



What ELSI is New?
Genomics Law Report
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What ELSI was New? Plenty.

From October 5 to December 8, 2009, the Genomics Law Report featured a series of thirty-six guest commentaries by industry, academic and thought leaders in the fields of genomics and personalized medicine. Entitled **What ELSI is New?**, the series asked each contributor to briefly respond to the following question: “*What do you believe is the most important ethical, legal or social issue (ELSI) that must be addressed by the fields of genomics and/or personalized medicine?*”

For better or worse, that’s where the instructions ended. The invited contributors identified the ELSI of their choice and discussed (or not) their rationale for so selecting as they saw fit. In addition to refraining from substantive editing, we intentionally avoided coordinating commentaries. Although we encouraged independent submissions from a variety of contributors and deprived them of any advance knowledge of what others in the series would say, one of our hopes was that consensus would begin to form around certain key ethical, legal and social issues.

To some degree this occurred. In collecting the series for the convenience of readers who would like to have all of the contributions in one place, we have ultimately settled on six broad topic headings for the commentaries, which are preceded by Jason Bobe’s call to arms for a new generation of “genomic astronauts.” **It’s Mine!** focuses on the privacy, ownership and access questions that continue to swirl around genomic information. **Personalized Medicine in the Real World** is wide-ranging, with commentaries that examine existing societal, scientific and governmental barriers to the implementation of personalized medicine, and several that propose specific solutions designed to eliminate certain of those barriers. In **Too Much Information**, the commentaries return to data and consider how individuals, clinicians, researchers and, ultimately, society will assimilate the coming deluge of personal genomic information. **Back to School** features several commentaries that make the case that improved educational models are the key to realizing the potential of genomics and personalized medicine. The commentaries in **No _____ Need Apply** focus on one of the most oft-discussed risks associated with personal genomic information—genetic discrimination. Finally, our commentators take a look to the future in **Testing the Limits?**, examining issues of genetic testing, modification and exceptionalism.

Although we have presented the series using these broad headings, as we undertook the task of gathering the commentaries and attempted to identify cross-cutting themes and trends, we came to appreciate even more the value of the series. That is, the commentaries simply do not fit into neat boxes. That was probably to be expected, given the variety and thoughtfulness of our contributors and the breadth and uncertainty that continues to surround the fields of genomics and personalized medicine.

Despite our organizational efforts, each of our contributors could easily have his or her own topic heading. And more than anything else, the diversity of ideas and opinions expressed in *What ELSI is New?* is its most significant contribution. After all, our other hope for the series was that the broad range of contributors would hold up seemingly familiar issues in new lights and see new connections. They sure did.

Dan Vorhaus
Editor, Genomics Law Report
December 2009

To the moon: In support of the genomic astronauts who will take us there.

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Jason Bobe](#), [Personal Genome Project](#).

At the frontiers of science and engineering, promising new technologies are becoming available that will help us address pressing problems in human health and well-being. As the recent [history of personal computing](#) has shown, technology is often the easy part. Once into the world, technologies tend to go careening down the well-beaten path toward “better, faster, and cheaper” on a journey that leads ultimately to everyone’s front door. Personal genomic technologies are no different. If you don’t have any DNA sequence of your own yet, you will soon and so will many of your family, friends, and neighbors (and [pets](#) too).

Making personal genomes useful is a much more formidable challenge. In medicine, we want to employ personal genomics in the development of therapies that eliminate disease and diagnostics that reduce illness through early detection or prevention. In our personal and family affairs, we want knowledge that enables us to lead fuller lives, to know how our own personal biology interacts with the varied environments and lifestyle choices that makes us who we are and connects us with others.

Low-cost sequencing technologies take us one small step toward achieving such translational goals, but a giant leap remains: connecting personal genomes with personal phenomes. And that, as David Houle [reminded us](#) recently, is why we got into this business in the first place:

“We did not begin to study genomes because we care about genotypes; we study genomes because we care about phenotypes, the health and well-being of humans and the diversity of life on Earth. Now is the time to begin to take the study of the phenotype as seriously as we take the study of the genotype. We must number, locate, and measure even the hairs of our heads, the details of the phenotype, so that we can understand which of those details matter.”

To get beyond databases comprised solely of disembodied DNA sequences, we will need the help of individuals who are willing to open up their personal lives and to share the details of their medical histories, physical traits, behaviors, and other phenotypes.

These individuals are the [astronauts of our era](#). By sharing their genomes and phenomes and making them broadly available through participation in [public genomics](#) research studies like the [Personal Genome Project](#) (PGP), these pioneers will radically accelerate our ability to explore new frontiers of human knowledge. In doing so, they face potential risks. They are putting it on the line for our benefit and for the benefit of future generations. They deserve our support. To the moon!

Chapter 1

It's Mine!

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To Share or Not to Share: That is the Question.

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Catherine A. McCarty, Marshfield Clinic Research Foundation](#).

I believe that the most important ethical, legal or social issue (ELSI) that must be addressed by the fields of genomics and/or personalized medicine is return of genetic results to research participants. Biobanks attached to electronic medical records are becoming very common and, as demonstrated through the [eMERGE network funded by NHGRI](#), are a cost- and time-effective approach to genetic discovery.

Where subjects have been actively consented to participate in a biobank, they are usually told that they will not have personal genetic results returned to them. The reason for this is two-fold: 1) the research is not being conducted in a CLIA-certified lab, and 2) research results would need to be verified in other labs before being considered to be potentially clinically relevant. The inability to act on results, e.g. there is no cure or treatment, also weighs into the decision not to return research results.

This approach worked well in the era of candidate gene and early genome-wide association studies. As genotyping costs decrease such that it may be possible to sequence the entire genome for \$1000, researchers will find that they have clinically relevant data for research subjects who were initially consented under the assumption that genetic results would not be returned to them.

Furthermore, as research findings are confirmed in other laboratories, results that were initially tentative may become truly significant and ideally clinically relevant. Researchers will then need to consider the ethics of withholding information known to be clinically relevant versus the fact that some subjects may have initially chosen to participate because they would not have information returned to them.

In addition to these basic ethical questions, the issue about how to adequately inform subjects about the meaning of the genetic results must be considered. It would be irresponsible to provide genetic information without appropriate genetic counseling. ELSI needs to keep pace with genetic technologies applied to biobanks.

It's my genome: should researchers be obliged to return genetic data to research participants?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Daniel MacArthur](#), [Wellcome Trust Sanger Institute](#) and [Genetic Future](#).

Alice signed up as a “healthy control” for a research project into the genetics of type 2 diabetes. During the project, researchers identified a variation in Alice's BRCA1 gene that is known to be associated with a high risk of breast cancer. Alice is unaware that she carries this variant, and if she was told about it she would be able to take steps to minimise her risk of cancer.

It is clearly in Alice's best interests to be given the option to be informed about this discovery – and yet in most research studies she would have no such opportunity. Instead, Alice is likely to have signed an informed consent form advising her that she will not receive any findings from the research study, and that indeed she has no automatic right to access any of the data generated from her DNA during the project.

As genetic research studies move into the era of whole-genome sequencing, and as cohorts of patients and controls grow ever larger, the frequency with which researchers uncover such medically relevant “incidental findings” will increase sharply.

Returning incidental findings poses major challenges for researchers: it requires disrupting well-established protocols for informed consent and subject anonymisation, and establishing new frameworks for responsible data return and counselling. Yet the alternative approach – withholding medically useful (or even simply intellectually interesting) information from research subjects even if they request it – is ethically problematic. In the absence of convincing evidence that disclosure of results causes harm, I would argue that the default position should be that research participants have complete access to their own genetic data if they request it.

In addition to the ethical imperative to return medically actionable data to participants, open return policies may well prove to have non-trivial practical benefits: the promise of receiving some tangible benefit from participation is likely to improve recruitment and retention rates.

In Support of Open Access for Genomic Research

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [George Church](#), [Harvard Medical School](#).

One of the recurring themes in this [ELSI series](#) has been the discussion of open-access vs. research-only models for genomic research (see [Bobe](#), [MacArthur](#), [McCarty](#), [Prainsack](#) and [Sweeney](#)). Below I discuss the characteristics and advantages of, as well as obstacles to, an open-access data model for genomic research.

A. Self-access: Open-access and freedom of information are increasingly required by law. Medical research is increasingly holistic -- integrating a variety of (identifiable) traits and molecular signatures. Genomics is just part of this, not particularly exceptional. Multi-purpose cohorts and biobanks are displacing single trait studies. Research volunteers are increasingly expecting to see their own data and what is being done with it. So with respect to such desired transparency, projects can be classified as ranging from 1) "no access" ([HapMap](#), [1000 Genomes](#), [dbGAP](#)), to 2) limited access and no vetting exam ([ClinSeq](#), [CPMC](#) and [REVEAL](#)), to 3) full access based on obtaining a 100% score on an exam covering risks of data sharing and re-identification ([PGP](#)).

