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ARTICLE

A qualitative study of prevalent laboratory information systems and data communication patterns for genetic test reporting

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PURPOSE: The availability of genetic test data within the electronic health record (EHR) is a pillar of the US vision for an interoperable health IT infrastructure and a learning health system. Although EHRs have been highly investigated, evaluation of the information systems used by the genetic labs has received less attention—but is necessary for achieving optimal interoperability. This study aimed to characterize how US genetic testing labs handle their information processing tasks.
METHODS: We followed a qualitative research method that included interviewing lab representatives and a panel discussion to characterize the information flow models.
RESULTS: Ten labs participated in the study. We identified three generic lab system models and their relevant characteristics: a backbone system with additional specialized systems for interpreting genetic results, a brokering system that handles housekeeping and communication, and a single primary system for results interpretation and report generation.
CONCLUSION: Labs have heterogeneous workflows and generally have a low adoption of standards when sending genetic test reports back to EHRs. Core interpretations are often delivered as free text, limiting their computational availability for clinical decision support tools. Increased provision of genetic test data in discrete and standard-based formats by labs will benefit individual and public health.

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INTRODUCTION

Genetic laboratory testing is a cornerstone of genomic medicine, with over 41,000 genetic tests for more than 12,400 conditions available through CLIA-certified labs in the United States [1, 2]. These tests can range from cytogenetic analysis and single variant testing to genome- and exome-wide sequencing results. Clinical genetic testing is a dynamic and information-intensive field, and the generated information can benefit individuals, their families, medical research, and public health [3, 4]. In 2011, the National Academy of Sciences released its report titled "Toward Precision Medicine: Building a Knowledge Network for Biomedical Research and a New Taxonomy of Disease." This report promotes and characterizes a network that integrates clinical genetic data with research findings to improve both the diagnosis and treatment of diseases [5]. This aligns with the United States' national vision and plan for an interoperable health IT infrastructure and a learning health system where genetic profiles and laboratory test results are computationally available within electronic health records (EHRs) for consumption by clinical applications such as clinical decision support systems (CDS), and to support clinical research [6–8].

Computable genetic testing results offer several opportunities for improved health-care services and research [9–12], and it is one of the items of Masys et al.'s desiderata for the integration of genomic data into EHRs [13]. However, few research studies have thoroughly investigated how laboratories process and exchange genetic testing information with health-care providers. Besides the medical value of understanding this area, there are business and strategic values to it. From a business point of view, the 2019 global market of laboratory information systems is estimated to be $1.8 billion and may reach $2.4 billion by 2024 [14]. Therefore, aligning these investments to national goals could increase health-care efficiency and decrease costs. In addition, reporting on progress toward interoperability and health information exchange (HIE) is part of the national roadmap [6].

To fill this knowledge gap, we conducted a two-part qualitative study. The first part aimed to characterize how genetic testing labs in the United States handle their information processing tasks, starting from receipt of test orders and ending with final results reporting. The second part investigated the labs' perspectives concerning biomedical informatics (BMI) interoperability standards and their expected benefits, implementation challenges, and possible motivational factors. The focus of the present report is on the first part, while the second part will be reported in a second publication.

MATERIALS AND METHODS

This study was focused more on BMI interoperability standards (e.g., Health Level Seven [HL7] standards, Logical Observation Identifiers Names and Codes [LOINC]) than on bioinformatics standards (e.g., sequencing data

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formats, and human genome reference versions). There are no sharp boundaries that can guide this differentiation (e.g., the standardization of gene nomenclatures, which we did not investigate). The interviewer tried to direct the interview to BMI standards that are more necessary for high-level interoperability by receiving clinical information systems. Throughout this paper, we use the word "standards" to refer to BMI interoperability standards, unless otherwise specified.

We followed a qualitative research method based on a thematic analysis of semistructured interviews [15, 16]. This method is appropriate to study HIE needs and processes where there is little prior research, as it is more likely to capture the full range of relevant themes [17, 18]. The study method consists of three steps:

Instrument development and implementation

Information was obtained from the laboratories in two parts: a pre-interview survey of objective background information followed by a telephone interview for discussion of more subjective questions. The University of Utah's REDCap platform was used to implement the pre-interview survey [19, 20]. The interview scripts and the survey questions were reviewed by the study team that was followed by peer reviewing and pilot testing. The actual interview scripts were customized according the survey results for each participating lab to avoid asking irrelevant questions. More details about the instrument development and implementation are provided in the second part of the study.

