INTEROPERABILITY’S ROLE IN STRIVING FOR PRECISION MEDICINE

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Abstract: President Obama announced the Precision Medicine Initiative in his 2015 State of the Union address, a research effort to find treatments that are tailored to specific genetic profiles and characteristics. This personalized and context-specific treatment approach will require big data analysis of patient outcomes and their genetic sequence, which must then be accessible and comprehensible to caregivers. The initiative must therefore be able to reconcile genetic data, demographic information, and health information, in the electronic health record. To achieve data interoperability, the organizations engaged in the initiative must confront questions of data privacy, standardize data exchange, and incentivize the adoption of data-sharing technology.

I. INTRODUCTION

When President Obama proposed the Precision Medicine Initiative (PMI) during his 2015 State of the Union address, interoperability was listed as a key investment. In fact, it is the central element to all of the PMI’s objectives. The PMI requires an integrated, interoperable network between laboratories, healthcare organizations, public

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1 The Precision Medicine Initiative is a long-term research endeavor, involving the National Institutes of Health (NIH) and multiple other research centers, which aims to understand how a person's genetics, environment, and lifestyle can help determine the best approach to prevent or treat disease. What is the Precision Medicine Initiative, National Institutes of Health, https://ghr.nlm.nih.gov/primer/precisionmedicine/initiative.


health registries, payers, and researchers. An extraordinary degree of collaboration is now underway, and will be necessary to enact new policies as the landscape of healthcare changes. Multiple agencies will need to work in concert with these developments to make personalized treatments available, as opposed to “one size fits all” healthcare treatment. The PMI not only aims for patients’ genetic sequences to be incorporated into their electronic medical records, but also aims to deliver “the right treatments, at the right time, every time to the right person.” This personalized approach to healthcare delivery requires big data analysis of patient outcomes to provide individualized and context-specific treatment, which must be accessible and comprehensible to healthcare providers.

This article highlights the efforts being made to promote data sharing and interoperability between different healthcare organizations in order to provide better patient outcomes. The ensuing discussion examines how federal agencies and healthcare organizations engaged in PMI are building upon such efforts to confront issues of


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privacy, standardization, and adoption. This article ends with a call for integration of genetic data that can incentivize interoperability for precision medicine.

II. CURRENT STATUS OF INTEROPERABILITY

A. Background of Healthcare Technology Interoperability

For genetic data to be applicable in healthcare, it is not enough to exist in a text document alone; the genetic data must be integrated with data in electronic health record systems (EHRs) in order to provide clinicians with effective clinical decision support.10 The Office of the National Coordinator for Health Information Technology (ONC) regulates, certifies, and promotes the use of EHRs through an incentive program.11 The EHR incentive program, Meaningful Use, which was authorized by the 2009 Health Information Technology for Economic and Clinical Health Act (HITECH), encouraged healthcare organizations to transition from paper charts to EHRs by providing incentives.12 The Meaningful Use program stipulates that healthcare providers must “meaningfully use” EHRs, evidenced by routinely performing a set of actions in the EHR, such as drug prescription ordering and lab result review, in order to receive certain

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12 See 75 FR 44314.
levels of reimbursements.\textsuperscript{13} By requiring these actionable tasks that depend on integrating with other systems such as laboratories and pharmacies, Meaningful Use compels interoperability with laboratory and prescription systems.\textsuperscript{14}

Prior to the adoption of EHRs, patient records were forwarded to external healthcare systems by mail or fax.\textsuperscript{15} A growing demand for the ability to share patients’ health information across organizations and public health registries electronically, now fuels the focus on interoperability between different EHRs.\textsuperscript{16} Such interoperability would allow healthcare organizations to transfer the full history of the patient, and would also more readily contribute patient data to public registries in order to advance population health measures.\textsuperscript{17} Data formats and qualitative standards can differ from one system to another, but to be interoperable, EHRs must utilize a semantic language.\textsuperscript{18} The ability to transmit patient health records to other healthcare systems using a standard format was added in 2014 as a requirement for the Meaningful Use program, requiring EHRs to be capable of allowing reconciliation of medications, problems, and allergies into the local

\begin{footnotesize}
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\item \textsuperscript{13} See 75 FR 44314.
\item \textsuperscript{14} See 75 FR 44314.
\item \textsuperscript{15} See Janice F. Mulligan and Mark R. VonderHaar, Is Pushing Health Care’s Green Button a Kill Switch for Patient Privacy?, 46 The Brief 20, 21 (2017).
\item \textsuperscript{17} See id.
\item \textsuperscript{18} See Rebecca S. Eisenberg, Robert and Barbara Luciano, Promoting healthcare innovation on the demand side, J Law Biosci (April 2017) 4 (1): 3-49.
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Various EHRs, however, are still working to achieve interoperability and adopt standardized terminologies to exchange electronic health information accurately.\textsuperscript{20} Thus, in the same way, a standard ontology that could incorporate genetic information with health information, make calculated assessments, and translate successfully into other systems, remains a goal of both PMI and of interoperable EHRs.\textsuperscript{21}