B. Sharing: Since individuals can now easily get their medical and genomic data in digital form (outside of their actual "medical records" or any research project), and since individuals can have motivations to share these data, we can let this happen with or without scientific / non-profit / IRB guidance. If we choose the "without" route, then we will likely see Facebook / for-profit / non-IRB "[DTC genomic research](#)" proliferate. Projects that choose the "with" route, such as PGP, aim to set higher standards for how much knowledge citizens demonstrate about genetics and research before they give or receive data. The risks for both individual and society of sharing data are likely lower than many occupations (e.g. police and taxi) and possibly lower than the risks of "not sharing," but those [risks still need to be communicated and appropriate guidance provided](#).

C. Science: Access to information can be restricted via fees, legal threats, technological censoring (e.g. GPS and encryption algorithms), and study design ([eliminating useful data linkages](#)). What has been the impact of such restrictions on science, on serendipity, collaboration, interdisciplinary research, etc. in the past? Will computer experts, (with artists, writers, etc.) create user interfaces, de-mystifying huge case-control studies in open-access systems or in closed? Will physicists and chemists make whole systems biology models if they can only see part of the data (or none of it)? Will social scientists (with ethicists and policy experts) discover alarming (or hopeful) trends in open-access systems or closed? Do we really know in advance who will contribute and who will not? Will we prioritize access based on willingness to jump through bureaucratic hoops? Is that likely to maximize the number of

creative interdisciplinarians or produce the [biggest out-of-the-box analytic breakthroughs](#)? This is not about mere inconvenience, it is about a series of totally missed opportunities. The predictable positive impact of open-access is huge, and add to that impacts far beyond what we can currently predict.

D. Politics:

1) *Retroactive activism*: One could argue that current case-control cohorts and biobanks have enough momentum that nothing new can compete. But monuments do topple. If enough volunteers request / demand their data, then [there may be pressure to give it to them, no matter what the original contract said](#). Any [claim that the data or cells are de-identified will be untenable](#), since past volunteers can inexpensively provide DNA identifiers (say 100 SNPs).

2) *Proactive*: More importantly, going forward, larger biobanks and cohorts will likely be the most useful and [new recruits may increasingly migrate to the most transparent and scientifically exciting projects](#).

3) *Reactive*: The press and the public will react to efforts that permit people to publicly share their own data; but any criticism is likely to be much less severe than the backlash following the [accidental \(or intentional\) release of multiple volunteers without their permission](#). Keeping secret data about people that they cannot access will perpetuate distrust of science. In contrast, celebrating volunteers willing to become informed and share their medical information might [inspire the public in a manner like astronauts in the 1960s](#).

Privacy & Ownership of an Individual's Personal Genetic Information

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by Jennifer Sweeney, [Knome, Inc.](#)

As personal genetic information becomes increasingly accessible and affordable, the ownership and privacy of such data will emerge as a central issue in genomics. Is personal genetic information, stored within a centralized database where a third-party gatekeeper determines who has access, really still private? Who owns such data? Should the individual have control over how, when and where their information is used? In such cases, it certainly seems the individual may have forfeited ownership and rights to control access to their own data.

Many of today's genomics companies are compiling vast databases of genetic data, phenotype information and medical histories obtained from their clients. Two important characteristics of genetic information make issues of privacy and ownership especially important – the durability of such data and its relevance to family members. An individual's genetic information does not change over time, so once it is disclosed there is no reclaiming it. Further, we share substantial portions of our genetics with family members – choosing to grant access to our own data impacts present and future family members without their consent.

As yet, industry and regulators have placed little focus on ownership and privacy concerns. GINA addresses discrimination once an employer or insurer already has access but is silent on who actually owns the genetic information. For the most part HIPAA's privacy protections do not apply because DTC genomics companies are not "covered entities" under the privacy rule. Corporations, namely DTC genomics companies, have become the default guardian of personal genetic information.

Surely we all benefit from individuals electing to share their genetic information with others – it is the basis for how future genetic discoveries will be made. However, granting unfettered access or forfeiting ownership of inherently private, personal genetic information exposes an individual in ways not yet fully understood. Careful consideration should be given to the best ways to ensure genetic information remains private, owned and controlled by the individual.

“So... One Can Have Their Complete Genome Sequenced...Should I?”

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Patrice M. Milos](#), [Helicos BioSciences Corporation](#).

I've had a fascinating time reading these posts and it has taken me awhile to articulate a question I imagine many of you might, or will, have. I believe the above question will be front and foremost on people's minds in the next two to three years. As genome sequencing costs continue to fall dramatically, we will arrive at our end goal of the \$1000 genome... or perhaps even \$100... shortly.

Yet it is just this one question which raises many more ELSI questions.

First and foremost – do I have a right to my genome sequence?

I have a fundamental belief that the answer is a resounding yes. Why should I have to worry about whether I can obtain the sequence of genes that may actually prove important for key decisions for me as well as my family?

Once this question is answered a key question on my mind is how can I ensure my sequence is accurate? What is the standard that I am to accept if I am to make sense of my sequence? Who will be responsible for ensuring the level of acceptable accuracy?

In all honesty I am still grappling with this question – is one in a million, one in a billion error rate acceptable? What if my one error suggests a mutation in a cardiovascular gene which predisposes to myocardial infarction? What if the error is in a drug metabolism gene resulting in the inactivation one of the genes? What if the error is in BRCA1? If there were an area where regulations could actually prove important, the ability to ensure your sequence is accurate is key.

Let's assume we obtain near flawless consensus accuracy – what decisions will I be able to make based on my individual genome variation?

We are at an early stage of making sense of the sequence of human genomes but the pace of knowledge is rapidly escalating. I remain confident as we accurately sequence hundreds to thousands of genomes that we will have a much better understanding of the unfolding story of our genome. This will come as the technological revolution around us continues.

Finally, how will I and my doctor use my genome to make my healthcare better?

This to me is the hardest question – sure we have examples but they haven't touched me yet. Is our healthcare system ready to fully realize the value of our genome – not today, but let's hope all our efforts will pave the way to the era of personalized healthcare.

I must say, I have not made my own decision whether or not I will sequence my genome and I am just not sure why I remain guarded. Perhaps those of you who know me might imagine that I would not want anyone to understand what secrets my genome holds and what it is that makes me unique... although as our technology matures at the same pace as our science I may just have to find out why as a female I am colorblind and why I have a funny toe and why I remain an eternal optimist. After all you still can't fully understand a person by their genome sequence alone.

Good luck to each of you in making your personal decision in the years ahead. I'll let you know when I am ready.

The participatory turn in medicine – Which letter in the alphabet?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Barbara Prainsack](#), [King's College London Centre for Biomedicine & Society](#).

Leroy Hood recently predicted the emergence of 'P4 medicine – predictive, personalized, preventive and participatory'.¹ Particularly the final P in 'P4' seems to hit a nerve: Craig Venter already hailed the 'democratization of genomics'² as part of a participatory turn in medicine, and 23andMe launched a 'Do-It-Yourself revolution' in disease research.³

It is indeed a welcome development that growing numbers of people can access genetic and other health information (personalised and otherwise) relatively easily, and that specialised medical knowledge is no longer the prerogative of those with a professional education. (The blurring of the divide between 'lay people' and professional experts, which currently takes place in personal genomics, arguably accounts for some of the latter's concern about this newly emerging market.) But the participatory turn in medicine is also indicative of an ongoing individualisation of responsibility in health care⁴: The more knowledge we can obtain, the more we will be expected to obtain, and to pay for.

If we get sick when we could have prevented it, social and financial costs are often the result. It is one of the most challenging, but also most crucial tasks of ELSI research to ensure that people's gain in power and agency will not be outweighed by the 'gain' in responsibility, new health duties, and blame. Taking the tenets of 'P4 medicine' seriously means that we should learn from people's experiences and expectations – and 'people' is not restricted here to those who already participate. Otherwise we might get R4 instead of P4: a kind of medicine which is limited to those of us who are responsible, resilient, rich, and RSS-fed.

¹ Leroy Hood. [A Doctor's Vision of the Future of Medicine](#). Newsweek (July 13, 2009).

² Craig Venter in an [interview with the San Francisco Chronicle](#), June 11, 2009, pE1.

³ 23andMe blog [Spittoon](#) on July 7, 2009.

⁴ Rose, N. *The Politics of Life Itself: Biomedicine, Power, and Subjectivity in the Twenty-First Century*. Princeton University Press (2006).

Chapter 2

Personalized Medicine in the Real World

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The Direct to Consumer Disconnect: Why the genomics community is going to have trouble talking to patients and doctors alike

*This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Matthew Herper](#), *Forbes*.*

We can sequence DNA cheaper and faster, and even deal with all the data.