Lab review and recruitment

A list of US-based genetic testing labs was obtained from the National Center for Biotechnology Information Genetic Testing Registry (NCBI-GTR) [21]. Additional labs were added through Internet searches. We reviewed the web page of each lab to identify their business category (e.g., commercial, university affiliated, and reference labs), representatives, and if they still provide their testing services. We excluded irrelevant labs from the invitation list, such as research labs and labs that had discontinued their services. More details about the lab review process, exclusion criteria, and results are discussed in the other part of the study.

Invitation emails were sent to candidate labs describing the research focus and requesting completion of the pre-interview survey and participation in a semistructured interview. The interview questions were customized according to the pre-interview results of each lab to avoid asking irrelevant questions and to save interview time for discussion of the most relevant content. At the end of the interview, participants were asked to rate their interoperability level according to three available choices:

Basic interoperability allows the transfer of data messages without automatic interpretation, that is, the message type and content need human interpretation

Syntactic interoperability allows the transfer of automatically computable types of data messages with predefined structure, but the content is not computationally recognizable, that is, the message type and structure are computationally recognizable but the message content needs human interpretation

Semantic interoperability allows the transfer of discrete data, that is, automatically computable message type, structure, and content

Each of these choices was defined and consistently explained by the interviewer through all the interviews. Also, the interviewer encouraged discussion of the interviewee's perception of the current status.

System models development

The interviews were recorded, de-identified, transcribed, and analyzed by A.K. to develop the first draft of the system model for each of the interviewed labs to depict the laboratory's information systems and their key elements. The models serve as abstract representations of information processing and flow between the labs and their customers. We used this simple modeling approach to identify relevant patterns and differences among genetic labs. The models were iteratively reviewed by S.M.H. and revised through several cycles. After addressing the comments and required edits after each review, some of the models were consolidated into a single generic model due to similar patterns in processes and principal systems.

The final versions of the generic models were reviewed and discussed by a panel of experts (C.C.M., J.H.G., G.D.F., M.S.W., S.B.B., and B.R.J.) in BMI, HIE, interoperability standards, genetic testing, and lab information systems. The panel has experience in both research and clinical practice. The panel checked the models for both accurate reflection of the recorded transcripts and clarity. A.K. and S.M.H. updated the models according to the collected comments and recommendations.

RESULTS

Participating labs

Overall, 302 genetic testing laboratories were identified. After exclusions, A.K. and S.M.H. sent email invitations to 188 laboratories to participate in the study. Thirteen laboratories completed the pre-interview survey, and ten laboratories (5.3% response rate) participated in the full interview. The final participants consisted of a mix of private (4 labs), specialized (7 labs), national reference (5 labs), hospital-based (6 labs), and university labs (6 labs). A lab may have more than one description. Specialized labs only run genetic testing or a category of genetic testing such as pharmacogenetics. Private labs are companies or are affiliated with companies.

Most of the interviewees had 3+ years in their current position in the lab, with working experience of 6+ years. For the rest of the labs, the lab manager or CIO recommended specific interviewees after learning more about the study aims and scope. Eight interviewees had a background in genetics, four in informatics and information technology, and the remainder in biology, medicine, and chemistry. More details of the participants are provided in the Supplemental material.

Lab system models

Three generic models were abstracted from the ten lab system models. The generic system models differ in their internal and external business processes.

Generic model 1: one backbone system with additional specialized systems for interpreting genetic results. In this model (Fig. 1), the lab performs almost all of the lab tests and interpretations internally using a central laboratory information management system (LIMS). However, it also outsources some infrequently requested tests to other specialized labs. The communication between the LIMS and the specialized lab systems may be automatic, manual, or both. Procedurally, the LIMS receives the test order, the raw test results are obtained, and then the lab staff or an automated electronic interface sends the required test results and patient information to one or more specialized systems for interpretation. After the lab personnel interpret the results, the final report is uploaded to the LIMS and then transmitted to the hospitals and clinics—either to the LIMS of the ordering health system or directly to the EHR.