\textbf{B. Clinical Genomics Interoperability}

Organizations and researchers are prevented from sharing and combining their data with each other because health information is not consistently translated from one health system to another, in addition to the fact that genomic data does not typically exist within the EHR.\textsuperscript{22} The ONC has recognized this insufficiency, and published an Interoperability Roadmap.\textsuperscript{23} The document declares that the goal of interoperability within the next several years is that healthcare providers “routinely use relevant info from a variety of sources, including environmental, occupational, genetic, human service, and cutting edge research evidence to tailor care to the individual.”\textsuperscript{24} This calls for emphasis on standards development organizations, which create and maintain standards and data

\begin{itemize}
\item \textsuperscript{19} See 75 FR 44314; 45 CFR § 170.315(b)(2).
\item \textsuperscript{20} The Office of the National Coordinator for Health Information Technology, \textit{Connecting Health and Care for the Nation: A Shared Nationwide Interoperability Roadmap}, October 6, 2015, at vi.
\item \textsuperscript{23} \textit{Connecting Health and Care for the Nation: A Shared Nationwide Interoperability Roadmap}, supra note 20, at vi-viii.
\item \textsuperscript{24} Id. at 47, 75.
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formats for semantic terminologies and codes, to provide more specific implementation
guides in order to facilitate EHR development in support of those standards for
widespread adoption. The ONC contends that adoption of technical standards will
prevent noncompliant access or authorization to data, even amidst changing and complex
legal regulations of privacy and security, which have admittedly hindered development of
interoperable technology.

III. ESTABLISHING A FRAMEWORK FOR GENOMIC INTEROPERABILITY

A. Addressing Privacy and Security Complications with Genomic Data

A discussion of data sharing and interoperability naturally involves determining
the rules with respect to privacy and security of data. The rules governing data and
privacy within the Genetic Information Nondiscrimination Act (GINA) and Health
Insurance Portability and Accountability Act (HIPAA) were established to allay the
public fear and concern over misuse, discrimination, or unauthorized disclosure of
genetic data. Nevertheless, GINA applies only to discrimination by employers or health
insurers, but does not address how physicians should share or withhold that information

25 Id. at 24-27.
26 Id. at 16, 18-19.
27 Elizabeth Snell, Maintaining Health Data Privacy in Precision Medicine Push, HealthIT Security
medicine-push.
from health insurers if genetic information does exist in the EHR.\textsuperscript{29} State and federal statutes may also impose varying restrictions on what kinds of data should be withheld or require special consent.\textsuperscript{30} Splitting data between healthcare providers and organizations can hinder access to a patient’s complete health record and impact quality of care.\textsuperscript{31}

Furthermore, it is reasonable to expect that previously de-identified genetic data may pose greater risks of re-identifying individuals, given advances in genomic research and increasing integration of genomic data with health information. Studies have already shown that readily-accessible information online, in combination with genomic sequence data from an open research database, can identify individuals.\textsuperscript{32} Entities not restricted by GINA, such as life or disability insurance insurers, could therefore access health information regarding a consumer’s propensities to genetic diseases. This concern calls for greater legal protection and security measures.\textsuperscript{33} Privacy thus remains a complication for interoperability and exchange of genetic information.\textsuperscript{34}

The ONC has also partnered with the National Institute of Standards and Technology (NIST) in producing the PMI Data Security Principles Implementation Guide

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\item See \textsuperscript{29} U.S.C.A. § 1182; 42 U.S.C.A. § 2000ff-5.
\item See \textsuperscript{30} 29 USCA § 1182; N.Y. Public Health § 2782 (McKinney 2017).
\item Kulynych, supra note 33.
\end{enumerate}
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to provide clarifications especially for those organizations participating in the Precision Medicine Initiative, regarding privacy and data security best practices.\(^{35}\) This document serves PMI organizations with a broad guideline, and does not encompass existing rules and guidelines required under HIPAA and GINA.\(^{36}\) PMI organizations that adhere to the implementation guide must take additional measures to ensure HIPAA compliance, and should still perform their own security risk assessments.

