Genetic Counseling Using Workflow-based EMRs

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Abstract—Widespread use of genetic tests for medical treatment and clinical genetic counseling–as a cost-effective treatment for an increasing number of hereditary disorders–has led to study of privacy and disclosure issues, and has compelled governments to limit disclosure of test results. To the best of our knowledge, no clinical workflows for genetic counseling apply applicable information disclosure laws have been documented and enforced in Electronic Medical Records (EMRs). To fill this void, herein we model a representative genetic counseling workflow and show how to simultaneously enforce privacy and informed consents in an open-source EMR. Our prototype provides workflow-guided counseling as well as consent management that enforces state and federal law-compliant genetic information sharing.

Index Terms—Genetic Privacy, Privacy Laws, Electronic Medical Records (EMRs), Workflow Management Systems, Ontology

I. INTRODUCTION

As genetics research advances, the list of predictable diseases is growing. For example, having been identified genetic mutations which associated with diseases include breast cancer, ovarian cancer, sickle cell anemia, etc. Studies have shown that preventive care costs significantly less than treatment upon diagnosis of a disease [18], [26]. Therefore, genetic tests, along with family history, are becoming a common practice in identifying risks of many hereditary conditions. clinical genetic services are the complex processes, usually involve genetic tests for finding gene mutations to make eventual disease onset predictable and Genetic counseling for explaining genetic test outcomes and suggest possible courses of action [12] to genetic tests requesters. Genetic test results are not only being used as indication basis for providing preventive and preemptive treatment for hereditary diseases, but also being broadly utilized for research purposes to discover more and more new findings. To compare with other medical researchers, researchers in genetic medicine need to use both genetic test results and their owners identifiable information, so more open accesses are required, e.g. using an opt-out consent that is much less rigorous in format for sharing data.

Genetic tests usually involve finding known changes - referred to as *mutations* - in a gene of a person that causes diseases. Researches have identified genetic mutations associated with various diseases such as breast cancer, ovarian cancer, sickle cell disease, β-thalassemia, left ventricular non-compaction cardiomyopathy (LVNC), and Alzheimer disease and many others. As new research on known gene mutations become available, and medically acceptable as indications, more diseases are added to the list for genetic tests that are available.

Several companies such as 23andMe [1], Gene by Gene [4], Color Genomics [3], and others, offer genetic tests and risk assessment services in the direct consumer market. Additionally, larger laboratories such as Myriad Genetics cater directly to health-care providers [8]. Information from these results are analyzed by professionals in genetic science who then provide counseling services to patients. With increased competition and lower costs of genetic tests [7], genetic counseling is going to play an important role in preventive care. This places genetic counselors in a critical path to explain the outcomes of genetic tests, and suggest possible courses of action [19].

Conducting genetic tests involves addressing ethical and privacy issues. Samples of human blood/tissue, and derived genetic information are able to precisely identify an individual and a group of related people that may be susceptible the same diseases as the original sample donor. Consequently, when genetic sequence information is shared without consent, lost, stolen, or used for a purpose other that which consent was obtained, the identity of a person is compromised. This information can be used by a third party to discriminate or, worse, harm the donor or a group of people. Prince et al. describe three practical genetic counseling cases that illustrate genetic discrimination [20]. Individuals may face discrimination in life, disability, and long-term care insurances. In other cases, when genetic information privacy is compromised, an individual may experience the stigma of having to carry a genetic marker for a disorder or disease.

Although, genetic tests have existed for a while, using genetic information for diagnosis and treatment is a part of a larger process that is being broadly termed as genetic counseling. Consequently, the precise processes (workflows) used by medical practitioners for genetic counseling is not very well defined. A good counter example is the workflow for hemodialysis where the a standard workflow for treatment is used by medical practitioners [24]. Although there is an increase in the bio-medical master degrees awarded by medical schools [14], less than 35 universities offer degrees with specialization in genetic counseling [2]. This results in difference in the process followed during genetic counseling. As a means of articulating different workflows currently emerging in genetic counseling and their larger usage in patient diagnosis and treatment, we have developed a prototype for genetic counseling with a flexible way of specifying and using

these workflows. In our prototype, we implement a workflow based on [17]. However, our tool can be easily modified to accommodate the changes in the workflow at a later stage.