Generic model 2: one or more brokering systems that handle final report housekeeping and communication. In this model (Fig. 2), the lab mainly uses the services of one or more external interpretation companies. The primary lab does the sequencing, identifies the genetic variants of importance, and sends the variant information to the interpretation company which sends back the final reports in PDF and structured formats, e.g., XML or JSON. Lab staff manually upload the report from the specialty lab into the primary LIMS. Next, the LIMS automatically sends the report (both in PDF and XML/JSON formats) to the affiliated hospital EHR, either through a dedicated HL7 V2.x message or an ad hoc proprietary interface.

Generic model 3: one primary system for results interpretation and report generation. In this model (Fig. 3), the lab performs the sequencing and interpretation of tests ordered by hospitals and clinics, but it also provides sequencing as a service to other labs. The lab has one information system for handling the interpretation and delivery of the results. The lab staff manually handle the

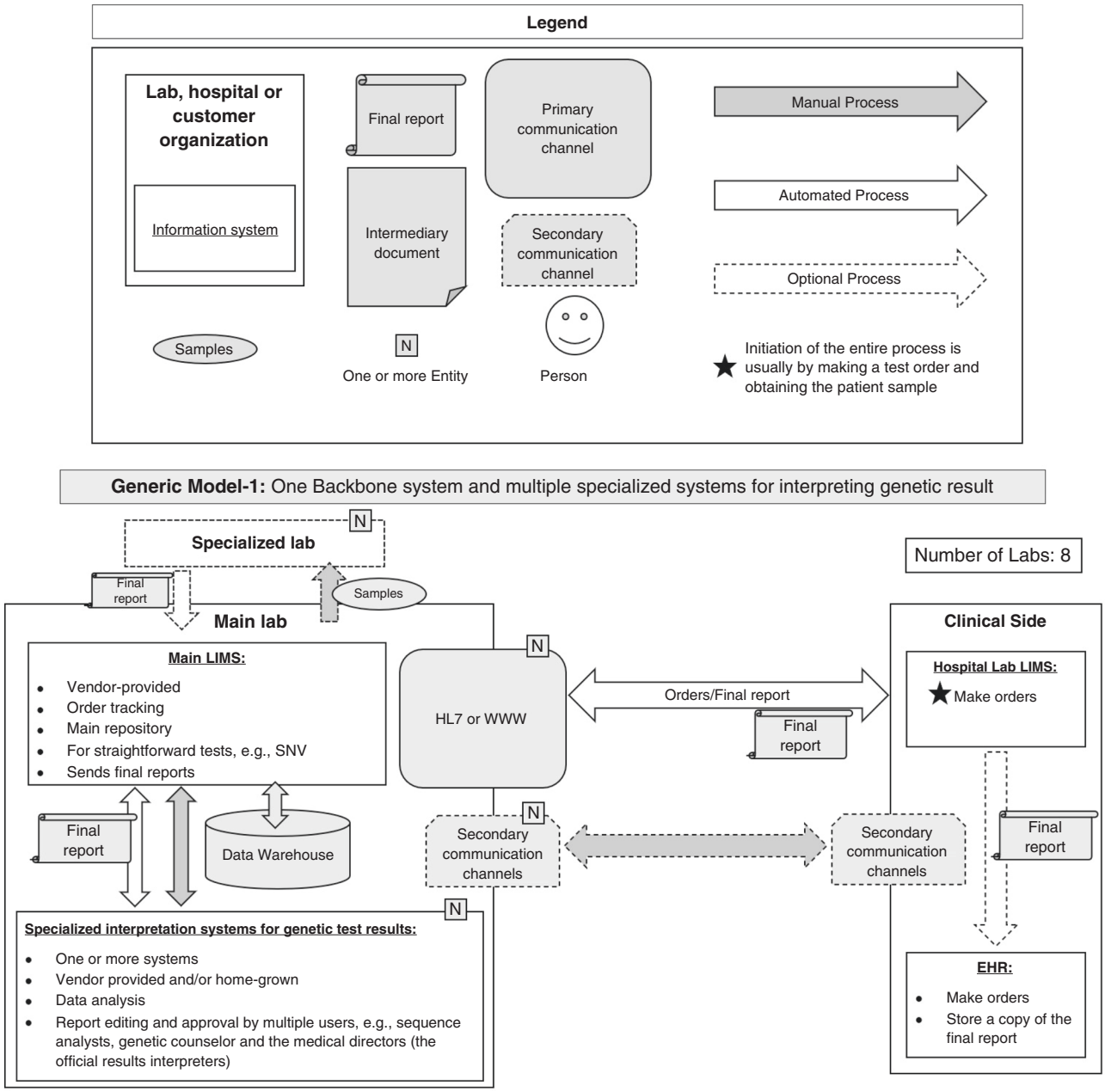

Fig. 1 Generic model-1 of the three identified models. In this model, there exists one backbone system with additional specialized systems for interpreting genetic results. A legend corresponding to all three generic models (Figs. 1–3) is provided at the top of this figure.