But if genomics is really going to impact medicine, we're going to have to start bridging the gap between the companies and scientists doing this early work and the traditional medical establishment. The genoscenti like to talk about how everyone should have access to his own DNA. Your genes are a part of you, they say. How could they belong to anyone else?

But that isn't how medicine has worked. I need a doctor as a go-between to find out my LDL or CRP or even what copy of the BRCA gene I have.

That's not to say that things shouldn't change. But bridging this yawning gap is going to be the big social challenge of genomics.

23andMe's big contribution has been to start this conversation, but we're still a long way from figuring out how genomics will fit into medical culture, no less into the regulatory framework. And how and whether innovation gets paid for will depend very much on those cultural and regulatory structures.

It's easy to say medicine and government have to change. But the reality is that they will do so only slowly. Right now, the big challenge for people in genomics may be to start thinking about how to silently wind their way into the existing medical culture.

Personalized Medicine: what is the added value to healthcare systems?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Katherine Payne](#), [The University of Manchester](#).

The Issue

Personalized medicine can potentially offer benefits to patients by targeting the safe and effective use of medicines by using DNA-based 'companion' diagnostic tests to inform prescribing decisions. These potential benefits are not costless. To understand the added value of personalized medicine it is necessary to identify and quantify the true costs and benefits of introducing such companion diagnostics and medicines into healthcare systems.

Response

My *raison d'être* as a health economist is to apply the concept of opportunity cost, which drives health policy-makers to consider the benefits forgone if one healthcare intervention is chosen as a treatment over the other available options. My challenge is to provide information about how to allocate scarce healthcare resources such that maximum patient benefit is obtained from every pound (dollar/euro) spent. One aim of personalized medicine is to target medicines only to patient populations that will derive a positive benefit (health improvement). In theory, it is the perfect solution to this challenge of getting value for money. Targeting (or personalizing) medicines in this way should stop scarce healthcare resources being wasted. The reality is that we know companion diagnostics are not 'perfect' and such tests provide 'probabilistic information' to prescribers.

The current regulatory climate for DNA-based diagnostics expects manufacturers to provide information on the analytical accuracy of the test. However, this information does not tell healthcare policy-makers about the added value of linking a medicine with a companion test. To be clear whether personalized medicine does add value it is necessary to understand the difference in costs and benefits of using a companion diagnostic compared to current prescribing practice. The benefits should be measured in terms of whether using the test to target the medicine does show increased health gain for the population of patients who are prescribed the medicine. Depending on the chosen viewpoint of the evaluation, the costs are those for the healthcare system but may also include patient costs. Evidence must therefore be collated, using robust research methods, on how the test affects the referral of patients to care pathways, subsequent services and treatments compared to the status quo. An important challenge remains. How, and who, to fund the research to generate sufficient evidence for policy-makers deciding how best to commission personalised medicine to gain maximum benefit from the available healthcare budget?

Personalized Genomic Medicine and Health Care Justice

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Michelle L. McGowan Ph.D.](#), [Case Western Reserve University Department of Bioethics](#).

The most significant challenge to the promise of personalized genomic medicine hinges on the realization of health care justice. From a social justice perspective, meeting the basic health care needs of the population – locally and globally – is an urgent objective that dwarfs the goal of a genomic approach to medicine.

The challenge to proponents of personalized genomic medicine is to find a way to frame their aspirations and actions in a way that simultaneously moves towards realizing personalized medicine and global health care justice. However, the terminology and current direction of personalized medicine has the potential to hinder this effort.

To date the promotional rhetoric of personalized medicine has emphasized how access to one's genomic information will be personally empowering, that it can lead to individually tailored prescriptions and therapies, and that knowledge of genomic risk susceptibilities can prompt preventive health measures and curb the manifestation of disease. The underbelly of these promises is its individualist orientation which invokes a neoliberal ideology toward health with obliges personal responsibility for health management.

This approach seems painfully out of step with the social debates on how to achieve access to basic health care for the population as a whole. It seems to me that the promise of individually-tailored health services can best be achieved in a context in which the entire population is ensured access to basic health care, because without it the realization of personalized medicine has the potential to further exacerbate existing health disparities and generate new forms of health inequities.

Personalized medicine, leave U.S. behind

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [David Dooling of the Genome Center at Washington University in St. Louis School of Medicine](#).

We are currently in the throes of a heated debate on the future of health care in the United States. Despite endless hours of discussion amongst politicians, health care experts, and pundits, very little time has been spent discussing the implications of the current and any proposed health care system on medical research. The passage of the [Genetic Information Nondiscrimination Act \(GINA\)](#) last year makes it illegal for insurance companies or employers to discriminate on the basis of genetic information, e.g., the results of genetic tests for breast cancer susceptibility. The passage of this law promised to usher in an era of genetic testing and personalized medicine, but that era has not begun. There have been announcements of [studies involving large cohorts of patients being genotyped as part of their normal course of treatment](#), but what percentage of patients will consent to these studies knowing that insurance companies can deny life-saving care because of clerical errors? While there have been some success stories applying genome sequencing to cancer treatment (ironically the above linked Nature News article cites an example from Canada), other more complex genetic diseases will need very large cohorts of patients to establish genotype/phenotype relationships.

These cohorts will not (and indeed should not) materialize until insurance companies cannot deny coverage for pre-existing conditions and cannot drop or limit coverage for the seriously ill. After all, genetic testing performed during treatment for one disease could turn up susceptibility to a whole host of other diseases; susceptibility to diseases that may induce insurance companies to look back through your forms for clerical errors. Fortunately, most members of Congress agree that the above are worthwhile reforms. Unfortunately, neither of these reforms do anything to control the cost of health insurance, and may even cause it to increase, leading to fewer insured and therefore fewer potential study participants unless there are other reforms to address skyrocketing health care costs (e.g., a strong public health insurance option). In short, without health care reform that guarantees coverage and controls costs, medical researchers in the United States will be at a significant disadvantage because the large-scale genetic studies required to understand complex diseases will not be available to them.

Without these studies our knowledge of the link between genetics and disease will remain vague probabilities that provide cocktail party fodder for those wealthy enough to afford health insurance and consumer genetic testing.

Potential increased health disparities related to genomic medicine

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [The Board of Directors](#) of the [National Society of Genetic Counselors](#).

There are many existing barriers to accessing healthcare. However, as the advances in genetic science and technologies are integrated into mainstream medicine, the gap between those who can afford and those who cannot afford healthcare will evolve into a new form of health disparities. There will always be those people who can afford the latest in medical technology and will have access to a \$1000 genome test and genetic counseling to help interpret the results and fully integrate the information into their personalized medical plan.

However, a significant portion of Americans, and arguably those who most need to target their healthcare dollars, will not have access to the information. The use of genomic data to tailor a care plan should become a mainstream component of medical care for all people. Prevention, early detection, and risk reduction options are valuable to all Americans regardless of their ability to pay.

We need to work together to expand access to genetic testing

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Jonathan T. Lord, Navigenics, Inc.](#)

At Navigenics, we know that the future of health care rests in preventing disease, not just treating it. And we know that personalized genomics will play a big part in transforming medicine from a “sick care” system to a true health care system, allowing doctors to tailor prevention plans and treatments to meet patients’ needs.

But in order to expand access to personalized genetic testing, we must first address the most pressing question – where and how to start?

With awareness. We need to raise awareness of genetic testing and set accurate expectations. At a time of high drama around healthcare reform, much of the debate has focused on reforming health care financing and not on fundamentally changing health care. We need to do a better job at highlighting the promise and importance of genetic testing. This could involve public health campaigns or even curriculum restructuring.

There are also questions of access, not only to reliable testing services but also to follow up health care and physicians. Unless there is universal access to basic services, the offer of genetic testing inevitably creates health care disparities.

We believe that genetic testing should become a standard service, covered through 3rd party payers. But it still needs to have greater acceptance by the medical establishment. Until this occurs, the promise of any broad utility will be stifled.

There is an opportunity to make a real difference for individuals’ health and to systemically make prevention a part of the health care scene. We see a time when genetic testing is an integral element in personal health records; a time when doctors use this information to guide prevention and treatment regimens; and a time when errors in the system are dramatically reduced by this promising tool. We need to work together to make this a reality.

Medical vs. “Recreational” Genomics: Drawing a Line in the Sand

This commentary in the Genomics Law Report’s ongoing series [What ELSI is New?](#) is contributed by [Allie Janson](#), [University Health Network/Mount Sinai Hospital’s Fred A. Litwin Family Centre in Genetic Medicine](#) and [DNA Exchange](#).