The two US federal laws that regulate sharing of genetic information are Health Insurance Portability and accountability act of 1996 (HIPAA) [10], and Genetic Information Non-discrimination act of 2008 (GINA) [5]. HIPAA considers genetic information to be confidential medical information and regulates health-care providers. GINA regulates employers and health insurance companies but not health-care providers in using genetic information and protects individuals form discrimination based on genetic conditions. However, GINA does not apply to federal government employees or employers with fewer than 15 employees. These complex laws, in addition to the fragmented laws in each state, form the basis for information sharing and consent management workflows of our prototype system. In creating our prototype system, we also noticed significant regulatory gaps that create additional burdens in providing automated workflow-based guidance in genetic counseling.

Challenges. The following are the challenges for implementing a workflow-based EMR for genetic Counseling:

- Genetic Counseling is a new and an emerging field where the workflow has not been standardized, although providing a basis to do so would facilitate this emerging area and the mission of training genetic counselors.
- Genetic information collected for tests and their sharing have to conform to HIPAA [10] and GINA [5] regulations.
- State laws to protect Genetic information vary and add complexity to the system. HIPAA specifies that stricter sharing laws mandated by state regulation can override HIPAA policies.
- Although commercial systems may include Genetic Counseling in their packaged EMRs, it is difficult to verify their workflow as they are closed source.

Contributions. In order to address the above limitations of existing EMR systems, we present an end-to-end managed EMR prototype for genetic counseling that can accommodate the emerging workflows and diversity of state regulations (or lack thereof) in a re-programmable way. To the best of our knowledge, the proposed system is the first of its kind. Our working prototype has the following features:

- Automatically suggests Genetic Counseling for known disease codes
- Enforces a standardized work-flow for Genetic Counseling
- Automates paperless information sharing and medical treatment consent in accordance with local laws

The rest of the paper is organized as follows: Section II describes the closely related work in this area. In section III we describe the implementation of our system. Finally, we present our conclusions in Section IV.

II. RELATED WORK

Electronic Medical records for Genetic Counseling

Electronic Medical Records (EMRs) plays a vital role of book keeping in the health-care industry. However, EMRs for genetic counseling present a unique set of challenges [13] as identified by Belmont et al. A major issue identified by the Belmont study the required uniformity in representing collected genetic data. Additionally, this study highlights the privacy, ethical and legal issues of handling genetic data in EMRs. Ours is a flexible freeware based platform to study these issues.

Scheuner et al. conduct a case study to verify if the current EMR systems meets genetic information needs [21]. This study involved results and conclusions gathered from discussion about 56 patient's electronic medical records with 10 EMR specialists, 16 medical geneticists, and 12 genetic counselors. An overall lack of support for functionality, structure, and tools for clinical decision making was an important finding.

A more recent study of the state of EMRs supporting genomics for personalized medicine again identifies structuring of data as a challenge [22]. The authors also identify clinical workflow management as a priority area that needs further research, development, and testing. Functionality, structure, and support for genetic information specific data is easily added to an EMR system. However, current EMR systems for genetic counseling still lack support for workflow enforcement and the ability to collect specialized genetic information sharing consents and enforce them on EMRs that contain the data.

Genetic Counseling ontology

The Gene Ontology Consortium has developed an ontology to store structured gene information in databases [16]. The ontology provides structured terms and vocabulary to store information regarding gene, gene products and sequences. The structure and terms developed by this team lacks support for capturing the terms used for informed consent requirements laid out by law. In a more closely related work, authors in [26] describe an ontology for treatment consent. The ontology presented is, however, insufficient for capturing the terms and vocabulary used in information disclosure consent. In this work, we develop the ontology containing the structure and terms required for information disclosure consent.