final delivery of the test results to clinics via email, and also upload the sequencing results to a secure cloud-based system.

Lab systems descriptions

Customers. The labs' customers vary significantly in terms of type and volume. Customers include affiliated or nonaffiliated health-care providers, researchers, and other labs. Some of the labs mainly serve a single type of customer, while others serve multiple types. Some labs serve external customers more than the affiliated hospital or provider. Another set of labs provide their services to other labs either to conduct a whole test or just to do the sequencing or interpretation. Nonaffiliated health-care providers may be other hospitals, cancer centers, or pediatric and adult care institutions and can be national or international. Some specialized labs may serve independent pharmacies or hospital pharmacies that want to offer pharmacogenetics tests.

Communication approaches. The interviewees from laboratories described that they communicate with their customers for two primary purposes. First, they need to collect some patient data that may help in interpreting the results. Second, they send the lab test reports back to the requesting clinicians. The following two sections explain the approaches used for each of these purposes.

Collecting patient data. Figs. 1–3 show some of the frequently used approaches to collect patient data by the corresponding generic lab model. However, the lab may collect relevant patient data using one or more of the following approaches:

- Paper requisition forms that are either attached to the test orders or specimens; or, the customers send scans of the paper requisition forms using email or fax.

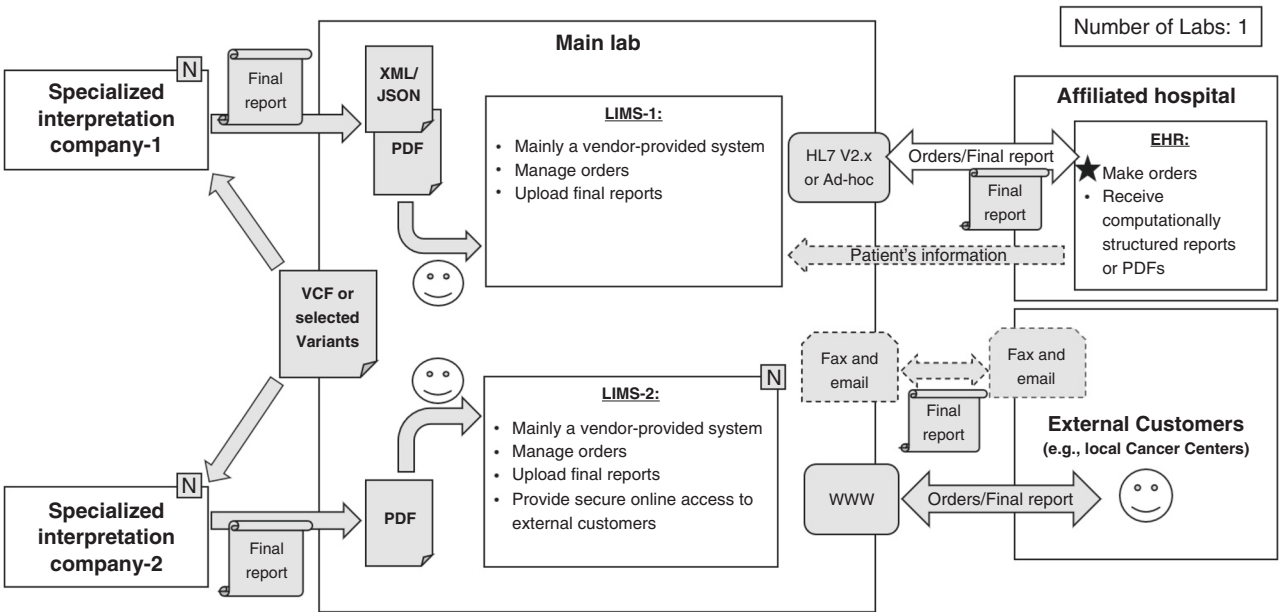

Fig. 2 Generic Model-2 of the three identified models. In this model, there exists one or more brokering systems that handle final report housekeeping and communication.