To me, one of the most interesting aspects of genomics is the potential for it to be both medical and recreational in nature. Direct-to-consumer companies have picked up on the recreational aspect of genomics and run with it—successfully marketing their service as a fun and interesting glimpse “inside oneself.” Understandably, the medical community has been, and will be, slower and more cautious about incorporating weak gene variant-disease associations into medical care. However, as the 1000\$ genome comes barrelling towards hospital doors, physicians may no longer have a choice. Patients will present with a symptom and a genome print-out in hand, and demand answers.

In addition to the logistical issues of resources, education and time, there is a more theoretical question at hand: How do we distinguish between genomic information that belongs in the medical setting and genomic information that does not? I think this issue extends beyond peer-reviewed publications and FDA approval. Historically, a key step in any genetics evaluation has involved defining the purpose of a genetic test. In the genomic era of medicine, however, defining a specific purpose may prove challenging, as many gene variants will have associations with multiple medical traits, or with both medical and non-medical traits. Who has the responsibility to report this information to the patient? And how do we manage patients who are interested in learning about certain risks but not others?

Just as genomics has the potential to alter the way an individual perceives oneself, it also has the potential to challenge the fundamental principles of medical care. For now, “recreational genomics” companies have the benefit of (however controversially) being able to sell their service as a non-medical product. But the medical community has a much more complicated task at hand. A task, I imagine, that will require some creative thinking, consensus building and flexibility on the part of physicians and patients alike.

Informed Health Decision-Making through a Registry for Genetic Tests

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Sharon F. Terry, Genetic Alliance](#).

Diagnostic testing is one of the first clinical fruits of genomics, and certainly the leading edge of personalized medicine. As such, the most critical ethical, legal and social issue that must be addressed by the field of genomics and personalized medicine is oversight of these tests. We focus here on one aspect of oversight: a registry for genetic tests to allow providers and consumers to make informed choices.

A registry should be developed and maintained that includes the name of the laboratory performing a specific test, the name of the laboratory or manufacturer that developed the test, and information to support claims about the analytical validity and clinical validity of that specific test or test method. Submission of information to this registry should be mandatory for all advanced diagnostic assays. This registry should be created and maintained by NIH since it has enormous capacity related to genomic informatics.

Such a registry would provide a framework for transparency to allow the field to contribute to clinical medicine in a meaningful way. Individuals, their providers and payers, would have the information they need to make informed decisions. We desire a transparent regulatory system. As Javitt et al state in a recent paper, the Secretary of HHS has “the explicit authority under both the FD&C Act and CLIA to develop a genetic test registry in order to protect public health.”[FN 1] Oversight of genetic tests requires a modern 21st century system involving FDA, CLIA, and many stakeholders, this is a necessary first step.

FDA should develop consistent evidentiary standards for post-market labeling changes to include pharmacogenetic information

*This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Stephanie Devaney](#), [Genetics and Public Policy Center at Johns Hopkins University](#), and [Gail Javitt](#), [Berman Institute of Bioethics at Johns Hopkins University](#) and [Sidley Austin LLP](#)**

A prescription drug's labeling is the primary vehicle used by drug manufacturers to inform providers of the conditions under which a drug is safe and effective for use, and must be approved by FDA at the time the drug is approved. Post-market labeling changes may also require FDA approval depending on the type of change. Pharmacogenetics is a relatively new and complex field that regulators and drug sponsors alike are trying to navigate. In recent post-market labeling changes to include pharmacogenetic information, FDA has been inconsistent in the type and amount of data it has required to support the new claim. A clearer regulatory path is critical to encourage drug sponsors to invest in pharmacogenetic research and to ensure that health care providers get the information they need to get patients the right drug at the right dose at the right time.

FDA encourages sponsors to include pharmacogenetic information as part of new drug applications. In 2005 the agency issued a guidance document titled "Pharmacogenomic Data Submissions", which explains when and how pharmacogenomic data should be submitted during drug review. However, there are several hurdles to the incorporation of this information post market, especially when the efficacy of the drug is affected thus narrowing the patient population. Although FDA appears to be willing to permit pharmacogenetic claims relating to safety that are supported by a modest level of evidence, data from randomized controlled trials are usually required for claims of reduced or increased efficacy. However, conducting a randomized controlled trial for every potential predictive genetic variant would be costly and risks hampering innovation. On the other hand, absent appropriate data collection and analytical methods, retrospective analyses of data from completed clinical trials run the risk of creating biased results.

The recent debate between FDA and sponsors of the colon cancer drugs Erbitux (cetuximab) and Vectibix (panitumumab) over whether the drugs' labeling should include KRAS mutant status as predictive of drug efficacy highlights FDA's discomfort with using retrospective data to support efficacy claims. However, the rapid pace of genomics research means that genetic effects on drug response will frequently be identified post market. Therefore, clear guidelines on when retrospective analyses will be adequate and what criteria such data must meet are necessary to facilitate incorporation of pharmacogenetic information into drug labeling. This position echoes the recent comments of Larry Lesko, director of the Office of Clinical Pharmacology at FDA, during the National Conference on Personalized Healthcare in early October 2009. He

commented that the KRAS experience could be used to set a framework to inform other drug sponsors on how to utilize retrospective analyses of genomic biomarkers and drug response.

*The views expressed in this article are exclusively those of the author and do not necessarily reflect those of Sidley Austin LLP and its partners.

The Hidden Legal Barriers to Scientific Research

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Thinh H. Nguyen](#), [Creative Commons Counsel for Science Commons](#).

When thinking about the legal issues associated with genomics, many people, particularly lawyers, tend to focus on patent issues. While there are legitimate concerns about patents, there is a growing body of sociological research to suggest that in the vast majority of cases, bench science is not impeded by fear of patent lawsuits, but rather by far more mundane legal and cultural barriers.

Such barriers can arise from private agreements between the participants in the exchange. For example, material transfer agreements (“MTAs”), which are bailment agreements that often govern the exchange of reagents or biological specimens, continue to generate significant negotiation costs, delays, and failed instances of sharing, despite an NIH mandate more than a decade ago to simplify and standardize the process of sharing biological resources.

The sharing of scientific data and databases on the Web represents an important opportunity to accelerate the process of scientific discovery, but it also breeds a proliferation of legal forms, including click-through data use agreements, Website terms of use, and other restrictions that are not only bewildering to scientists but also to the lawyers who support them. The use of such restrictive agreements, often unnecessary, can present a significant problem for research projects that must depend on data aggregation or integration, an important tool of genomics research.

The increasing extent to which legal formalities, and thus lawyers, intermediate scientific sharing, not only between academia and industry but increasingly between academics themselves, represents a cultural sea-change with important consequences for how science is conducted in the future. The public and unrestricted availability of genomic databases and resources, or the “commons”, must be cultivated and defended, through a robust community consensus, or it can easily fragment into legally constructed “walled gardens” without public right of way.

Chapter 3

Too Much Information

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How will medicine and its regulation adapt to the information age?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Andro Hsu, 23andMe, Inc.](#)

Jorge Luis Borges once [compared](#) scientific endeavor to the making of a map as large as the territory the map depicted. As the price of generating DNA sequence and other data types decreases more quickly than Moore's Law, we are well on our way to creating a map even larger than reality.

To handle these vast amounts of biological data, we will eventually need computers not only for storage, but also to run algorithms that can simultaneously process, synthesize, and evaluate thousands of interacting biological and environmental variables—tasks beyond the capabilities of any human brain.

The problem? Our notion of evidence—and the regulatory framework that governs its use in setting clinical guidelines, approving drugs and medical devices, and making medical decisions—was developed during a reductionist era of low-hanging informational fruit, when tiny sample sizes sufficed to demonstrate large relative risks. Today's holistic approach uses higher-resolution, system-wide data to power sophisticated statistical models that produce incremental gains in prognosis, but require ever larger samples to validate.

So what happens when we can obtain so much unique information about a person that the only appropriate population for validating predictions made from those data is the subject herself? Conversely: even if we had comprehensive biological and environmental data for the seven billion humans in the world, how could we provide sufficient evidence when rigorous validation of all possible interactions requires a sample size larger than the world itself?

To benefit from increasingly detailed data, we need to cooperate in finding a way for regulators, health care professionals, and patients to get comfortable with multivariate algorithms, computational models incorporating correlations without underlying causation, and tests that measure and combine analyte levels to produce individualized answers in the form of probabilities. Welcome to systems biology, a field as messy as life.

The balance of experiment and theory is shifting in genomics; this matters for ELSI

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Robert Cook-Deegan](#), [Duke University Institute for Genome Sciences & Policy](#).