Genetic Counseling informed consent

Obtaining informed consent for diagnosis (including testing) and treatments is a very well studied area and a mandatory requirement on care providers. In particular, care providers are required to obtain informed consent for genetic counseling as much as any other treatment. A preliminary study convened jointly by the National Institute of Health (NIH) and the Center for Disease Control (CDC) [15] presents the risks and ethical issues involved in collecting and storing tissue samples for genetic tests in a research setting. Ethical and legal issues are similarly present in a clinical genetic counseling process.

Authors in [25] present an automated and paperless informed consent management system for medical treatments.

This work provides a generic framework to enforce informed consent for minors. The work presented in [25] only enforces general treatment consent. This work does not specifically address the additional issues related to genetic counseling and sharing of genetic information. Genetic counseling requires enforcement of information disclosure, research and sample and genetic information retention consents in addition to treatment consent. The objective of this work is to create a workflow that enforces all types of mandated informed consent requirements in the genetic counseling process to comply with local, state and federal regulations.

III. SYSTEM

This section presents details of our prototype genetic counseling EMR system within a sample enforced workflow. We first describe genetic counseling workflow and the challenges involved in modeling the process. Then we present a model of this workflow using a work-flow engine. Next, we describe the different types of informed consent for genetic counseling. We then collate the federal and state laws that regulate disclosure of genetic information. Then we describe how we enforce the disclosure consent for genetic counseling to comply with various state and federal laws. Lastly, we describe how we integrate the workflow enforcement with an open-source Medical Record System (OpenMRS).

A. Genetic Counseling Workflow

The Genetic Counseling Definition Task Force defines *Genetic counseling* as *The process of helping people understand and adapt to medical, psychological, and familial implications of genetic contributions to disease*. A team of health care workers involving, but not limited to, Medical Practitioners, Bio-curators, Genetic Counselors, Molecular Pathologists, Medical Geneticists play an important role in Genetic Counseling. Similar to other medical procedures, genetic Counseling follows a well defined protocol. This protocol involves several tasks performed by the caregivers in a particular sequence called the workflow.

Although genetic counseling has been around for a while, we were unable to find a documented and modeled workflow for this process. Therefore, we present a detailed documentation for clinical genetic counseling. We model our workflow based on a presentation about clinical genetic counseling from Stanford's Clinical Genomics program [17]. In addition to the previous presentation, we refer to another presentation by O'Daniel et al. on Genomic Medicine [18] for additional details. Here we provide a detailed documentation and overview of the Genetic Counseling workflow. Figure 1 shows the workflow for genetic counseling modeled using a workflow editor, created using an open-source workflow system YAWL [23].

Our modeling of the workflow is tailored to genetic counseling for Hereditary Cancer Syndromes. However, this can be modified to accommodate any other genetic disorder. The following list walks through the details of the individual tasks involved in the process of providing genetic counseling to a patient. The following list focuses on describing individual tasks involved in the genetic counseling process. We present the details of consent management later in section III-B.

1) **Collection of Medical Records, Family History, and Social History:**

   - *Require Genetic Counseling:* A genetic counseling case originates with a physician's referral for a genetic counseling. Alternatively, in other cases a patient self requests genetic counseling after learning of a manifestation of a genetic disorder in a family member.
   - *Patient Walk-in:* In the case where a patient self requests genetic counseling, upon arriving at the genetic counselors office, the patient is asked to provide social and family history. Additionally, if copies of medical records are provided, they are recorded in the system. In our workflow model, the patient is presented with a detailed family and social history questionnaire to assess the risk of a suspected Hereditary Cancer Syndrome. We model this questionnaire based on forms developed by the Virginia Women's Center [11].
   - *Physician's Referral:* When a physician refers a patient for genetic counseling, the patient is asked to sign release forms for medical records from the referring physician (if not already provided). Else, if the patient brings in copies of medical records, they are recorded in the system.
   - *Requirements Review:* Information collected from the patient is reviewed for completeness. If any, missing information is collected from the patient, patient's primary care physician or public sources of ancestry information. This is shown in the 'Collect More Information' process in the modeled workflow (Fig. 1). Once all of the information (to the best of patients knowledge) is collected, one or more of following people review the collected information: Genetic Counselor, Molecular Pathologist, Medical Geneticist. Next, they verify if a genetic test is available to answer the questions posed by the patient or the treating physician.

