Clinical Practice Guidelines
Formalization for Personalized Medicine

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ABSTRACT

Clinical guidelines are important means to improve quality of health care while limiting cost and supporting the medical staff. They are written as free text with tables and figures. Transforming them into a formal, computer-processable representation is a difficult task requiring both computer scientist skills and medical knowledge. In this paper the authors describe a CDSS designed to assist physicians for personalized care, and methodology for integration in the clinical workflow. A reasoning method for interacting heterogeneous knowledge and data is a necessity in the context of personalized medicine to achieve its potential and improve the quality, safety and efficiency of healthcare.

Keywords: Artificial Intelligence, Clinical Guidelines, Decision Support System, Personalized Medicine, Quality of Health, Semantic Web

1. INTRODUCTION

Personalized medicine may be considered as an extension of traditional approaches to understanding and treating diseases, but with greater precision (Hamburg & Collins, 2010). A profile of a patient’s genetic variation can guide the selection of drugs or treatment protocols that minimize harmful side effects or ensure a more successful outcome (Landau, Bollag, & Kraft, 2012).

Genetic profiles can better discern different subgroups of diseases, guiding physicians to select the best treatment protocol. Technological developments allow supporting personalized medicine, including computerization of patient data and bioinformatics. The explosion of data and medical knowledge is a major problem for reuse and integration of knowledge in clinical decision support system to provide personalized assistance to practitioners.

Potentially, electronic health records (EHR) will have important roles in the delivery of health care, particularly to improve quality and effectiveness. Among the most important features offered by EHR are electronic exchange and

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interoperability of patient health information; new tools for carrying out some care delivery functions, like drug prescribing; supporting physicians in the delivery of evidence-based care, including incorporation of evolving practice guidelines into point-of-care accessible information formats (Downing, Boyle, Brinner, & Osheroff, n.d.).

Many adverse drug reactions (ADRs) are the result of variations in genes coding for the cytochrome P450 (CYP450) family of enzymes and other metabolizing enzymes (Phillips et al., 2001; Blue Cross Blue Shield Technology Evaluation Center, 2004). These variants may cause a drug to be metabolized more quickly or slowly than in the general population. As a result, some individuals may have trouble eliminating a drug from their bodies, leading in essence to an overdose as it accumulates, while others eliminate the drug before it has a chance to act.

While improvements in quality of care involve all of these functions, it is particularly in the area of clinical decision support system (CDSS) what EHR-based technology is expected to improve delivery of care in a continuously evolving manner. Adaptation of CDSS for personalized medicine is not only to improve semantic interoperability, data access but also to develop reasoning methods that manage heterogeneous knowledge, including clinical, genetic, biological knowledge (Douali, De Roo, & Jaulent, 2012).

This paper describes the new methodology allow CDSS to support Genomic and Personalized Medicine. The development is based semantic web tools.

2. BACKGROUND

2.1. Genomic and Personalized Medicine Challenges

Despite the great promise of the clinical use of genomics, many obstacles lie in the way of integrating genomic and personalized medicine into routine clinical care. These challenges include the need for greater oversight and quality assurance of genetic testing (Phillips et al., 2001). There are limited availability of rigorous, prospectively collected evidence on the clinical value and cost effectiveness of genetic and genomic assays (Khoury, Gwinn, Yoon, Dowling, Moore, & Bradley, 2007; Khoury et al., 2007). That concern among pharmaceutical companies that the tailoring of pharmaceuticals to genetic sub-populations may segment the marketplace and reduce the number of patients for whom a given medication is indicated and prescribed (Ginsburg, Konstance, Allsbrook, & Schulman, 2005). More over the limited knowledge of, and comfort with, genetics and genomics among health-care professionals (Kawamoto, Lobach, Willard, & Ginsburg, n.d.).

2.2. Clinical Decision Support System

CDSS form a significant part of the field of medical knowledge management technologies through their capacity to support the clinical process and use of knowledge, from diagnosis and investigation through treatment and long-term care. Their role and acceptance in daily clinical practice is increasing (Berner, 2007). A several studies (de Clercq, Blomb, Korstenb, & Hasmana, 2004) showed that CDSS can improve physician performance and accuracy, while the quality of each system may depend on the technical approach they use to model medical information.

Despite the potential for genomics to transform health-care, past experience indicates that new genomic interventions, like any new medical intervention, will remain substantially underutilized for many years unless a robust infrastructure is established for supporting their appropriate use. Clinical research results require an average of 20 years to be routinely implemented in clinical practice (Peleg & Tu, 2006). Moreover, genomic interventions may face even greater barriers to clinical adoption compared to more traditional medical interventions, due to such factors as limited clinician familiarity with genomics and the volume and complexity of the underlying data that may need to be considered.
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