I am teaching Darwin this semester with a colleague from the English Department. It's a real gas to go back to the roots of modern biology and see the shift from pure field observation and categorization to theoretically driven argument. Since Warren Weaver coined the term and then primed the pump for molecular biology in 1938 at the Rockefeller Foundation, biology has become a much more experimental science. Many lineages of embryology, genetics, and other parts of biology always were experimental, but after World War II, molecular approaches became dominant, driven in no small part by a succession of new technologies: ultracentrifuges, radio-isotopes incorporated into macromolecules, protein sequencing and synthesis, recombinant DNA, and nucleic acid sequencing. Sequencing really caught fire in the 1980s, and joined forces with the broader revolution in computing and communication using the Internet and computational firepower following Gordon Moore's trajectory of faster, cheaper, and smaller computing power.

Many other parts of biology are changing too, and this is not a claim of genomic exceptionalism. It is merely a claim that things are changing, and fast, not that they are only changing in genomics (just think of stem cells). As I think about "what's new," I find myself drawn to one hugely important change: the log-a-year improvements in sequencing technology that suggest we'll be generating one whale of a lot of DNA sequence data, and not just about *Homo sapiens* (I hope). I don't pretend to know what that means. But I am drawn to two lines of research that seem to give us a glimpse of some major changes ahead that we would do well to be thinking about. I don't quite know what to say about them, except to say that my antennae are quivering with a sense that they are really important.

One development is about what used to be called regulation of gene expression. It's getting too complicated for that to really capture the meaning, but the general idea is that the genome is not even close to static, or the passive repository of Mendel's particles of inheritance. The ENCODE project is throwing up all sorts of new insights, casting doubt on assumptions that conserved elements are selected for function and unconserved sequences are adrift—or that unexpressed regions can be called "junk." Sydney Brenner used to joke that "junk" does not mean useless, but the stuff you keep in the attic. Seems to me like this "junk DNA" is all over the kitchen, family room, and not least the bedroom, but there's not nearly so much up in the attic as we thought.

Why does this matter for ELSI? Let me count the ways, but start with just one to illustrate. On the Law axis of ELSI, we have 50,000 DNA patents largely premised on what we thought was true about the genome. Remember when we thought there were over 100,000 genes, or that one gene encoded one RNA encoding one protein? That's so yesterday. This legacy of patents may begin to expire before the stakes are worth fighting over in court—or they may not. But one

thing's for sure, a cell is not a bag of genes that can be specified and patented under the Central Dogma of molecular biology. Yet parse the fractured English of many patent claims and you'll see Watson, Crick, Beadle and Tatum laid bare.

The other tectonic movement in biology is beautifully illustrated by the paper from the Broad Institute on the human population genetic history of India by David Reich and his colleagues. It's mind-blowing how off-the-shelf chips derived from common variations discovered in the HapMap can generate data on 125 people to reach powerful inferences using fancy math. Hypotheses about population relatedness can be tested and falsified. That's using technology that began to take off a decade ago. Imagine now what we'll be contending with as sequencing enters the fray. We should have an order or two more informative data, made even richer if we preserve the information about who passed genes to whom in the data sets. It seems quite likely that the granularity of human population genetics is poised to rocket because it won't take all that many people to make inferences about any human lineage that we study.

I was once naive enough to think that the study of human population genetics would conquer racism, or at least strongly challenge it. I thought science might put a stake through the dark heart of eugenics and racial hygiene. That's because I didn't know much about racism except some of the genetics of human inheritance. Genomics won't conquer racism any more than evolutionary biology has beaten back Creationism, and Maynard Olson, among others, in "Davenport's Dream" has noted that we can't count on the facts reinforcing every Liberal Dream. We may find some phenotype-genotype associations that we find downright uncomfortable that cannot be attributed to schlock science. It is not a given, but it is a possibility. I just have to think there are many, many issues that are going to arise as it becomes cheap and fast to study the full genomes of lots of people, and as we can reconstruct our common and our uncommon ancestry with levels of specificity vastly beyond what we have been able to do until now.

How will we handle the rapidly approaching flood of genomic information on individual patients and consumers?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Hank Greely, Stanford Law School](#).

The cost of advanced genome analysis is falling rapidly. The fabled “thousand dollar genome” is less than five years away. It will be as cheap to order a whole genome as to order any one genetic test. As the price falls, many people will buy “their genomes,” or, at least, information on billions of base pairs, millions of SNPs, and unknown numbers of copy number variations and translocations. What will we do with that information?

Some may think we already are facing this problem with the “consumer genomics” companies like Navigenics and 23andMe, but those SNP-chip-based companies have the “advantage” that they produce weak information, linked to only small variations in disease risk. Fully detailed genomic information will unearth something frightening in all humans’ genomes, for themselves or for their (existing or possible) children and other family members.

Patients need to understand the true implications of this genomic information, but how will we accomplish that? Some primary care physicians may be able to make sense of information about a few famous disease-related genomic variations – perhaps well-known mutations in BRCA 1 or 2, the expanded CAG repeats of Huntington’s disease, or the most common cystic fibrosis-associated mutations of CTFR1. Most won’t know even that; none have a clue about the thousands of rarer genetic disease associations, let alone the hundreds of published pharmacogenomic associations. Even clinical geneticists and genetic counselors will not know all the important variations, and in any event, those professionals are far too rare to handle any significant part of the demand. Even if we had the capacity to provide counseling, our current regulatory scheme does not require that genome consumer get any professional explanation – good, bad, or indifferent – to genome consumers. The age of cheap full genomes is almost upon us – and we are not close to ready for it.

Dear Dr. Board-Certified Clinical Geneticist

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Misha Angrist, Duke University Institute for Genome Sciences & Policy](#).

Dear Dr. Board-Certified Clinical Geneticist:

First of all, thank you! Sure, you could have bailed after pediatrics or internal medicine and made more money, but you chose to stick around for a couple more years, incur still more debt, and make less. Much less! (Fortunately, you are a doctor and not a financial adviser.) And a fine doctor you are, schooled in the ancient art of dysmorphology, which is probably as inscrutable to your molecular-fetishist colleagues as exon-capture protocols are to you.

Anyway, here you are, languishing at the bottom of the physician food chain. But hey, at least you have the psychiatrists for company!

I know: You rarely do procedures beyond physical exams (albeit terrific physical exams). Otherwise, mostly you talk to people. And order tests that generate bupkes for your office.

For patients with highly penetrant Mendelian disorders or syndromic conditions, you are a lifesaver. But of course, CMS can't wrap its dysplastic little brain around this and so it doesn't like to pay for your services. Meanwhile, genetic counseling has been around for decades, yet you still have no ingenious way to bill for it. (God knows where you'd be without those trusty "physician extenders.") To paraphrase Three 6 Mafia, "It's hard out here for a geneticist."

But I have good news: this is all about to change! Because this is your One Shining Moment! We are hurtling toward the \$1000 genome! If you thought the line of patients coming in with their 23andMe reports was annoying, just wait until they show up with their raw sequence from Illumina! Heh. I kid because I care.

And because I know you're up to the challenge. As Dr. McKusick said, you are the last generalist. You're all about incorporating as many disparate forms of data as you can into diagnosis and treatment. You recognize that the time has come for your guild to begin to think about diabetes, MS, Alzheimer's, arthritis and MI. You are ready to coach your community on the benefits of prevention and to begin to parse SNP chip data, CGH arrays and yes, whole-genome sequences.

And I feel certain that you're ready to roll up your sleeves and start revamping your med school curriculum, your training programs and your pay scale in order to better your own lot and to recruit the most promising young physician-scientists in the world. Just imagine: a universe in which patients don't need to wait six months or a year to see you!

Listen, it's not the surgeons, oncologists and cardiologists who have the keys to the kingdom. It's you, Doc! You've already forgotten more about phenotype than your gene-jockey neighbors will ever know. So come on, Doc, leverage that power!

Oh, and get this: one of your own is now Director of the NIH! I'd bet you dollars to donuts that no one wants you to succeed more than he does. Try him!

This is your time. Carpe diem, babe!

Sincerely,

Misha

Personalized Medicine in the Web 2.0 Era

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Bertalan Mesko](#), [Webicina.com](#).

There have been huge developments in the field of personalized medicine in the last few years but the major problems we have to face have just now become clear. People are ordering direct-to-consumer genetic tests from home; they expect general practitioners to answer genomics-related questions that might affect their health management or lead to medical decisions, and they are publishing information that should never leave one's computer.

Educating medical professionals and preparing the public to be able to provide informed consent for these activities is crucial to this next phase; as is the accessibility of the enormous and growing amount of genomic data. A new generation of web services, the so-called web 2.0, seems to be playing an important role in this movement. Through such tools, people can interact with their doctors and each other easily and they can share the results of genomic tests, resulting in a self-maintained database of human genomic information. Examples include the research project of 23andMe.com or the amyotrophic lateral sclerosis research of Patientslikeme.com.