2) **Pre-test counseling:** Once an insurance or payment authorization is obtained, the patient meets with a genetic counselor for pre-test counseling. The genetic counselor performs the following sub-tasks for pre-test counseling:

   - An informed consent is obtained for counseling, i.e., Genetic Counseling Treatment Consent. As a part of this informed treatment consent, the counselor provides the patient with information about the risks and benefits of this process.
   - The family, social, and medical history provided by the patient is reviewed.
   - The counselor discusses the expected range of results and their impact on the patient and their relatives. Additionally, the counselor discusses other potential incidental findings that may be in the
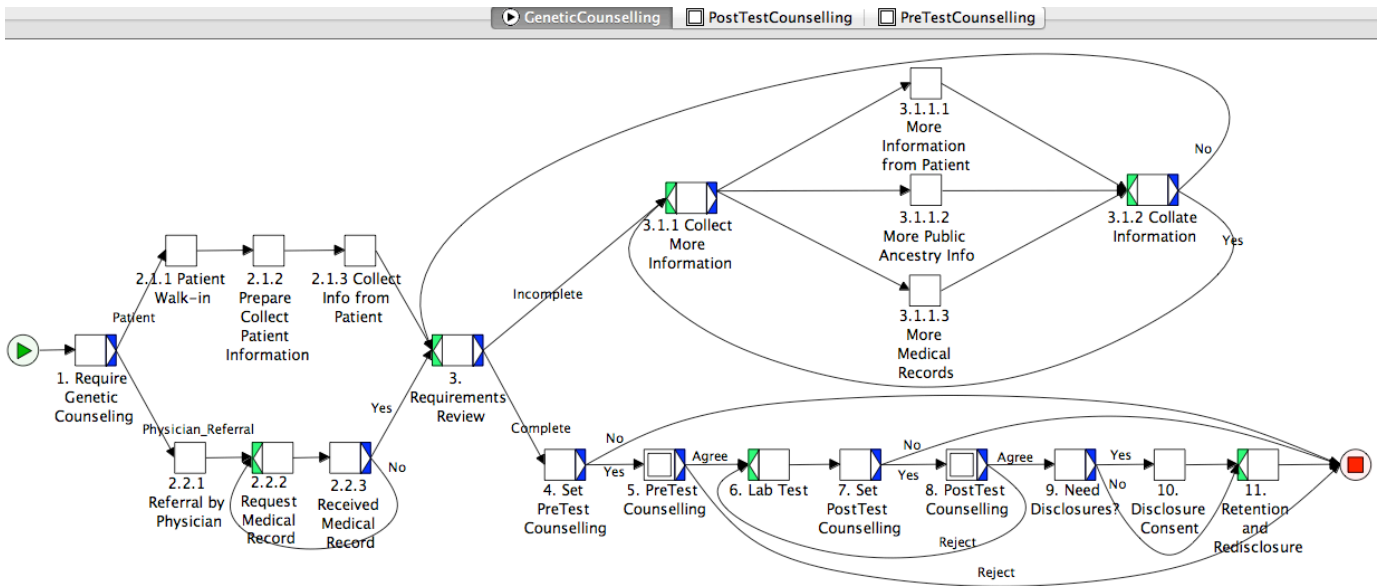
Fig. 1: Genetic Counseling Workflow

report. Incidental findings are those findings in the test results that are not associated with the primary reason for conducting the test, but indicative of other genetic disorders. The counselor also discusses ethical issues that could arise from conducting or disclosing these test results. Before ordering the tests, any questions or clarifications from the patient are answered.

- If applicable, consent for sharing information for research purposes is collected.