- Electronic forms that are filled out online through the lab web portal.
- Electronic data interfaces between the customer's EHR and the LIMS of the testing lab. These interfaces may use the HL7 V2.x messaging standard and LOINC codes. In other cases, communication happens via an ad hoc communication channel through a proprietary method, or the LIMS and the EHR are from the same vendor.
- PDF notes about the patient (e.g., clinical notes, history) sent via fax or email.
- Telephone calls.
- Online or face-to-face consultations.
- Lab staff access the patient's EHR and review/collect the required information using their credentials. This scenario is mainly for labs affiliated with hospitals.
- The lab staff glean information from previous testing done on the same patient.

Most of the interviewed labs collect required patient information as free text. However, this text may be formatted and organized into semistructured fields and sections. BMI standards are not used widely in this step. After retrieving these narratives, lab staff may extract some data and store them in a structured form using terminology standards in a LIMS (see "BMI interoperability standards" section below). This data extraction and coding may serve billing purposes or support the internal lab management of tests and analysis/interpretation.

Sending report to clinicians. The lab can use one or more of the following approaches to communicate final approved results:

- HL7 V2.x-based interfaces with the LIMS or EHR of the customer. In many cases, the HL7 message represents the results in PDF or scanned image format. Usually, these interfaces are implemented for affiliated hospitals or large/frequently ordering external customers.
- Downloadable PDFs from a secure web portal.
- Telephone calls in emergency/urgent cases to communicate preliminary results to the health-care provider. Usually, the lab will send a more detailed formal report later.
- Face-to-face or audio/video online consultations to explain the results in more detail.
- Faxing to a predefined HIPAA-compliant fax number.
- Secure emails, including results as PDF, MS Word, or PowerPoint files.
- A shared cloud-based storage system to share raw data with other labs that actually do the interpretive analysis. This is also seen in the research setting.
- An ad hoc communication channel for LIMS from the same vendor of the affiliated hospital EHR, the lab may provide the results as PDF or a structured XML or JSON format, which may include standard terminologies.
- LIMS user interface accessible from within the affiliated hospital EHR and some external customers. This user interface presents the genetic reports into the EHR as a dedicated tab or section. Also, results may be updated as soon as new evidence becomes available changing the interpretation of the results.
- Lab staff may upload the final report directly to the affiliated hospital EHR using the parent enterprise credentials. The uploaded report is typically in a PDF format.
- Delivery of hardcopy reports. This service is rarely used and mainly for international customers.

BMI interoperability standards. The following is a list of standards currently used by the interviewed labs as mentioned by the interviewees:

- LOINC for lab test names (i.e., not for document types, or specific information types). One interviewee doubted that the provider's information system uses the LOINC codes.
- Systematized Nomenclature of Medicine–Clinical Terms (SNOMED-CT) mainly to represent the diagnoses, i.e., not symptoms, procedures, or treatments. A combination of LOINC and SNOMED-CT is used by one of the labs to describe the test indication and the site of the specimen collection.
- International Classification of Diseases, Ninth, and Tenth Clinical Modification (ICD-9 and ICD-10-CM) are used mainly to represent the diagnosis for billing and insurance purposes, and sometimes for representing the indication for the test. ICD codes may be added to the electronic test order or may be added later by the testing lab staff, e.g., a technician or a coder. One of the lab information systems has an automated mapping between ICD-10 and SNOMED-CT. Sometimes, the