The main challenge to personalized medicine over the coming years is to find a way to combine these pieces of information with a public library of scientific findings in order to create a comprehensive clinical database of genotype/phenotype associations. We must also make it easier for patients to access reliable websites focusing on personalized medicine and help medical professionals understand where and how they can locate clinically relevant information about new therapies or diagnostic tests, because it is all too easy for patients and even researchers to become lost in a cloud of scientific publications, without mentioning language problems.

By involving web 2.0 tools such as secure communities, new communicational channels of social media or collaboration-based sites, personalized medicine may soon achieve its rightful place in the global medical palette.

Chapter 4

Back to School

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We must revolutionize our communication of science to non-scientists

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Chris Gunter](#), [HudsonAlpha Institute for Biotechnology](#).

So many issues are intertwined that it's extremely difficult to pick just one, but I suggest we must revolutionize our communication of science to non-scientists. In my past job as an editor for Nature, a daily task was to translate the most cutting-edge life science discoveries into language for multiple purposes: marketing, press release, highlighting in other journals, and so on. Regularly I would be dismayed at the end results, and resolve to somehow explain things better next time. Common ground between scientific papers and non-scientists can be harder to find than the "missing heritability."

Now I work at a nonprofit institute and spend much time giving public lectures. I am struck by the absolute hunger of the public to understand genomics and personalized medicine. Our institute's outreach activities are generating much-needed support from politicians, local universities, regional clinical centers, and school groups of all ages. People I meet often want to donate their time, money, or samples – but first and foremost, they want to understand.

Thus I propose our field engage in our own form of personalization: using education and media in all forms to convert the energy of the public into an army for science. Studies report non-scientists are likely to hear about genomics from their doctors or the media. Let's incentivize scientists to use all forms of media we have now (and then invent some new ones) to engage the public. Let's require two-way communication between health professionals and scientists on what applications or materials would be most useful and feasible in clinical settings. Let's convince publishing firms to find new ways of making research available and accessible, and enable journalists and educators to be creative and accurate in conveying research results. Without such a revolution, I submit there will be no "full realization" of personalized medicine.

Education, Not Regulation, Will Benefit Consumers of Recreational Genetics

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Blaine Bettinger, Bond, Schoeneck & King](#) and publisher of [The Genetic Genealogist](#).

Genetic ancestry testing, the use of DNA to explore an individual's recent or ancient genetic contributors, has been available to customers for almost a decade but has recently been a topic of much debate among bioethicists. The concerns often center around topics such as privacy, definitions of race, and emotional or psychological effects of test results, among others. These concerns, together with continuing advances in personalized genomics that have the potential to make our DNA an important part of how we shape our identity and interact with society, lead to the question of whether recreational genetics should be regulated in order to prevent any potential harms to consumers. Or, as some argue, will regulations hinder the field without providing any real benefit?

Although perhaps not the most important ethical or social issue faced by the field of personal genomics, it is a concern shared by many of the estimated 1 million people who have purchased a genetic ancestry test to date. Genetic genealogy was the first branch of personal genomics to develop into a commercial product, and many who have benefited from testing vehemently oppose any regulation which might hinder an individual's ability explore his or her own DNA.

There are indeed ethical issues faced by the field of genetic ancestry testing, but the solution to these issues, as with so many other aspects of personal genomics, is education rather than regulation. It is impossible for every consumer to be an expert, and as a result genetic ancestry testing companies must continue to make every effort to educate them. Many of the ancestry testing companies provide their customers with the relevant research and resources necessary to make sense of the results, but educating the consumer cannot solely be the responsibility of the companies. Our education system currently provides most students with only a cursory examination of genetics and fails to provide them with the tools needed to understand and evaluate even the most basic science underlying personal genomics. With the potential for personal genomics to revolutionize healthcare and play an important role in so many other fields, it is vital that everyone possess these tools.

Although the push for regulation is admirably aimed at preventing consumer harm, regulation will likely not provide consumers with the one thing they need most as they face a world changed by personal genomics – education.

The Risk of Communicating Risk

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Zoe McDougall](#) and [Dr. Gavin Harper](#), [Oxford Nanopore Technologies](#).

“We teach our children the mathematics of certainty but not about how to handle uncertainty and risk in the world”. David Spiegelhalter, Professor of the Public Understanding of Risk, University of Cambridge, UK.

In April 2009, the UK's Meteorology Office forecasted a 65% chance of above-average temperatures in the UK for the summer. The media grasped this statement and declared an odds-on ‘barbecue summer’. The public took action on the basis of these headlines; sales of domestic holidays went stellar. By late July, weeks of rain had caused a national public backlash against the Met Office. Few people understood or recalled that they had been advised that there was a 35% chance of a washout summer. Ironically, the temperatures had been above average despite the rain.

This episode is illustrative of the risk of communicating risk. Assume that an authority figure truly understands the probabilities surrounding a scenario. It is a substantial challenge for them to educate their audience to a degree that they understand that risk, can make a truly informed decision, and accept responsibility for that decision if the dice don't roll their way.

As we enter an era of personalized, genomic medicine, the understanding and communication of probabilities is likely to be a stumbling block, not only for the public but for clinicians too. As genome science accelerates, scientists and clinicians will be faced with evolving risk scenarios. Particularly when looking at risk of common disease, most newly discovered variants are probabilistic rather than deterministic. Integrating these into public health policies or presenting probabilities to individual patients will require enormous skill and should stimulate debate about responsibilities for decision making. Given the complexity of genomic data there may be many scenarios in which consent can never be truly informed, risking paralysis in the system.

How to provide context, interpretation and counselling around these complex sets of probabilities is a new challenge in statistics, ethics and psychology. We could start, as Professor Spiegelhalter recommends, by teaching our children more about the mathematics of uncertainty.

Why Are We Missing What Is Important In Personalized Medicine?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Steven Murphy](#), [Helix Health of CT](#).

A 33 year old man with anxiety comes to the office. He says he has some worsening of his anxiety. It causes chest pressure. I saw his brother last week for a physical. He told me his father had a heart attack at 50.

The TIMI score for risk puts him at a 0.8% all cause 30 day mortality risk. No big deal right? His Reynolds Risk? 2%. Do I send him for a genetic test? No. I send him for a stress test. Why? Family History.

There are more people with Family History, than with faulty SNPs.

The results? Grossly abnormal test. He now has a coronary artery stent.

The algorithms aren't refined for personalized medicine or family history. How can we expect Genomic data to change these algorithms?

How can we teach doctors genetics? How can we help doctors integrate it into care?

The attention is on the wrong things. How can we focus on what saves lives? How can we focus on what reduces costs? How can we focus on what is important with all the noise in this space?

Why are we failing physicians by lack of education? How can we demonstrate that this is a tool to augment care? Why are we failing patients by failing to educate their providers? That is the key here, not Open Bars, Blimps, TweetUps, or Public Relations.

Those things just create noise in the hurried clinician's mind. Unless of course they (the clinicians) have already tuned out anything with the name "Genetic/Genomic" in it.....

The next generation is in high school.

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Dana Waring](#) and [Ting Wu](#), co-founders of the [Personal Genetics Education Project](#).

Personal genetics is just that, it's personal. Regardless of how many laws or societal norms are in play, any decision an individual makes about knowing or using personal genetic information will ultimately be a personal one, informed by the richness of that individual's own history and circumstances. Therefore, in order for personal genetics to be adopted in a fair and ethical way, broad-based education is critical. We argue here that the education of high school students may be our most important responsibility and, coincidentally, the most far-reaching, cost-effective, and timely approach for achieving worldwide understanding of the benefits, risks, and ethics of personal genetics.

Let's assume that predictions are correct, and genome sequencing will be available for less than \$1000 US, or equivalent, within five years. If so, current high school students will form the first generation to come of age and face, en masse, the opportunities and consequences of personal genetics. They will be securing first jobs, buying insurance, finding partners, and starting families just as genome sequencing becomes mainstream, allowing them to know themselves, their partners, and their children in unprecedented detail. Will they be prepared?

By focusing educational efforts on high school students, we will ensure that the vast majority of our next generation will have had a chance to understand and debate the complexities of personal genetics. Because infrastructures for educating our youth are in place worldwide, conduits for introducing the concepts of responsible genetics are immediately available. Furthermore, the extant curricula of science, sociology, history, literature, health, and community service offer natural venues for teaching and debate. Our experience with pgEd indicates that high school students find the implications of personal genetics gripping, that targeting educational efforts to them will ensure that the next and subsequent generations will be informed, engaged, and prepared.

Chapter 5

No _____ Need Apply

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Finding The Proper Place for Genetics in Insurance

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by Mark Henderson, Science Editor for [The Times](#) and author of [50 Genetics Ideas You Really Need to Know](#).

It is a mark of the quality of the science fiction movie Gattaca that, 12 years after its release, so many ethical issues surrounding genetics are still seen through its prism. In Andrew Niccol's dystopian vision, DNA divides society into "valids" and "invalids", some of whom are decidedly more equal than others.

