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# The genome-enabled electronic medical record

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## Abstract

The integration of patient-specific genomic information into the electronic medical record (EMR) will create many opportunities to improve patient care. Key to the successful incorporation of genomic information into the EMR will be the development of laboratory information systems capable of appropriately formatting molecular diagnostic and cytogenetic findings in the EMR. Due to the lack of granular genomics-related content in existing medical vocabularies, the adoption of new standards for describing clinically significant genomic information will be an important step toward recognizing the genome-enabled EMR. Appropriate capture of patient-specific genomic results in the EMR will generate new opportunities to utilize this information in clinical decision support, including automated response to pharmacogenomic-based risks.

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Rapid advances in our understanding of the human genome have raised expectations that the delivery of patient care soon will be improved by these developments. Resources such as OMIM and PharmGKB [1,2] are useful tools for retrieving deep information about the clinical and biological issues involved in a given clinical condition but are not structured to support the operational delivery of patient care. Despite promising pilot studies [3], the integration of clinical and genomic information to support the delivery of patient care is not yet routine. In their current state, electronic medical records (EMRs) and clinical information systems are not suited to the optimal management of patient-specific genomic information. Most EMR systems were designed to optimize access to diagnostic results of transient value, such as blood chemistry results. Genomic results have lifelong value and (other than those for somatic malignancies) can affect decision making for family members. Likewise, the unique biological knowledge associated with genomic findings is not currently reflected in most existing EMR solutions. Three developments are necessary for the realization of the

“Genome-enabled electronic medical record” in patient care: improved tools to support the capture of genomic results as generated by molecular diagnostic and cytogenetic methods, a controlled vocabulary appropriate for the description of clinically significant genomic findings, and applications capable of enabling clinicians to utilize these results to support their decision making. Together these advances will create the foundation upon which genomics-based capabilities can be built.

Currently, most molecular diagnostics and cytogenetics laboratories use one of three primary methods to manage results. One of the most common methods is the use of anatomic pathology reporting systems. These systems manage text-based reports and are not designed to support the discrete information generated by a genetics laboratory. The second method is the development of a highly customized system built using off-the-shelf software components, often with in-house software development personnel or consultants. These systems are often difficult to support and are not readily extensible. The third method is the use of software developed by niche-specific companies. Such applications typically were not designed to integrate into the broader clinical systems used by many health care

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## Evolution of Reporting

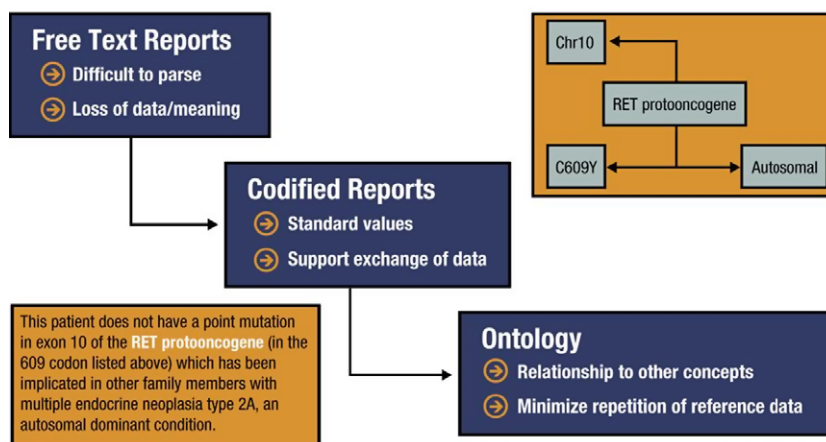


Fig. 1. Phases in the development of structured reporting.

organizations. The current off-the-shelf and niche approaches often do not fully support compliance with the Healthcare Insurance Portability and Accountability Act (HIPAA). For example, they frequently lack the ability to log transactions and queries specific to a patient record.

The current state of reporting for molecular diagnostic results is summarized in Fig. 1, in which reporting of molecular diagnostic findings is either managed as purely textual data or, at best, codified using the limited standards currently available. There is a need for solutions that satisfy the unique workflow requirements of the molecular or cytogenetics laboratory and capture information in a method that retains the discrete details of molecular findings while also supporting the capability of generating a clear report to the ordering clinician. Among the unique workflow requirements of the molecular diagnostics laboratory is the need to document significant amounts of non-clinical information generated during the processing of a sample, for example, during DNA isolation or PCR analysis. An additional requirement is the ability to offer significant flexibility in ordering and canceling tests and procedures in response to poor quality specimens or other procedural failures. These systems should also be able to integrate seamlessly with other laboratory sections, for example, microbiology or anatomic pathology, in order to support the interdisciplinary nature of clinical genomics. For instance, the management of a chronic myelogenous leukemia (CML) case in the hematopathology section requires the smooth integration of clinical pathology (complete blood count) and molecular diagnostic results (BCR-ABL gene expression levels). At least one supplier of integrated clinical information systems has released a laboratory information system module designed to integrate clinical genomics findings into the EMR while also providing the capabilities needed to support the workflow of the molecular diagnostics laboratory.<sup>1</sup>

