DNA & EHR – Six Letters Spelling the Future of Health Care

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In an effort to provide quality healthcare to millions of uninsured Americans, President Obama recently pledged to allocate $10 billion per year for the next five years as an incentive for healthcare providers to adopt Electronic Health Record (EHR) systems.¹ Proponents suggest that EHR implementation has the propensity to simultaneously reduce cost and improve the quality of care; thus, ultimately resulting in broader access to the United States healthcare delivery system.² Alternatively, as many politicians and healthcare industry representatives are quick to cite, the potential for rapid EHR implementation could exacerbate existing problems and pose enormous risks if the systems are

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adopted in haste and without adequate systemic structures in place to receive them.³

This article will discuss the role EHRs will play in the efficacy of 21st century medical care innovations and provide an overview of the intersection of genetic information (GI), defined as heritable biological information determined by nucleotide sequencing tests for purposes of this paper⁴, and EHR. Part one identifies and explores the therapeutic and research applications of genetics and EHR. Part two appraises the potential benefits of those applications, such as reducing healthcare costs and improving access to care.

The final part examines potential concerns that arise from implementing GI in EHR systems. Specifically, it discusses the legal framework and its role to maximize interface benefits, while preventing potential abuse and gesturing toward the deficiencies in the current system. Finally, this article identifies the tension between genetics and EHR. This intersection can improve access to care by reducing error, improving care and eliminating diseases, but could also facilitate discrimination by insurers in the absence of adequate protections, thereby reducing access to care.

I. GENETICS & EHR: APPLICATIONS

There are several applications for electronically available GI in the modern medical environment. Generally, these applications can be divided into two subcategories: those that facilitate diagnostic and therapeutic clinical medicine and those facilitate research. Although we can conceptually divide the clinical setting from the research setting to dissect each, this is an artificial dichotomy. Because clinical care for the individual patient is facilitated and improved by EHRs, this enables health information to be used in research and thus, results in diagnostic and treatment advances that improve patient care.

First, the ability to diagnose unexpressed genetic conditions before the individual becomes symptomatic enables preventative treatment for several conditions. For instance, some genetically inherited diseases can be delayed if not entirely avoided by preemptive interventions. Recent research indicates that if detected early, the onset of Alzheimer’s disease can potentially be delayed through health behavior counseling. These types of medical interventions are facilitated by incorporating genetic profiles into vast databases that aid physicians to identify “at risk” individuals and populations for the prophylaxis and screening

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5 See infra p.3.
7 See supra p. 2.
Interventions in early disease stages reduce healthcare costs, improve outcomes for patients, and reduce the stress on the healthcare system.

Second, GI electronic applications allow clinicians to have access to a patient’s GI along with other relevant health information that is imperative in the U.S. healthcare delivery system, which has gradually become more fragmented. For example, due to the psychological and cognitive elements of the disease, an Alzheimer’s patient diagnosed by a general practitioner would benefit from a mental health professional having the same access to the patient’s GI because it may facilitate anticipation, assessment, and treatment of certain symptoms.

Furthermore, when applying GI electronically in a therapeutic setting, the availability of this information plays a crucial role in genetics research. Information in individual patient files, such as genetic data and clinical scenarios, can be stripped of identifiable information and pooled into massive databases. These massive databases are used for dense statistical research projects as well as diagnostic tools that are essential to future medical progress.

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13 Interview with Lama Eldahdah, Genetics Counselor, Reproductive Genetics Institute, in Chicago, IL. (Mar. 27, 2009).
14 See infra pp. 3-4.
16 Id.
In the United States, academic and research institutions, such as Vanderbilt University and Marshfield Clinic, have developed systems to pool EHRs by de-identifying GI obtained during clinical diagnosis and treatment. To obtain access to the information, opt-out provisions in annual Health Information Portability and Accountability Act (HIPAA) mandated consent forms are used.\textsuperscript{17} These systems utilize complex algorithms to strip the “shadow” versions of the main EHR databases from all identifying information.\textsuperscript{18} Last year, Vanderbilt’s system contained information for over 50,000 patients, and acquired an average of 700 new records per week.\textsuperscript{19} Vanderbilt’s system is comprised by two distinct, but mutually supporting databanks where the EHR system and the “shadow-databank” DNA repository had de-identified information compiled through discarded blood samples.\textsuperscript{20} Similar to Vanderbilt’s design system, the Marshfield Clinic Genetic Biobank is at the center of a Wisconsin-wide genetics research initiative that began in October of 2007.\textsuperscript{21} The initiative goals include using database information to target certain diseases, including attempting to determine genetic components to increased risk for heart attack, as well as diabetes and its complications.\textsuperscript{22}
II. APPRAISING THE BENEFITS