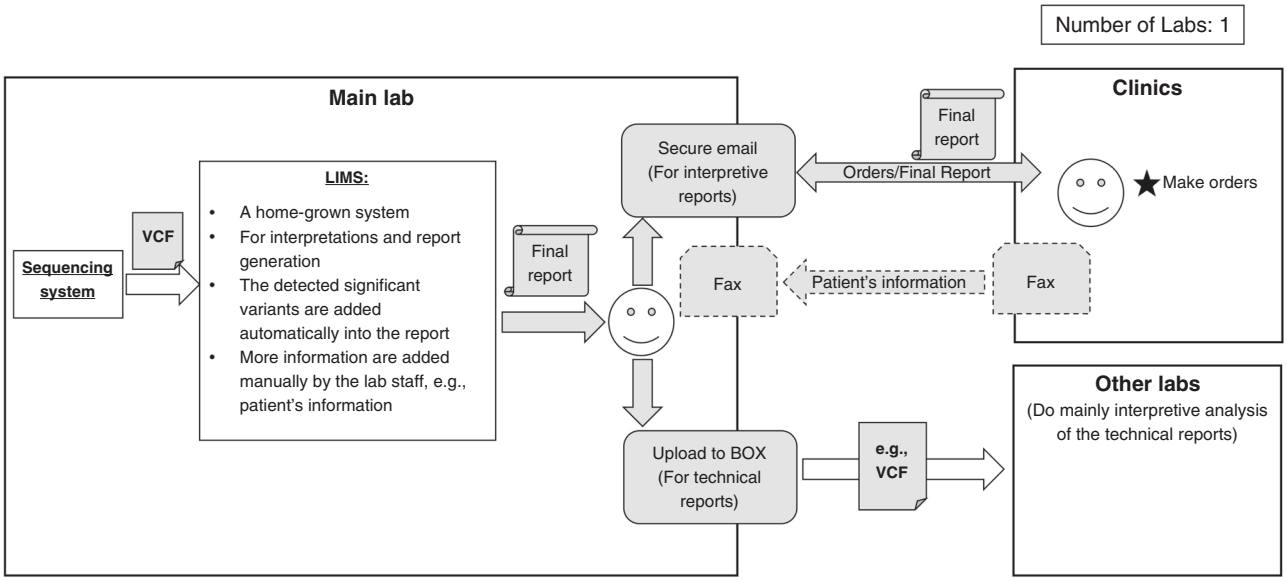

Fig. 3 Generic Model-3 of the three identified models. In this model, there exists one primary system for results interpretation and report generation.

lab returns the disease/condition name only, without the ICD code.

- Current Procedural Terminology (CPT), used exclusively for billing purposes. The codes are linked to specific descriptions of the methodology used for testing.
- Health Level Seven Version 2.x (HL7 V2.x) and HL7 V3 are used to send the final report from the testing lab to the provider system through a data interface. In almost all cases, the final report is included in the HL7 message as a PDF or scanned image. One of the interviewed labs built a dedicated customer-specific HL7 V2.x interface for each test type, when high test volumes justified the effort.
- HL7 Fast Healthcare Interoperability Resources (FHIR) are used to support medical applications under development by one of the labs. A specific FHIR genomic standard is under development.
- Human Phenotype Ontology (HPO) is used by two labs. One lab uses it to support downstream research groups from the same university. The other lab is a private sector specialized lab that conducts an intensive analysis of each testing case to provide a comprehensive report for critical decision making. Therefore, HPO terms are added to support the analysis process itself rather than for clinical decision making.
- RxNorm, a drug coding system, is sometimes used within pharmacogenomic test reports.

A few of the interviewed labs mentioned that they are currently not using any BMI standards, but they are planning to implement them in the future. No other standards than those described above were identified. However, ICD-10, CPT, and HPO were mentioned more frequently by labs that currently do not implement standards.

Paper and electronic forms. Although there is no use of paper forms internally in most of the interviewed labs, some paper forms still exist, e.g., requisition forms from customers, paper trails of the wet-lab phase of testing, and backup paper records of electronic documents. Additionally, labs may use faxes, document scans, PDF files, and MS Word documents, limiting the computational availability of the included information. The labs that are still using paper forms mentioned that they plan to implement a fully electronic information transfer system.

Outsourcing genetic tests. Genetic labs have various business models for performing genetic tests. Some labs perform all tasks related to genetic testing in house, including sample collection, wet-lab, sequencing, analysis/interpretation, and reporting. Others outsource some of these tasks to other labs or companies.

Some labs do the most frequently ordered tests in house while sending specialized or low frequency tests to a specialized lab to perform the analysis and the interpretation. Later, they receive the final report and route it to the ordering provider. Other labs do the sequencing of the genetic samples by themselves and then send the raw genetic results to a specialized company to perform the interpretation, e.g., pharmacogenetic testing. Later, they receive the final report, review and approve it, then enter or upload it to the affiliated hospital EHR or send it to their external customers. Labs that offer their genetic sequencing services to other genetic labs may be separate entities (e.g., private companies) or be affiliated with the ordering labs. For example, they may be from the same health-care provider, hospital, or university.

