Integrating Genetic Information Resources with an EHR

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As the knowledge about the genetic factors associated with clinical conditions increases, access to information that can help practicing clinicians better understand these factors becomes essential for optimal care and communication with patients. We describe the implementation of "infobuttons" from the problem list module of an electronic health record (EHR) to on-line genetic resources.

INTRODUCTION
The integration between the electronic health record (EHR) and on-line information resources, using tools such as “infobuttons,” is considered a promising solution to fulfill clinicians' information needs at the point-of-care [1]. We describe the implementation of “infobuttons” in HELP2, Intermountain’s web-based EHR, to two genetic information reference resources: the National Library of Medicine's Genetics Home Reference (GHR) and GeneTests.

IMPLEMENTATION
HELP2 infobutton requests are handled by an application known as "E-resources Manager" (ERM). Infobuttons can be easily configured using the ERM, requiring only the creation of a new resource profile. A profile defines the relevant clinical context of the resource and provides instructions on how to generate properly formatted and encoded requests.

In November of 2005, we initiated a project to implement infobutton links from the HELP2 problem list to genetic resources. The configuration process included two steps: 1) establishing links between the coded problems used in the problem list and the coded topics used in GeneTests and GHR; and 2) development of ERM profiles for GHR and GeneTests.

The implementation was based on a list of 50 clinically relevant genetic conditions available in either GeneTests or GHR, such as “Cystic Fibrosis” and “Marfan Syndrome.” This list of conditions was created by a co-author who is a clinical geneticist (MSW). The clinical problems and the genetic conditions were linked using a new semantic relationship, labeled as "has_reference." These links were created by the terminology engineer (NM) responsible for the domain of problem concepts.

A minimalistic approach was taken as an attempt to restrict the number of links derived from a single problem concept. The intent was not to confuse the clinician with too many possible infobutton links. Whenever possible, a problem was linked only to the genetic topic that had the largest amount of relevant information. For example, in many cases clinical problems could be associated with high-level topics applicable to multiple genetic conditions (e.g., “Spinocerebellar Ataxia”), as well as topics applicable to subtypes of these same conditions (e.g., Spinocerebellar Ataxia 1, 2, 3, and 6). In these cases, we chose to link problems only to high-level topics, despite the fact that the subtopics could also have provided useful information in specific scenarios.

After the analysis of all 50 genetic conditions, different types of links were created. “Equivalent” links were used when topics corresponded to only one problem (e.g., "Congenital Hypothyroidism"). “Broad” links were used when topics had a broader meaning than the corresponding problem concepts (e.g., "Dystrophinopathies" topic to "Duchenne Muscular Dystrophy" and "Becker Muscular Dystrophy" problems). “Narrow” links were used when topics had a narrower meaning than the corresponding problem (e.g., "Neurofibromatosis 1" and "Neurofibromatosis 2" topics and the problem "Neurofibromatosis"). In a few cases, the appropriate semantic correspondence was not possible, leading to the creation of new problem concepts (e.g., "Aceruloplasminemia").

The new infobuttons to genetic topics were made available to HELP2 users in January of 2006. Before resuming the concept mapping process to include the remaining genetic topics available in GHR and GeneTests, we will evaluate if these new infobuttons are being effectively used by our clinicians.

REFERENCES