The essential benefits of electronically stored GI are similar to the general benefits of EHRs, which include a greater efficiency and the potential to reduce medical error. As noted above, the accessibility to an individual’s health information improves the care coordination across a variety of providers and points of access. Thus, electronically stored GI has the potential to eliminate many costly risks of conflicting orders, duplication of treatments, and medical errors that harm patients, which can increase treatment costs. This is crucial protection in a society where four out of every five physicians are specialists, and a Medicare patient with a single chronic condition will see on average seven physicians per year.

Another primary justification to adopt EHR systems is the ability to reduce institutional costs. Studies suggest that successful EHR implementation can reduce costs in a variety of ways. For example, direct cost-savings occur, such as administrative costs associated with redundant medical tests, when the EHR system is substituted for the traditional paper system. Costs-savings are enhanced by indirect benefits, such as care coordination or reduced patient safety risks that improve the quality of care, and therefore, results in long-term cost-

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23 Emanuel, supra note 11, at 60-61.
24 See supra p. 3.
27 Emanuel, supra note 11, at 61.
28 See supra p. 3.
29 Hillestad et al., supra note 26, at 1103-04.
savings to the patient. These healthcare cost reductions have the potential to reduce the immense stress on our current healthcare system and consequently, improve access to care.

Furthermore, the availability of EHR allows patients to become more actively involved in their own care. Allowing patients to observe documented changes in their health behaviors, such as lower cholesterol because of diet change and exercise, provides immediate, positive reinforcement, and encourages compliance. This also provides physicians a forum to supply “information therapy,” making pre-screened information available to patients, thereby aiding their understanding medical conditions, treatments, and other relevant information. This improves the physician-patient dynamic, facilitates communication by helping to identify additional health concerns, allows the freedom to choose the maximally effective treatments, and engages the patient in their care.

Similar potential exists for greater care coordination, increased patient involvement, and enhanced patient autonomy with regard to electronically available GI. Because access to GI facilitates reproductive autonomy, this

31 Samuel J. Wang et al., A Cost-Benefit Analysis of Electronic Medical Records in Primary Care, 114 AM. J. MED. 397, 397 (2003).
32 See EMANUEL, supra note 11, at 126.
33 Laura Landro, Online Records Get Patients Involved in Care, WALL ST. J., Mar. 18, 2009, at D1.
35 Steve Lohr, A Hospital is Offering Digital Records, N.Y. TIMES, Apr. 6, 2009, at B3.
36 Landro, supra note 33, at D1.
37 See supra pp. 5-6.
enables parents to make more informed decisions about health risks that might affect potential children.\textsuperscript{38} For instance, Preimplantation Genetic Diagnosis, (PGD) enables genetics professionals to evaluate embryos for genetic markers before in vitro fertilization is performed.\textsuperscript{39} Furthermore, amniocentesis, a prenatal testing procedure that identifies the probability of genetic abnormalities, facilitates reproductive autonomy by allowing parents to choose whether to terminate a pregnancy when the fetus has a high likelihood or confirmed diagnosis of a genetic disease.\textsuperscript{40}

“Information therapy,” a patient resource supported by patient-accessible records that utilizes internet applications, is a valuable tool for educating patients about already expressed genetic conditions, and can guide patients in making decisions based on GI.\textsuperscript{41} Patients who possess genetic risk factors for certain diseases may need a lot of support to make informed reproductive decisions.\textsuperscript{42} In addition, difficult decisions about prophylactic treatments, such as radical mastectomies for patients that carry certain mutations that indicate a high likelihood that they will develop breast cancer (also known as “BRCA analysis”),


\textsuperscript{39} Id.