Collecting feedback. Generally, labs survey their customers (i.e., clinicians or patients) as a part of the College of American Pathologists (CAP) certification requirements [22]. Some of the labs collect this feedback proactively through regular surveys, while others may collect input only when there is a complaint or a recommendation for improvement. Some hospital-based labs receive feedback through personal contact or monthly meetings. The labs use feedback to implement a range of enhancements, including:

- Optimizing the report formatting, organization, degree of detail, complexity of the language, and including more information or clarifications
- Enhancing the communication of the changes in variant classifications when they become available
- Developing new software and updating existing software

Overall interoperability level. Six of the interviewed labs rated their overall interoperability level as basic, two at the syntactic level, and one between syntactic and semantic levels. One lab did not answer this question.

The three labs that described their overall interoperability level as syntactic or syntactic-to-semantic interoperability are hospital-based university-affiliated labs. They use either an HL7 V2.x interface or dedicated software with a clinical interface for sending lab test reports. Usually, these interfaces are built for high volume customers. The only lab that described its overall interoperability level as syntactic-to-semantic uses a LIMS provided by the same vendor as the affiliated hospital EHR.

DISCUSSION

Although genetic testing labs share common goals and may provide similar services, we found important variations in terms of the adopted workflow, the information systems used, and how results are shared with their clientele. Most labs operate at a basic level of interoperability with a few sending genetic results as discrete data at the syntactic level. The most common implemented standards are HL7 Version 2 messages and ICD codes. There is a very low adoption of terminology standards to represent the actual genetic results and no labs had yet implemented HL7 FHIR. This variability in the systems used and the generally low rate of incorporated standardized discrete data create challenges in interoperability and compromise the availability of test results in computable form. This is problematic because computable results are necessary for commonly used informatics tools, such as automated CDS. To maximize the benefits of the genetic test results, labs need to adopt common terminology and messaging standards. Although some of the results may be known by professionals working in this area, this study systematically polls existing system architecture and the interoperability infrastructure that supports genetic testing. We expect these findings to guide the implementation of standards-based HIE solutions for computable genetic lab test results, so that these test results can be consumed by applications such as CDS.

Although there are common elements, laboratory workflows are still very heterogeneous. Three models were constructed, each associated with some level of local customization. Other noninterviewed labs may utilize more than one of the models. Although electronic information systems are in use, manual tasks still exist in many cases, including data entry, report transfer from one system to another, or sending reports to ordering clinicians. While labs use the HL7 V2.X standard to receive orders and to report results, they usually present the core genetic interpretations as free text or in a semistructured format. This approach severely limits the computational availability of discrete genetic results. This finding aligns with previous studies concerning communication and use of patient information by hospitals in the United States, where data are exchanged between different health-care entities, but with suboptimal support for clinical decision making [23].

It is important to note that HL7 has provided a number of useful resources that analyzed genetic test reporting and provides solutions for many of the identified gaps such as the HL7 V2 Implementation Guide for Clinical Genomics [24], CDA Implementation Guide for Genetic Testing Reports [25], and the more recent FHIR Genomics Implementation Guide [26]. The adoption of interoperability standards and comparable LIMSs make genetic reporting more efficient and improve health-care services. In addition, the efficient utilization of genomic data entails other factors such as data security, privacy, lifetime storage, and the dynamic nature of genetic data and knowledge [27–29]. However, current EHR systems have limitations of processing and development of discrete and codified genetic data such as test result interpretation and clinical recommendations [10]. Also, there is a need for new models to have transparent and sharable CDS that make use of emerging AI tools [27, 30]. Both LIMS and EHRs should also consider the various needs of its stakeholders including patients and clinicians of various clinical specialties.

The perceived interoperability levels of the interviewed labs and the described future directions reflect a commitment to improve the interoperability offered by their current information systems. Some labs have well-established plans for improving the interoperability of their lab test reports. Other labs have established general directions but with no specific plans. There is a lag in the implementation of these plans, but exploration of the implementation barriers was beyond the scope of the paper.