Concerns about such genetic discrimination are relevant to many ELSI discussions, but rarely more so than when the subject is insurance. The great fear is that genetic data could create an uninsurable underclass, denied mortgages and life and health cover because of genomic flaws beyond their control.

The consensus that this would be wrong is so strong that the Genetic Information Non-Discrimination Act (GINA) passed the US Congress with barely a murmur of opposition. A UK moratorium on the use of genetic tests by insurers has also recently been renewed. But it is far from clear that the issues that genomics raises for fair insurance are either new or especially problematic.

Insurers have used genetic information for as long as there has been insurance. Until GINA they were quite entitled to ask about family health history — a proxy for genetics, and in the UK they still are. And it is not controversial to charge higher premiums to drivers with Y chromosomes. Neither is genetic injustice confined to consumers. An individual who discovers she has a raised risk of a degenerative disease like Alzheimer's has [an incentive to buy long-term care insurance](#), at a premium that is fair to neither the provider nor its other customers.

Insurers may also be less interested in genetic discrimination than is often imagined. Gil Baldwin, managing director of Aviva Health, has [told me he would be happy in principle to give discounts to customers who have themselves sequenced](#), even if they do not share the results. His logic is that they will be better motivated to change their lifestyles to lower both their risks and his.

Market mechanisms like this, indeed, should be capable of meeting many of the insurance challenges of the genomic age. Nobody, after all, has a perfect genome, and insurers are businesses that need customers. We all have above-average inherited risks of some disorder or other, and companies that set the genetic bar too high will soon go bust.

I'm not saying that the insurance industry requires no regulation in its use of genetics. There may well be a need, for instance, for a safety net for people who carry rare mutations and risk

profiles. The knee-jerk argument that DNA has no legitimate place in insurance, however, is not as compelling as it may seem.

Personal genomics and genetic information nondiscrimination legislation: Are we ready?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by David Gurwitz, [Tel-Aviv University Department of Human Molecular Genetics and Biochemistry](#).

The age of personal genomics has arrived faster than most have expected. While the purchase of full genome sequencing services may today be too costly for most to consider, full genome sequencing costs are projected to fall under US\$1000 within less than a decade. But are we ready for the personal genomics age and its far-reaching societal implications? There are countless ELSI matters that need to be considered in the context of readily available personal genomes. One key aspect is the need for more comprehensive genetic information non-discrimination legislation.

Legislation is a notoriously slow process; when it comes to handling new technologies, legislators may take many years to catch up. Israel was among the first States to pass – already in 2000 – a comprehensive genetic information non-discrimination law; some European States soon followed. In the US, however, a similar legislation process suffered a lengthy birth: The Genetic Information Nondiscrimination Act (GINA) of 2008 has been in the making for over a decade (1), in spite of strong support from research associations and groups such as the NIH Pharmacogenomics Research Network (2). GINA, finally signed into law on May 21, 2008, protects Americans against discrimination in health coverage and employment based on individual genetic information (3). However it does not protect individuals against misuse of genetic information by life or disability insurance providers, banks, schools, or immigration authorities (4-6). Amending GINA for assuring wider protections seems urgent: personal genome sequences coming from proliferating electronic health records and government forensics databases, and other sources of personal sequences, including Facebook-like websites, are being contained in innumerable internet-based resources. Safeguards are typically in place to ensure that only qualified persons may access personal genetic profile records, but no safeguards are failsafe (7).

We are the citizens of an ever more closely connected global village. Individuals may migrate to new countries in pursuit of better jobs, security, or love. The accessibility of personal genomic data on the internet, however, knows no boundaries. Individuals may find themselves living in countries where privacy protections are below the level they have been accustomed to in their homeland country, while their personal genetic data acquired at home will accompany them to their new base. There is thus a far more urgent need: a global agreement prohibiting discrimination based on personal genetic information.

This remarkable task is doable: the global ban on human cloning is a fine example. A Declaration or International Bill – ideally issued by the United Nations – may be the most

efficient way forward. Article 2 of the Universal Declaration of Human Rights, the basis for the International Bill of Human Rights, states that

“Everyone is entitled to all the rights and freedoms set forth in this Declaration, without distinction of any kind, such as race, colour, sex, language, religion, political or other opinion, national or social origin, property, birth or other status.”

Ideally, this list should also include, ‘personal genetic information’. Let us hope this will not take too long – so we can be less concerned about the age of personal genomics.

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Genetic discrimination: problem or paradox?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Alison Hall](#) and [Caroline Wright, PHG Foundation](#).

Over the last decade, our acquisition of genetic knowledge has gathered pace; whole human genome sequencing is now within reach, as accessibility increases and prices tumble. One consequence has been to challenge existing conceptions of what the term 'genetic' actually implies. The popular perception that a person is shaped solely by their genes has been undermined by the sheer volume, complexity and mundane nature of much genomic information. Whilst genomics and personalised medicine have the potential to unlock copious biological secrets and yield enormous medical benefits, other forms of information (such as family history, medical imaging or other biomarkers) may be equally useful, predictive and personally sensitive.

It therefore seems all the more perplexing that 'genetic exceptionalism' – namely the notion that genetic material is special and distinctive – remains an enduring belief amongst the general public and relevant professional disciplines. The concern that third parties, such as employers or insurers, who have access to this information could use it in ways that discriminate against the individual to his detriment has already prompted some jurisdictions to impose legislation banning 'genetic discrimination'. Such legislation is aimed at preventing employers from excluding those susceptible to future disease from potential employment, or insurers from using the results of genetic tests to identify and exclude at-risk individuals, ultimately leading to the creation of an unemployable and uninsurable genetic underclass.

The spectre of genetic discrimination seems likely to restrict generalised access to new genomic technologies. Relegating personalised medicine to the monopoly of professional elites is, in our view, misguided, unenforceable and unnecessarily paternalistic. However, worries about genetic discrimination still cast a shadow over the development of proportionate regulation, which requires education of citizens and institutions alike. Concerns over genetic discrimination must therefore be addressed in a practical and appropriately nuanced manner before the full promise of genomics and personalised medicine can be realised.

Does a genomics that does not work for some mean a genomics that will not work for all?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Jenny Reardon](#), [Center of Biomolecular Science and Engineering at UC Santa Cruz](#), and [Stephanie M. Fullerton](#), [Department of Bioethics and Humanities at the University of Washington](#).

The future promise of personalized medicine rests precariously on the care with which we pursue genomic research in the current moment. While a focus on legal protections and open access are important, we must also attend to fundamental questions about the constitution of human diversity at the genomic and social level.

This is clearly evident in the recent collision of open source genomics with privacy rights. While calls for unfettered data sharing have formed the ground for much biomedical research, the achievement of a genomic commons may create its own kind of blockages. For example, although it is finally against the law to use genomic information to discriminate in healthcare or employment, the Genetic Information Non-Discrimination Act does not protect against misuses related to long term care coverage, life insurance, membership in federally-recognized groups, or immigration. For these and related reasons many people remain wary of involvement in genomic research. “Open access,” in other words, may inadvertently close the door for many.

Does this form of genomics, one that may not work for some, mean a genomics that will not work for all? Answering this question requires addressing questions about the ordering of human beings that are at once scientific and social. Scientifically, the limited ability of GWAS to explain trait variation has called into question the predominant role of common variation in disease risk. This has placed a renewed premium on the identification of rare variants. Only significant effort will achieve the participation of the broader and more diverse range of human beings required for such research. Understanding why some people participate, and many do not, will demand understanding the specific ways in which genomic ideas and practices form from and re-form social practices of racism and inequality—issues that remain with us despite the last decade of proclamations about the anti-racist and equalitarian features of genomics.

If the subjects of genomic research remain primarily those categorized as “white” or “European,” and those who have the means to afford genetic testing, then genomics will fall short of its goals to be a human science that meets human needs. To avoid this future requires that scientists more carefully attend and respond to questions about the ordering of nature and society that their field poses.

Chapter 6

Testing the Limits?

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How should we deal with the arrival of very common prenatal testing for a broad set of genetic characteristics?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Hank Greely, Stanford University](#).

Each year, about 60,000 pregnant women undergo prenatal genetic testing in the United States, out of more than 4.2 million live births. Within five years, new testing methods, made possible by the rise of cheap forms of genomic analysis, will be able to test cell-free fetal DNA from the pregnant woman's blood. These tests, feasible as early as the fifth week of pregnancy, will require a simple 10 milliliter blood draw, avoiding the invasive procedures of amniocentesis or chorionic villi sampling, with their attendant high costs, discomfort, and miscarriage risks.