**B. Application Programming Interfaces**

To advance one of the major objectives of the PMI, collecting patient health information for extensive genomic research, the ONC has partnered with the National Institutes of Health (NIH), the Office of Science and Technology Policy, and EHR vendors under the Sync for Science program.\(^{37}\) This collaboration will design the functionality and framework that will allow patient research participants to easily sync their health data from the EHR to the genomic research database.\(^{38}\) The framework relies on community-developed standards to create libraries of application programming interfaces (APIs), which serve as bridges of communication between different software

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\(^{36}\) Id.


APIs will allow various EHRs, external applications, and patients to share data without requiring extensive development collaboration.\textsuperscript{39} APIs must still depend on a common standard to allow data exchange to flow.\textsuperscript{40} The community-developed standard, Fast Health Interoperability Resource (FHIR), was developed by a privately funded workgroup of EHR vendors and healthcare organizations.\textsuperscript{41} FHIR provides a modular semantic language to exchange common health data elements between systems.\textsuperscript{42} While Sync for Science aims to extract health information into research databases, its sister program Sync for Genes aims to integrate genomic data into the EHR systems.\textsuperscript{43} Sync for Genes will also build on the FHIR infrastructure using APIs to permit genomic information to be pulled into the EHR at the point of care, with appropriate clinical context given to the healthcare provider.\textsuperscript{44}

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\item \textsuperscript{39} Elizabeth O'Dowd, \textit{Why Application Programming Interfaces Are Key for Healthcare}, https://hitinfrastructure.com/features/why-application-programming-interfaces-are-key-for-healthcare (last visited November 8, 2017).
\item \textsuperscript{40} O'Dowd, supra note 39; White, supra note 38.
\item \textsuperscript{44} Sync4Genes, http://www.sync4genes.org/ (last visited November 3, 2017).
\item \textsuperscript{45} Genomics Implementation Guidance, https://www.hl7.org/fhir/genomics.html (last visited November 8, 2017); Sync4Genes, supra note 44.
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IV. INTEROPERABILITY FOR CLINICAL GENOMICS

A. Incentives for Interoperability

The Precision Medicine Initiative is being spearheaded by Vanderbilt University, along with several other healthcare organizations, having received funding from NIH to pursue PMI. With the support of NIH funding, the PMI cohort organizations will initiate patient engagement and use APIs to exchange health information with the participants. APIs expectedly will be implemented on a larger scale by EHRs in the next few years but not necessarily for the purpose of advancing precision medicine. The adoption of APIs, which will become mandatory in 2019 as a criteria for Meaningful Use EHRs, will serve to institutionalize patients’ ability to extract their health information from the EHR to share with other compatible applications. Whether this interoperability requirement has a particular function or purpose is not specified in the Meaningful Use criteria. If APIs become commonplace among EHRs, the technical hurdles facing PMI

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48 See 42 C.F.R. § 495.24.
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are lowered but will still require EHR vendors and healthcare organizations to address the usability of the data.\textsuperscript{50}

Assembling, analyzing, and meaningfully presenting data requires significant resources.\textsuperscript{51} The ONC also acknowledges that the EHR incentive program alone is not a sufficient measure that will establish interoperability, but that policy and funding incentives must also be addressed to reduce pushback on information sharing.\textsuperscript{52} Shared savings programs and accountable care organizations, which depend on value-based payments, have helped to shift the focus onto general interoperability because of its ability to foster more efficient care coordination.\textsuperscript{53}

Because the FDA already performs analysis on the health and claims data provided by payers, and ultimately controls approvals for drugs and diagnostic tests, the FDA may be the driving force of introducing gene-targeted medicine practices.\textsuperscript{54} Healthcare payers have been suggested as a source for incentivizing clinical genomic applications, which require a marriage of genomic and health data.\textsuperscript{55} If payers agreed to cover the cost of genetic tests and collected data that can confirm the clinical validity of


\textsuperscript{54} See USC § 355(k)3; Eisenberg, supra note 50.