This study has several strengths. First, it addresses an important gap in the informatics literature related to the exchange of clinical and genetic data between labs and health-care providers. Second, we included a broad range of labs, spread across the United States, with various business models and specialties. Third, study participants have decades of experience in their fields and were able to articulate detailed descriptions of the information system ecosystem in place at their organizations. Last, we followed a rigorous qualitative methodology including in-depth interviews and rigorous content analysis that resulted in a detailed understanding of information system and data exchange workflow patterns used at participating labs.

This study has limitations, most notably that the results are based on a relatively small sample of labs and hence it is unclear whether the study findings are representative of and capture all substantial variations across US genetic testing labs. To mitigate this limitation, a panel of experts reviewed and discussed the identified models. They felt that the two models, each reflecting the situation of only a single lab, were likely representative of a minority, though these models may be representative of a substantial number of other labs which did not participate in the interview survey. However, the heterogeneity of the workflows did make it difficult to assess whether thematic saturation was reached. In addition, the results reflect only the interviewees' knowledge of their organizations' information systems as well as subjective experiences and points of view, which may not be accurate or complete. One interviewee who had been at the lab a short time invited other members of the lab to participate in the interview to ensure responses were complete and accurate. Yet, the fact that all interviewees had several years of experience working at their organizations as well as their self-reported background and training mitigates this limitation. Future quantitative and qualitative studies are needed to provide a more complete and in-depth understanding of the current state, besides the quantification of the identified sources of variation.

Conclusion

Genetic testing labs develop and implement heterogeneous workflows and generally have a low adoption of standards to send test reports back to health-care providers and systems. The core genetic interpretations are delivered as free text, which limits their computational availability for CDS tools. While genetic testing labs exert great effort to provide their services in the most efficient way to meet their customers' needs, these efforts need to be aligned to achieve the national HIE roadmap's goals. Labs need to provide genetic results and interpretation in a discrete and standards-based format for the benefit of individual health outcomes and improved public health. Understanding the current state provides guidance for the implementation of better informatics solutions that support the practice of genomic medicine and learning health systems as a national goal. Building a reliable genomic medicine HIE infrastructure is paramount to achieving its ultimate aims of better health and a more cost-effective health-care process.

DATA AVAILABILITY

IRB restrictions do not permit the sharing of individual data. However, aggregate data is available within the article and in the supplemental material.

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Conceptualization: A.K., J.H.G., M.S.W., G.D.F., S.B.B., S.M.H. Data curation: A.K. Formal analysis: A.K., C.C.M., J.H.G., M.S.W., G.D.F., B.R.J., S.B.B., S.M.H. Investigation: A.K., C.C.M., J.H.G., M.S.W., G.D.F., B.R.J., S.B.B., S.M.H. Methodology: A.K., J.H.G., M.S.W., G.D.F., S.B.B., S.M.H. Resources: A.K., J.H.G., M.S.W., S.M.H. Supervision: S.M.H. Validation: A.K., C.C.M., J.H.G., M.S.W., G.D.F., B.R.J., S.B.B., S.M.H. Visualization: A.K., J.H.G., S.M.H. Writing—original draft: A.K., S.M.H. Writing—review & editing: A.K., C.C.M., J.H.G., M.S.W., G.D.F., B.R.J., S.B.B., S.M.H.

Compliance with ethical standards

ETHICS DECLARATION

The study was reviewed by the University of Utah Institutional Review Board and deemed exempt in May 2018. For consent, the pre-interview survey included the following statement at the beginning of the survey: "Consent information-by completing this survey you agree and consent to participate in this study and to use the collected information (except contacts and identifiers) for research purposes." For the semistructured interview, the interviewer had declared the following statement before starting the interview: "By agreeing to participate in this study interview, you consent for us to use the provided data for this research. This study is completely voluntary and you may withdraw at any point. Do you consent to participate?" All interviewees had agreed to participate.

COMPETING INTERESTS

B.R.J. receives salary support from ARUP Laboratories, a nonprofit enterprise of the University of Utah. S.B.B. is an employee of Genome Medical Incorporated and he has stock options in the company. C.C.M. was funded by the Pediatric Cancer Program which is supported by the Intermountain Healthcare and Primary Children's Hospital Foundations and the Department of Pediatrics at the University of Utah. The other authors declare no competing interests.

ADDITIONAL INFORMATION

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