Cell-free fetal DNA testing will be able to reveal aneuploidies, single gene diseases, broad disease risks, and some non-disease traits: certainly sex and probably skin, eye, and hair color; hair type; likely height; and male pattern baldness, among others. The price of this analysis should be \$1000 or less. At that price, insurers will likely find this testing cost-effective for every pregnancy, not just high-risk pregnancies. Safe, early, comfortable, broad, and fully insured fetal genetic testing is likely to be used by far more than the current 1.5 percent of pregnant women – probably fifty to eighty percent. As a result, abortions for serious genetic conditions will increase substantially and children born with such conditions are likely to be concentrated in populations particularly opposed to genetic testing or abortion.

How should our society react? Should we encourage, discourage, or view as neutral this kind of testing? What kind of quality regulation should we impose on this testing? Should we – can we – impose other, non-quality regulation, aimed either at preventing prospective parents from terminating pregnancies based on some genetic characteristics or at preventing testing (or communication of test results) for some kinds of genetic risks? We all will soon have to answer these questions, but we are only beginning to pose them.

Pre-implantation Genetic Screening: Socioeconomic Stratification and Equality of Opportunity

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by Austin Alexander, Siftgen, Inc.

As the era of personal genomics comes of age, genomic information will play an increasingly important role not only in medical decisions but also in reproductive decisions. Already, preimplantation genetic screening (“PGS”, also referred to as preimplantation genetic diagnosis or “PGD”) is being used to screen embryos prior to implantation to select those without known genetic diseases such as cystic fibrosis and Huntington’s disease. As the understanding of the genetic contribution to diseases and traits increases, and as the cost of full genome sequencing decreases, it will become feasible to use PGS to target the full spectrum of genetic diseases.

With time, it will also become possible to preferentially select for beneficial genetically-influenced traits such as athletic talents or high intelligence. Aside from the ethical and religious concerns with such a technology, one of the main social concerns is the potential for increased socioeconomic stratification. If access to PGS were limited to only those prospective parents with the financial means to pay for it, their progeny could gain a perpetual advantage over those whose parents could not afford it. Reactionary fear of this possibility has led to a backlash against PGS, with various groups calling for restrictions or outright bans on its use. This backlash is likely to increase with the number of traits that can be screened for, which could result in diminished access to those families who could most benefit from this technology. In order to ensure continued access to PGS while mitigating the risk of increased socioeconomic inequality, a new policy of universal access will be needed.

To-date, most of the debate surrounding PGS has been focused on whether and how to restrict access. Religious groups such as the Catholic Church have come out against it. In the UK, its use is governed and limited by the HFEA. In a few other countries such as Germany, Ireland and Switzerland, PGS is banned. This approach of limiting reproductive freedom denies parents the opportunity to avoid passing on deleterious mutations to their offspring and can result in unnecessary abortions with parents forced to choose after pregnancy has commenced rather than at the less destructive pre-implantation stage. These limitations also deny future generations the opportunity for improved health and quality of life and would result in many children born with diseases that could otherwise have been avoided. Furthermore, this policy could have the unintended effect of exacerbating one of the very problems it is intended to solve: by limiting access, it promotes increased reproductive tourism by affluent potential parents who can afford to travel to more favorable jurisdictions in order to gain access, thereby creating an even greater financial hurdle and increasing socioeconomic stratification.

A better approach would be focusing on how to make the option of PGS accessible to all those who wish to use it. With equal access, parental financial differences would become irrelevant and the playing field would be leveled across socioeconomic groups. While the choice of whether to use PGS should remain a personal one made by the parents, its widespread use could also result in public health benefits with the potential for numerous heritable diseases to be eliminated in much the same way as the widespread use of vaccines has eradicated many major infectious diseases in the developed world. Although some may argue that providing increased access is unaffordable, the costs of PGS could be more than offset by the lifetime healthcare cost savings of its beneficiaries. These benefits and the opportunity to minimize socioeconomic divergence all point to the need for policies that promote increased access for all groups, in direct opposition to those policies of restriction that have been pursued so far.

How will we handle the capability and responsibility of creating human life according to spec?

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Esther Dyson](#), [EDVenture Holdings](#).

Until now, we have been able to create humans but not to design them. We know that in many cultures, female infanticide is practiced for the purpose of having a son who can care for the parents in their old age. That practice is frowned upon by most.

But it gets creepier. We already have something of a taboo against creating humans for a purpose other than their own existence. That was behind the fascination with Jon Benet Ramsay, the little murdered girl who seemed to exist more for her mother's gratification than for her own. A recent movie, "My Sister's Keeper," posited a girl who had been conceived in vitro to provide tissues and body parts for an ailing sister (but the movie punted this question in the end). Like so many modern crimes, the issues revolves less around objective facts than around intent and responsibility....

We can look to euthanasia for some parallels and differences. Euthanasia may look like pain relief – or pain relief may look like euthanasia. Doctors are often involved and can properly guide people in making the right choices. In the end, individuals should have the right to control their own life and death. That choice is not available to the individual at the beginning of life; therefore, would-be parents need to be encouraged to be clear about their own motivations. This is not a new issue, but it becomes a bigger one when parents can choose not merely to **have** a child, but what kind of child to have. Parents have always had to resist the temptation to control their children's lives. Now they have an even greater ability to do so – and the burden of living with their choices.

So, I think it would be reasonable to have a law against creating a person for any other reason than that person's own existence. The point of the law is certainly enforcement, but it's also to have a clear statement of what is acceptable and what is not. A person who exists primarily for another's benefit is a slave, and we condemn and outlaw that – even though such exploitative relationships certainly exist and may look consensual from the outside.

Laws are an expression of society's norms, and this norm of non-exploitation deserves clear affirmation.

Self Explorimentation

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Mike Cariaso](#), [SNPedia](#).

Anyone with \$400 is now able to learn much about their own DNA. This door is already open, but we've not yet had time to determine where it leads. The first few steps are small, and seem to be within the bounds of what is broadly acceptable. Stepping further seems to go beyond what we can reliably know today.

A DNA variant named [rs3892097](#) increases risk of Parkinson's Disease when exposed to pesticides. Is my insurer/employer allowed/obligated to test for this genotype and to prevent me from working with materials which are particularly hazardous to me? Do I have the right to work around pesticides? Similarly [rs1799807](#) increases sensitivity to nerve agents such as VX and Sarin, how should the military factor this into their planning?

Today's grey market for performance enhancing drugs

<http://www.guardian.co.uk/science/2009/sep/20/neuroenhancers-us-brain-power-drugs>

adopts tomorrow's genetic engineering medicines. An injection restores normal vision to the color blind

<http://www.nature.com/nature/journal/vaop/ncurrent/full/nature08401.html>

Using a slightly different DNA pattern makes it just as easy to give

<http://en.wikipedia.org/wiki/Tetrachromacy>

to those afflicted with merely normal vision. Adding fully functional gills to the human body becomes a safe and similar procedure. World hunger ends when we add chloroplasts.

Genetic Exceptionalism and the Precautionary Principle

This commentary in the Genomics Law Report's ongoing series [What ELSI is New?](#) is contributed by [Alan E. Dow](#), Vice President of Intellectual Property and Legal Affairs, [Complete Genomics, Inc.](#)

One important issue that must be addressed in the field of genomics and / or personalized medicine is “genetic exceptionalism.”

Genetic exceptionalism is the concept that genetic information is inherently unique and should be treated differently in law than other forms of personal or medical information.

There are several reasons for such special consideration: genetic information can predict disease occurrence in a person and their blood relatives; it uniquely identifies a person; and it can be used to discriminate and stigmatize individuals. While these issues deserve attention and steps should be taken to protect people, over-regulation could limit our ability to investigate how genetic information predicts disease and improve medical outcomes.

Some people appear to equate genetic information and personal identity. For such people, protecting an individual's genetic information is tantamount to preserving their “respect... as an individual and as a member of the human species and recognizing the importance of ensuring the dignity of the human being” (Convention for the Protection of Human Rights and the Dignity of the Human Being with Regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, 1997). Such a belief can translate into over-regulation.

Opponents of a new technology can fight its introduction by applying the “precautionary principle.” A strong form of the precautionary principle would require that, when “potential adverse affects [of an activity] are not fully understood, the activities should not proceed” (United Nations World Charter for Nature, 1982). The burden then shifts to proponents of the new technology to prove that it will not cause harm. A more nuanced version of the precautionary principle would require “cost-effective measures” in the face of threats of serious or irreversible damage, even where there is a lack of full scientific certainty regarding the threat (e.g., Principle 15 of the Rio Declaration, 1992). What is considered to be “serious damage” or a “cost-effective measure” can appear very different to legislators and those being regulated – as would be appreciated readily, for example, by companies that have sought to introduce genetically-modified crops into Europe since the 1980s.

It would be tragic if genetic exceptionalism led to regulation that denies the public the tremendous benefits of the genomics revolution as a result of misplaced concern for human respect and dignity.

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