3) **Lab work:** Blood or tissue samples are collected from the patient for genome sequencing. Generally, 1 tube of blood is collected for sequencing. An additional tube may be collected for confirmatory studies. Another tube of blood may be collected from the patient with appropriate consent for research purposes. The blood and tissue samples are then sent to a laboratory for sequencing and the sequenced genetic information is returned to the genetic counseling team. We do not model the workflow for genome sequencing in the laboratory in this paper.

4) **Post-test counseling:** Once the sequencing results are reported back to the genetic counseling team, a draft report is prepared. This draft report is reviewed by a review team consisting of Genetic Counselors and/or Medical Geneticists, Physicians, and/or content experts. Once reviewed, the final report is uploaded into the EMR and is ready for the patient. When the patient meets the counselor to discuss the test results, the following sub-tasks are performed:

- The test results are reviewed by the counselor with the patient. Once the review is complete, the counselor assess the comprehension and coping skills of the patient.

- If the additional evaluation or referral for treatment (e.g. prophylactic mastectomy) is warranted, referrals or orders are issued by the genetic counselors. Alternatively, the patient may decline to accept the recommendations of the genetic counselor. If the recommendations are declined, it is recorded in the EMR.
- The counselor finally discusses implications of the findings on related family members. At this point, the genetic counselor may recommend the patient to personally inform related family members about the test results and treatment options by providing templates. Alternatively, they may obtain consent to disclose test results to affected family members.
- Lastly, if required, the counselor may schedule follow up appointments .

5) **Disclosure and other Consent:** At the end of the genetic counseling process, the care provider may be required to obtain consent to retain samples and/or genetic information (sequencing results) and genetic test reports based on state and federal regulation. Additionally, the care provider may require additional disclosure consents to share information with other individuals or entities as required by law. If consent to retain genetic samples and test information is denied, they must be destroyed and/or purged from the EMR records. Once these housekeeping tasks are complete, the genetic counseling workflow comes to an end.

B. Informed Consent in Genetic Counseling

As discussed earlier, if a proper workflow is not enforced for genetic counseling in the EMRs, it may leave health-care providers and/or health-care organizations open to lawsuits. Existing laws limiting genetic information disclosures do not

generally consider ethical issues involved during the disclosure process. Ethical issues present an abstract concept that need to be carefully considered by genetic counselors on a case-by-case basis. Additionally, an organization's ethical guidelines my regulate disclosure of genetic information, which upon disclosure may tangibly affect a third party. Lack of clear federal and state legislation covering ethical issues and individual Organizational ethics boards having different requirements makes it difficult to model them as workflows as of this writing. Therefore, we leave documenting the workflow for ethical disclosures as future work.

One stark difference between other medical treatments such as eye surgery, or hemo-dialysis is that the patient can stop genetic counseling at any stage of the workflow. Patients are permitted to revoke a consent at in point during the counseling process. Once a patient revokes a consent or declines to continue treatment, the care provider is required to document this in the EMR. Workflow in Figure 1 allows patients to revoke or decline treatment at specific check-points in the workflow.

In this part we focus on enforcing informed consent requirements stipulated by state and federal laws. Managing and documenting informed consent during the genetic counseling process is different from other general treatments. A patch work of state and federal laws and regulations to be followed by the care provider in each state adds additional complexity to the workflow of genetic counseling. The following types of informed consent is required for Genetic Counseling:

- **Treatment Consent:** Treatment consent for genetic counseling helps the patient understand the risks, benefits, and limitations of genetic counseling and are modeled in our workflow. Treatment consent regulation varies greatly among states, the details of which are captured in [25]. This consent is different from generic treatment consent as the test results may have the following outcomes: Positive, Negative, and Uncertain. For example, if a patient is being tested for Hereditary Breast Cancer markers BRCA1 or BRCA2, the results may be positive for mutations that increase the risk of breast cancer. The results may be negative for known mutations or it may contain unknown mutations that have not been studied. These test results are classified as medically actionable (e.g. prophylactic mastectomy) or in-actionable (no treatment is available). A separate consent is recommended for finding incidental mutations that may be actionable or in-actionable. Consent is also required for collecting blood and tissue samples from the patient prior to conducting a test. State laws provide for exemptions to requiring treatment consent in certain cases such as Paternity tests.
- **Information Disclosure Consent:** Information disclosure consent is regulated at both state and federal level. HIPAA, GINA, and individual state laws such Delaware Code §16.2.120 - §16.2.1227, protect patients from unauthorized disclosure of genetic information and test results. Laws usually require that the patient is provided with information about the type of information being disclosed