\textsuperscript{55} See Eisenberg, supra note 50.
those tests, payers could accelerate the arrival of targeted therapies and companion diagnostic tests into the healthcare space.\textsuperscript{56} While this strategy may not alone convince payers, the promise of improved quality through precision medicine is becoming an increasingly stronger argument.\textsuperscript{57} The Medicare Access and CHIP Reauthorization Act (MACRA) introduced a new payment model that indicates a shift away from a fee-for-service model towards a merit-based payment system and alternative payment models, both of which focus on quality and improved healthcare outcomes.\textsuperscript{58} The funding provided by NIH might provide the resources and incentive to progress PMI, but consistent support may still be restrained by future budget proposals.\textsuperscript{59}

\textit{B. Measuring Interoperability}

The Meaningful Use program of the HITECH Act can be attributed as a significant factor that increased EHR adoption since 2011, having provided financial incentives to adopt EHR systems that meet certification requirements of the ONC.\textsuperscript{60} However, HITECH did not properly define or encourage interoperability and

\begin{flushleft}
\textsuperscript{56} Id.
\textsuperscript{57} See Erin C. Fuse Brown and Jaime S. King, \textit{The Double-Edged Sword of Health Care Integration: Consolidation and Cost Control}, 92 Ind. L.J. 55, 64-65 (2016); Eisenberg, supra note 50.
\textsuperscript{58} See 114 P.L. 10 § 101; \textit{Connecting Health and Care for the Nation: A Shared Nationwide Interoperability Roadmap}, supra note 18, at 1-2.
\textsuperscript{60} Dustin Charles PMH, Meghan Gabriel PhD, Talisha Searcy, PMA MA, The Office of the National Coordinator for Health Information Technology, \textit{Adoption of Electronic Health Record Systems among U.S. Non Federal Acute Care Hospitals: 2008-2014}, April 2015.
\end{flushleft}
standardization at the outset, and currently health systems are readjusting to engineer and adopt standard ontologies in order to make their EHR systems transmit data meaningfully.\(^6^1\)

ONC’s Interoperability Roadmap pleads for government and private stakeholders to help expand health IT certification programs to develop and use testing tools that will regularly assess interoperability performance.\(^6^2\) This need is clearly demonstrated by the finding in 2017 that one EHR vendor was discovered to have faked ONC certification by coding certain drug codes into the software instead of integrating with a database, and by failing to satisfy requirements in transmitting patient data.\(^6^3\) To avoid falsifying interoperability, any certification requirements and incentives to promote precision medicine should be deliberate in defining and measuring interoperability.\(^6^4\) MACRA has set a requirement for widespread EHR interoperability by December 2018, and the ONC has defined two interoperability metrics simplistically: the proportion of healthcare providers who send, query, receive data; and the proportion of healthcare providers who use data incorporated from other sources.\(^6^5\) The ONC Interoperability Roadmap implies

\(^6^1\) See 75 FR 44314-01, July 28, 2010.
\(^6^2\) Connecting Health and Care for the Nation: A Shared Nationwide Interoperability Roadmap, supra note 18, at 21-22.
\(^6^4\) See Bloomberg BNA Q&A: Mass. Health Data Exec Looks at Future of Data Sharing, Apps, supra note 54.
that improved patient outcomes that result from interoperability are also measures of interoperability success, but development of those metrics will evolve over time through discovery.\textsuperscript{66}

V. CONCLUSION

In a world where precision medicine is fully realized, genetic test results and diagnostics will inform primary care physicians and specialist providers on best forms of treatment and prevention for a range of healthcare issues.\textsuperscript{67} There are limitless opportunities to continue funding research and incentivizing new patents in drug treatment and companion diagnostic tests,\textsuperscript{68} but meaningful scaled progress in patient care will only be made if the genomic and family history information is accessible in the healthcare setting, accomplished via interoperability standards.\textsuperscript{69}

\textsuperscript{66} See The Office of the National Coordinator for Health Information Technology, \textit{Connecting Health and Care for the Nation: A Shared Nationwide Interoperability Roadmap}, October 6, 2015, at 48.


\textsuperscript{69} See Teri A. Manolio MD, PhD, \textit{Implementing genomic medicine in the clinic: the future is here}, Genetics in Medicine 15, 258–267 (2013).
The current landscape of health information is still largely adjusting to interoperable technology and standards. The ONC’s proposed changes regarding EHR certification and API development should reduce the burden of implementing new interoperable technology. However, the drive to achieve interoperability between public registries, EHRs, and research databases, has largely arose out of the need to exchange patient health information. PMI organizations that have been provided with funding and collaboration to demonstrate PMI’s goal of sharing and using genetic data and family history between patients, researchers, and providers, and developing privacy and security policies. But for widespread adoption, the ONC must draw more attention to precision medicine, particularly to use cases of precision medicine that require integration of genetic data, and must create the demand for genetic data that can incentivize interoperability for precision medicine.

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70 See The Office of the National Coordinator for Health Information Technology, Connecting Health and Care for the Nation: A Shared Nationwide Interoperability Roadmap, October 6, 2015, at vi.
71 Id. at 13.