core/Core_Comps_English_2007.pdf
- **Good for health professionals to know what their expectations are in genetics.**
- 114 US Centers for Disease Control and Prevention: Genomics translation, genomic workforce competencies 2001 (2007)
www.cdc.gov/genomics/translation/competencies/index.htm
- 115 US Food and Drug Administration: FDA News Release, FDA approves updated warfarin (Coumadin) prescribing information (2007)
www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/2007/ucm108967.htm
- 116 Ginsburg GS: A primer on genomic and personalized medicine: how will it affect your practice? MedscapeCME (2009)
<http://cme.medscape.com/viewarticle/570861>
- **A well-written tutorial for physicians.**
- 117 US Centers for Disease Control and Prevention: Genomics resources (2009)
www.cdc.gov/genomics/resources/index.htm
- 118 Karolinska Institute: Infidelity gene? Genetic link to relationship difficulties found. ScienceDaily (2009)
www.sciencedaily.com/releases/2008/09/080902161213.htm
- 119 Chang E: In China, DNA tests on kids ID genetic gifts, careers, CNN (2009)
www.cnn.com/2009/WORLD/asiapcf/08/03/china.dna.children.ability/
- 120 Randerson J: The day I had my genes tested. The Guardian (2008)
www.guardian.co.uk/lifeandstyle/2008/dec/09/healthandwellbeing-medicalresearch
- 121 Help A Reporter Out
www.helpareporter.com/haro/index.php?r=story/create
- 122 American Society of Human Genetics: Reporting genetics news: resources for the media
www.ashg.org/press/news_genetics.shtml#2
- 123 World Congress of Science Journalists (2009)
www.wcsjnews.org/