and the name of the entity to which it is disclosed. In certain cases, the reason for disclosure may also be provided. HIPAA, and state laws provide for exemptions from this type of consent (e.g., Identification of bodies). Certain states also require that a consent be obtained for each instance of disclosure called re-disclosure consent. We model these exemptions and re-disclosure consent in our workflow.

- **Research Consent:** In most cases an informed consent is required from the patient to conduct a research. Research consents explains how the genetic information and samples collected would be used in the study and the potential outcomes of the study. They may also specify if the patient will be informed about any medically actionable findings from the research study and the extent of information that will be shared with the participants. Research consents are mostly governed by Internal Review Boards of individual organizations. Most organizations require a research consent for genetic research as a best practice. However, certain states exempt research organizations from research consent were only de-identified information is collected and/or disclosed as a part of the research.
- **Retention Consent:** Retention consent pertains to retaining blood or tissue samples and genetic information or test results once the test is complete and results are shared with the patient. Some states require blood and tissue samples be destroyed at the end of the test. Since blood and tissue samples are collected and sent to the laboratory for processing we do not capture retention consent in this particular case in our workflow. We capture in our workflow, cases where states require a retention consent for storing genetic information or test results. If retention consent is denied, information and test results are purged at the end of the genetic counseling process.
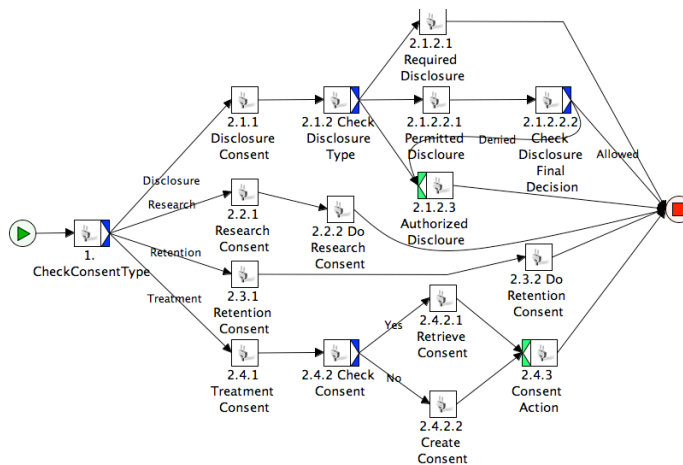

Fig. 2: Consent Workflow

1) Consent Management in the Workflow: In this section, we describe how we enforce the required consents in the genetic counseling workflow. Although we focus on the enforcement of disclosure consent, other types of consent may

be enforced in the workflow very similarly to the disclosure consent.

Figure 2 shows the workflow specification for the different types of consent modeled in YAWL. The consent workflow runs as a separate service in the workflow engine. When a task requires consent in the genetic counseling workflow, the consent workflow is invoked to manage the consent requirements. In the case of information disclosure consent, the workflow checks the purpose for the information request. HIPAA classifies these purposes into different disclosure categories described later in this section. If the disclosure type requires a consent, the consent workflow enforces this requirement. For simplicity, our modeled workflow enforces retention and research consent for all states as it is a best practice. For a detailed working of the treatment consent tasks, refer to [25].