Codification of molecular diagnostic or cytogenetic results using existing medical vocabularies will prove difficult as these lack sufficient terms for the description of molecular findings. For example, the Systematic Nomenclature of Medicine (SNOMED<sup>®</sup>) has minimal codes related to the description of molecular diagnostic findings. The Logical Observation Identifiers Names and Codes (LOINC<sup>®</sup>) vocabulary has recently added a significant number of molecular pathology terms, but it lacks the rich context-defining relationships provided by an ontology. The Clinical Bioinformatics Ontology<sup>™</sup> (CBO) addresses this gap by providing uniquely identified concepts related to clinically significant molecular findings [4,5]. The CBO is a curated ontology that was developed as a semantic network providing relationships that provide greater context to a codified term. For example, Fig. 1 shows the association between the gene concept for the RET protooncogene and relationships to context-defining concepts representing chromosome 10, the autosomal mode of inheritance and an amino acid sequence substitution in which the cysteine at position 609 is substituted for tyrosine. Currently, the CBO consists of nearly 7000 concepts, each of which is associated with a global unique identifier, offering the benefits of a controlled vocabulary. These concepts are associated by more than 15,500 relationships. The scope of the CBO is currently limited to clinical genetics, molecular pathology, molecular detection, and classification of infectious diseases and cytogenetics.

The combination of new laboratory information system capabilities and the ability to standardize clinical genetics results to a controlled vocabulary create the capability of developing novel decision support applications that can be widely deployed. For instance, codified result values describing HIV mutations associated with antiretroviral drug resistance can be used as the basis for building decision support logic that is embedded in computerized provider order entry (CPOE) systems. This allows the system to intercept medication orders that are contraindicated by

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genotype results. Extending this concept to other pharmacogenomic associations, such as those between the cytochrome p450 genes and many medications, is a logical development and will support the movement toward “personalized medicine.”

Inclusion of structured family history information (generally lacking in current implementations of clinical information systems) will further extend the possible scope of genome-enabled decision support. Enhancements to structured clinical documentation tools to better support the standardized description of family history information will be a key step toward this goal and will better position systems for the support of family practice. Among the extensions required are improved integration with both existing and emerging standard vocabularies and integration with pedigree visualization tools.

Enabling decision support applications to guide providers in determining when a genetic test is warranted will also be a key benefit of the “Genome-Enabled Electronic Medical Record.” Enabling the system to evaluate the discrete information stored in a longitudinal medical record will make the automated recognition of patterns that warrant further genetic analysis feasible. Finally, genomics is an area in which the ability to generate new interpretations of historic results is particularly important as our understanding of the clinical significance of single nucleotide polymorphisms (SNPs) grows. Storage of these results in a discrete, standardized, format that supports periodic automated re-analysis will be one of the key benefits of embedding genomic information in the EMR.

The complexity and dynamic nature of genomic information will require clinical information systems to provide physicians and other care providers with easy access to reference materials, whether web sites such as Genetests [6] or abstracts of primary literature such as those provided by PubMed. Providing access to those resources most appropriate for the case under consideration by the clinician will be a key requirement in order to filter the often overwhelming variety of resources.

The operational capture of genomic information during the delivery of patient care will create new opportunities for enhancing the discovery process. Subject to IRB approval and HIPAA compliance requirements, blinded data warehouses combining traditional clinical and genomic information results can provide a resource for deter-

mining how to prioritize basic research activities, strengthening the currently weak “bedside to bench” component of translational research, which has been more focused on “bench to bedside” efforts. Early large-scale efforts to integrate genomic and clinical information, such as the work of deCode Genetics in Iceland [7], have relied on dual entry of clinical information into one system used to support the delivery of care and another used for research. This duplication of effort can be reduced as standardization between the systems used in the delivery of patient care and those used for research purposes is improved.

Together these developments—the integration of genomics into the laboratory information system, the standardization of molecular findings, and the deployment of clinical applications that simplify the use of genomic information—will create the foundation for improvements in the electronic medical record. These new capabilities must be implemented in software environments fully capable of supporting and enforcing institutional and statutory privacy protections. The successful development of these enabling technologies will facilitate improved patient care by enabling physicians to utilize advanced diagnostic and prognostic capabilities in the delivery of patient care.

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