\textsuperscript{41} Guilherme Del Fiol et. al., Integrating Genetic Information Resources With an EHR, ANNUAL SYMPOSIUM PROCEEDINGS ARCHIVE, AM. MED. INFO. ASS’N, 904 (2006).

\textsuperscript{42} Interview with Lama Eldahdah, supra note 13.
require a patient to understand complicated information regarding different treatment options prior to making life-altering treatment decisions.\textsuperscript{43}

Furthermore, electronic GI can improve coordination within clinical genetics.\textsuperscript{44} In many instances, genetic counselors provide pre-testing counseling, and then request that a physician write an order for genetic testing.\textsuperscript{45} Increased coordination of GI exchanges in the clinical setting would avoid situations where physicians incorrectly receive laboratory test results for patients that they did not counsel, and thereby eliminate the risk of giving an inaccurate interpretation or misapplication.\textsuperscript{46} This situation is especially problematic when the results are interpreted by a physician without training in genetic counseling.\textsuperscript{47}

In other scenarios, physicians without adequate genetics knowledge order tests without consulting genetic professionals.\textsuperscript{48} EHRs increase the ability for geneticists and other genetic professionals who work in laboratory settings to discover orders for inappropriate tests, which can avoid unnecessary fiscal costs to the patients and make genetic testing more affordable.\textsuperscript{49} The Mayo Clinic adopted an EHR system that includes its genetics laboratories to avoid the negative outcomes in the aforementioned clinical genetics scenarios.\textsuperscript{50} This can also spare patients the unnecessary emotional strain of being informed that they

\textsuperscript{44} See infra p. 7.
\textsuperscript{45} Interview with Lama Eldahdah, supra note 13.
\textsuperscript{46} Id.
\textsuperscript{47} Id.
\textsuperscript{48} Id.
\textsuperscript{49} Id.
tested positive for an unpreventable condition due to a test that was performed against the patient’s wishes.\footnote{Interview with Lama Eldahdah, supra note 13.}

\section*{III. Assessing the Risks & Developing the Law}

The development of treatments and prevention for genetic diseases, which enhance social justice by eliminating genetic disease and acting to level the genetic playing field, is aided by technology such as EHRs.\footnote{\textsc{Allen Buchanan et al.}, \textit{From Chance to Choice: Genetics & Justice}, 16 (Cambridge University Press 2000) (2000).} However, these technologies are susceptible to being manipulated as a justification for discriminatory motives,\footnote{Karen H. Rothenberg & Sharon F. Terry, \textit{Before It’s Too Late—Addressing Fear of Genetic Information}, 297 \textit{Science}, 196, 196-97, (July 2002).} such as the potential denial of insurance coverage.

Moreover, professional ethical questions arise from the significant uncertainty surrounding the meaning of many genetic diagnostic tests.\footnote{See \textsc{Buchanan et al.}, supra note 52, at 313.} For instance, many genetic tests enable physicians to inform patients of their rough statistical likelihood of developing a particular condition based on their genetic profile.\footnote{\textit{Id.}} For instance, a BRCA analysis determines a woman’s genetic predisposition and statistical probability of developing breast and ovarian cancer by identifying certain mutations.\footnote{\textit{Id.}} BRCA analysis alerts high risk women in early adulthood so that they can benefit from earlier and more regular screening.\footnote{\textit{Id.}} However, these test results may also induce women to have imprudent
prophylactic mastectomies, which have serious physical, psychological, and emotional consequences.\textsuperscript{58} Beyond intrapersonal costs, these unnecessary procedures are increased and result in avoidable monetary resource expenditures for both the patient and insurance companies.