In order for the workflow to enforce all of the above consents for genetic counseling, we use a rule base to codify laws. We do so by first modeling the term structure we use to specify rules and then use OWL Description Logic (DL) to specify these rules using the terms created in our ontology. We use the open source ontology editor Protégé and the Pellet reasoner to create the rules and specify the rules respectively. During the counseling process, the workflow engine YAWL invokes the rule bases's run-time to determine the required consents and generates the required consent forms that are displayed to the patient. Once the patient signs the forms, the workflow proceeds to the next step, thereby enforcing federal and applicable state laws during the counseling process.
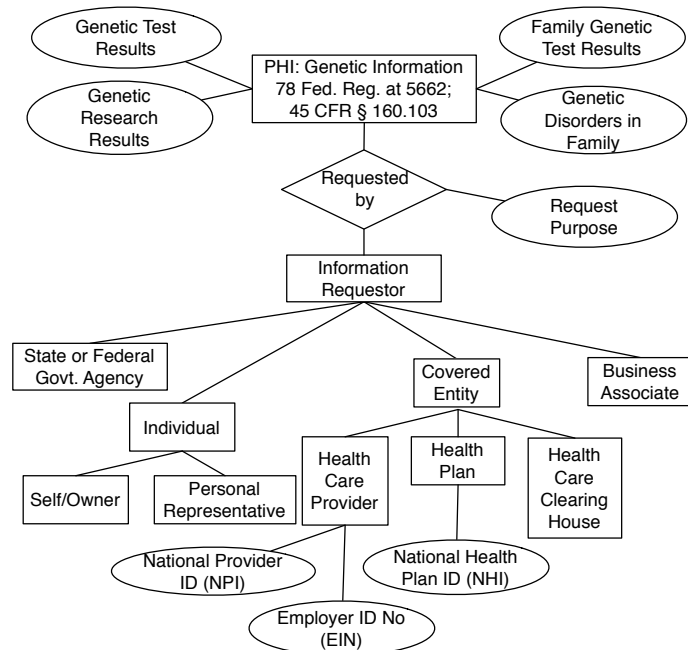

Fig. 3: Entity-Relationship Diagram for Genetic Information

In order to model the terms structure for genetic information disclosure, we start by identifying the relationship between the information requester and Protected Health Information (PHI) as defined by HIPAA. Figure 3 shows the ER diagram

for information disclosure consent in genetic counseling. The figure shows the *requests* relationship between an *information requester* and *Genetic Information*. Genetic Information is considered as Protected Health Information (PHI) according to HIPAA. Definitions pertaining to genetic information are included in detail in this legislation. An individual's genetic information is defined to include the following:

- Genetic test results of the patient
- Genetic research results of the patient
- Genetic test results of a related family member
- Information about manifestation of a genetic disorder in a family member

HIPAA permits or denies information disclosure of PHI and states the informed consent requirements based on the type of information requester and the purpose for which the information is being requested. HIPAA further classifies an information requester as State/Federal Agencies, Individuals, Covered Entities, and Business Associates. These sub types are shown in Figure 3. Based on type of the requester requesting access to genetic information, HIPAA classifies request purpose into the following categories:

- **Required Disclosures:** Required disclosures do not require any consent from the patient and are required to be disclosed. E.g. HHS Compliance Review
- **Permitted Disclosures:** These disclosures are permitted by law without consent. However, an organization may choose to implement policies and procedures requiring additional consent requirements. Examples of permitted disclosures include Treatment, Health care Operations, Public Interest and Beneficial Activities, etc.
- **Authorized Disclosures:** Authorized disclosures require informed consent from the patient, such as for General Research, Marketing, Psychotherapy Notes, etc.

The complete set of Privacy Rules were obtained from HIPAA located at 45 C.F.R. Part 160. These rules are then written into the Pellet reasoner for enforcing information disclosure consent requirements. In addition to the federal information disclosure requirements described in HIPAA. State laws regulate genetic information disclosure. They may have additional or fewer requirements when compared to HIPAA. For example, Delaware has a comprehensive code on regulating disclosure of genetic information and test results. On the other hand, states such as Alabama have no specific legislation.

