Towards Interoperable and Extendable Clinical Pedigrees in Healthcare Information Systems

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INTRODUCTION

Since ancient times, it has been evident that several diseases are more prevalent in some families. It is now know that genetic traits are passed from generation to generation and that many common diseases and clinical conditions are linked to genetic factors. This allows some degree of predictability when dealing with such diseases, provided that clinical family history data is available. Recording patients' clinical family histories is therefore recognized as an important step in the diagnosis and risk assessment of many diseases. In some cases, clinical family histories also allow the identification of at-risk individuals before diseases manifest, potentially minimizing or completely avoiding diseases or symptoms. Though there are several ways to record clinical family histories, such as checklists, forms, descriptive test, etc., clinical pedigrees are a particularly well-accepted tool, standing out for their balance of expressivity and ease of use. These graphical representations illustrate both family structure and clinical conditions of family members, allowing important information to be quickly assessed by observers, such as disease heredity patterns, penetrability, mortality and at-risk individuals.

Given the usefulness of clinical family histories, one would expect this tool to be widely used by practitioners, yet recent studies attest otherwise. Existing literature suggests that one of the main reasons for this underutilization is the fact that most Healthcare Information Systems (HIS) do not yet include adequate tools to record this information. Though clinical pedigrees can easily be drawn by hand or in external drawing software packages – and indeed some practitioners do resort to these alternatives – this results in representations that are disconnected from current health records and consequently difficult to integrate with existing HIS and keep up-to-date.

Some proprietary solutions do exist that allow the creation of clinical pedigrees within certain HIS, but their closed-source nature makes the integration with other HIS a difficult and potentially expensive task.

This chapter presents an alternative solution: the usage of open, interoperable and extendable clinical pedigree information systems that can be easily and freely integrated with existing HIS.

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BACKGROUND

It has long been observed that a number of diseases and clinical conditions are more prevalent in some families than in others. In colloquial terms, it is said that such conditions "run in families". It is now known that genetic are being passed from generation to generation. In fact, developments in Genetics have unveiled links between genetic factors and hundreds of common diseases, such as diabetes, Alzheimer, deafness, schizophrenia and many others (Kmiecik & Sanders, 2009; Rich et al., 2004). It is likely that efforts in human DNA sequencing and gene mutation identification, collection and interpretation, such as those carried out by Human Variome Project (2014) and Human Genome Project (U.S. Department of Energy Human Genome Project, 2014), will expose more and more links between genetic factors and medical conditions.

The significance of combining family history information with patients' clinical data has also been recognized for a long time, before specific links between genetics and diseased were even established. Based solely on the observation of increased occurrence of certain medical conditions within families, Hippocrates (460 B.C. – 370 B.C.) reportedly included family history information in "case studies", complementing the clinical evaluation of disease manifestations and providing an early form of risk stratification, (Hinton, 2008).

Naturally, a better understanding of the genetic nature of some conditions has increased the importance of family history information in modern healthcare. On the one hand, this information provides additional insight to patients' clinical conditions, which may ease prognostics and promote optimal choice of treatment (Morales, Cowan, Dagua, & Hershberger, 2008; Rich et al., 2004). On the other hand, family history information allows the identification of at-risk individuals within the patient's family, when genetic links exist, serving as a cost-effective risk assessment tool (R.L. Bennett, 2010). This discoverability of at-risk individuals fosters the application of predictive medicine, namely monitoring, counseling, genetic testing, suggestion of behavior changes or a combination of these, which can delay, diminish or completely avoid diseases or their symptoms (Frezzo, Rubinstein, Dunham, & Ormond, 2003).

CLINICAL FAMILY HISTORY REPRESENTATIONS

Clinical family history information can be recorded in several formats with varying degrees of detail. Text formats, such as forms, checklists or free text are common (American College of Obstetricians and Gynecologists, 2011) and relatively easy to register but lack the immediate expressivity that visual representations may provide. Free text in particular lacks structure, and important clues about conditions or family structure may be overlooked. Forms and checklists may also be limiting in the presence of uncommon symptoms or family structures. Graphical formats are also used to record family history information, such as genograms, ecomaps and pedigrees. Genograms and ecomaps include not only family structure and clinical conditions but also social relations with external individuals, as well and data regarding emotional symptoms, educational achievements, occupational history, ethnicity, religion, race, migration, class, and sexual orientation (Butler, 2008; Rempel, Neufeld, & Kushner, 2007). Not surprisingly, these tools are commonly used by therapists and clinical practice with families but, as Bennet (2010) puts it, they are not as multifunctional as pedigrees, particularly for disease risk assessment, since non-clinical information frequently clutters the representation, making relevant health information difficult to discern.