Many practical considerations must be made prior to implementing widespread electronic GI into the medical profession.\textsuperscript{59} For instance, there is a need to make sections of patients’ health records confidential and protected through restricted access.\textsuperscript{60} Thus, several infrastructural shortcomings must be addressed to ensure patient privacy is not violated and not to provide pharmaceutical companies a vehicle to conduct market research.\textsuperscript{61}

In 1996, HIPAA imposed restrictions to the availability of individual identifiable health information, or health information that is traceable to the patient.\textsuperscript{62} HIPAA has provisions for genetic-specific protections against discrimination and prevents insurers from precluding eligibility because of GI\textsuperscript{63} Furthermore, the “Privacy Rule” enforces requirements for information distribution that is classified as protected health information.\textsuperscript{64} Under HIPAA, “protected health information” is individually identifiable health information

\begin{itemize}
\item \textsuperscript{58} Id.
\item \textsuperscript{59} See Gross, supra note 3.
\item \textsuperscript{60} Id.
\item \textsuperscript{61} E.g., Matt Fedoruk, DeCodeing Iceland’s DNA, 4 THE SCI. CREATIVE Q. Aug. 2003, http://www.scq.ubc.ca/decodeing-icelands-dna/.
\item \textsuperscript{63} Id. at § 702(b)(1)(B).
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created or received by a “covered entity” such as healthcare providers, employers, life insurers, healthcare clearinghouses, and others. Protected health information also relates to a patient’s physical or mental health condition and payment information.

Congress enacted the Genetic Information Nondiscrimination Act (GINA) to facilitate the development of genetic technologies, to continue research, to enhance basic genetic knowledge, and to protect individuals from discriminatory misuse of GI for health insurance and employment purposes. GINA prohibits group health plans and health insurance providers offering group health insurance coverage from establishing rules of eligibility or continued eligibility for an individual to enroll under a plan based on several factors, including GI. Furthermore, this legislation prohibits a group health plan to require higher premiums, collect contribution payments from individuals with certain genetic conditions, and require genetic testing.

GINA prohibits discrimination against enrollees in large group health plans on the basis of GI. However, it does not offer protection for individuals seeking private or small business health insurance, life insurance, disability insurance, long-term care insurance, nor does it address discrimination against patients that are already symptomatic of a genetic disorder, and those that learn

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65 Id.
66 See id.
68 Id.
69 Id.
70 Id.
they have a condition through non-genetic predictive testing.\textsuperscript{71} Thus, large segments of the population, including approximately twelve million individuals who have private insurance at any given time, will not be protected from discrimination that may bar them from health insurance.\textsuperscript{72}

When GINA’s first health insurance regulations take effect in May of 2009,\textsuperscript{73} followed by employment regulations in November of 2009,\textsuperscript{74} it will become more evident where its protections fall short. Without sufficient protection from GI discrimination, improvements in access to care that might be achieved through the increased continuity of care, reduction of error, and the benefits of preventative treatment\textsuperscript{75} might be supplanted by the reduced access caused by discriminatory insurance practices.

IV. CONCLUSION

This article demonstrated that electronic applications of genetic technology afford many opportunities for modern societies to reduce or eliminate genetic diseases that drain limited healthcare resources.\textsuperscript{76} First, the intersection of genetics and EHR presents the potential to foster enhanced individualized

\textsuperscript{71} See id.
\textsuperscript{72} See GARY CLAXTON, HOW PRIVATE INSURANCE WORKS: A PRIMER 1(Kaiser Family Foundation 2002) (Apr. 2002).
\textsuperscript{73} GINA, supra note 4.
\textsuperscript{75} See supra pp. 2-6.
\textsuperscript{76} BUCHANAN ET AL., supra note 52, at 8-11.
medicine. For instance, by combining genetic knowledge with EHR, physicians gain the ability to utilize vast GI databases to aid them in treating patients. Second, these databases would facilitate otherwise impossible research, allow scientific advancements, and potentially eradicate devastating genetic diseases that result in decreased personal and healthcare costs.

However, these tremendous potentials for growth and improvement are harboring hidden risks of discrimination, privacy violations, and a slew of other negative consequences. Thus, continued development of protective legislation and public policy, led by robust public discourse, must guide the course of this technology. This is particularly evident with the EHR application as a way to store and exchange GI, where the great potential to reduce the stress on the healthcare system is tempered by concerns about opportunities for discrimination, particularly on the basis of health insurance.

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77 Hillestad et al., supra note 26, at 1113.
78 Glaser, supra note 10.
79 Id.
80 See supra p. 5.
81 See supra pp. 6-10.