In 2008, only 27 of the 50 states had specific laws requiring consent to disclose genetic information [6]. As of 2014, this number had risen to 35 [9]. Other states are acting swiftly to protect address the issue of genetic information privacy of its citizens. We gather all pertinent state laws governing genetic information passed until 2014 from [9]. Table I shows genetic information disclosure laws for a sample of selected states. These laws were then transformed into a simple algorithm to permit or deny disclosures of genetic information. Finally, these algorithms were translated into rules to enforce a state laws in the reasoner as described above.

State	Informed Disclosure Consent Law	Required/Permitted Disclosures
Alabama	N/A	N/A
...		
Delaware	- Unlimited access by subject to their own genetic information - Disclosures are to be authorized by obtaining informed consent of the tested individual describing the information to be disclosed and to whom	- Disclosure is necessary for the purposes of a criminal or death investigation or a criminal or juvenile proceeding or to protect the interests of an issuer in the detection or prevention of fraud, material misrepresentation or material nondisclosure - Disclosure is necessary to determine paternity - Disclosure is authorized by order of a court of competent jurisdiction - Disclosure is made pursuant to the DNA analysis and data bank requirements of §4713 of Title 29 - Disclosure is for the purpose of furnishing genetic information relating to a decedent for medical diagnosis of blood relatives of the decedent - Disclosure is for the purpose of identifying bodies - Disclosure is pursuant to newborn screening requirements established by state or federal law - Disclosure is authorized by federal law for the identification of persons - Disclosure is by an insurer to an insurance regulatory authority - Disclosure is authorized in accordance with §1201(4)d. of this title - Disclosure is otherwise permitted by law
...		
Florida	Informed consent is required to disclose genetic test results	Public entities are exempt from disclosure restrictions. Pursuant to Florida Statutes §119.07(1) and Statutes 42(a), Article 1 of the Florida Constitution
...		
Wyoming	N/A	N/A

TABLE I: State Laws limiting Genetic Information Disclosure

Flowchart 4 shows how genetic information disclosures are allowed or denied in a based on organizational policies, State, and Federal laws. When genetic information or test results are requested, information about the requester, Request purpose, and genetic information are collected in the EMR system. These are then passed to the modeled workflow, which uses the reasoner to assess whether a consent is required. The workflow then triggers the EMR system to display the appropriate consent forms. When both state and federal laws exist, HIPAA resolves eventual contentions by providing precedence to the law that does not require a consent for information disclosure. This precedence rule is implemented in the reasoner. The procedures modeled in flowchart 4 allow for individual organizational policies in the information disclosure process. However, our implementation does not model any organizational information disclosure policies.

2) Workflow enforced EMR: In the last step, we modify the source code of OpenMRS, an open source Electronic Medical Record System, to provide specialized interface for genetic counseling. We add JSP scripts to OpenMRS to generate treatment, information disclosure, and research consent. The EMR communicates with the YAWL run-time which in-turn communicates with the reasoner to complete our prototype system. Our system is able to enforce the genetic counseling workfow and consent requirements entailed in these workflows in accordance with state and federal laws.

IV. CONCLUSIONS

In order to model the genetic counseling workflow, we present and document the details of the sequence of tasks performed by the care providers during genetic counseling process. We also study and extract rules from federal and state laws that limit the disclosure of genetic information. We use these rules to implement a workflow-enforced Electronic Medical Record System tailored for working with Genetic Counseling.

To the best of our knowledge, our work presents a first working open-source prototype EMR for genetic counseling. Our genetic counseling EMR supports automatic paperless enforcement of treatment consent, information disclosure consent, research consent, and retention consent. This workflow enforced genetic counseling EMR would enable genetic counseling services provided by care providers and health care organizations to comply with state and federal laws concerning genetic information privacy. As a result, our EMR saves care providers and health care organizations from unnecessary litigation that would arise when proper procedures are not followed. Additionally, the electronic audit trail left by our EMR would help care providers and health care organizations during eventual litigations.

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Fig. 4: Flowchart for allowing or denying Genetic Information Disclosures

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