Integrating genetic information resources with an EHR

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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.

This article describes the implementation of "infobutton" links from a problem list of a web-based EHR to two on-line genetic information resources: Genetics Home Reference (GHR) and GeneTests.

MATERIALS & METHODS

GHR and GeneTests:

- Topics are associated with unique identifier
- Search API allows content to be retrieved based on topic identifier or search keyword.
- Our approach: "translate" problem list concept codes to GHR and GeneTests topic identifiers rather than keywords.

Configuration of Infobutton links consisted of two steps:

- Establish semantic links between the coded problems used in the problem list and the equivalent topics used in GHR and GeneTests.
- Configuration of profiles for GHR and GeneTests in an Infobutton Manager component.

First phase: focus on a list of 50 important genetic conditions available in either GeneTests or GHR (e.g., "Cystic Fibrosis," "Marfan Syndrome").

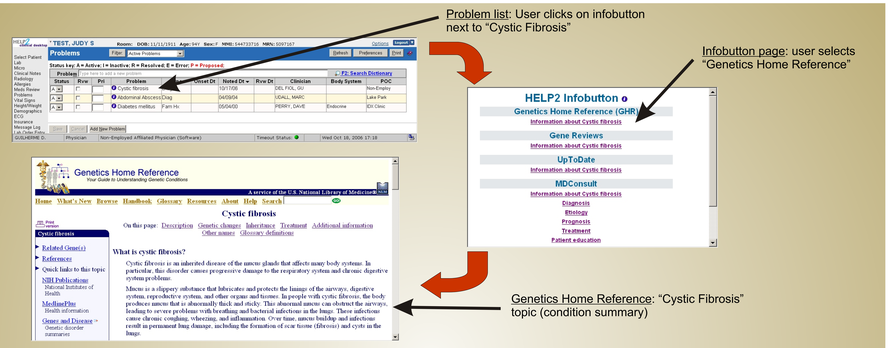

Figure 1 – Sequence of screen shots depicting a problem list infobutton linking to the "Cystic Fibrosis" condition summary in Genetics Home Reference.

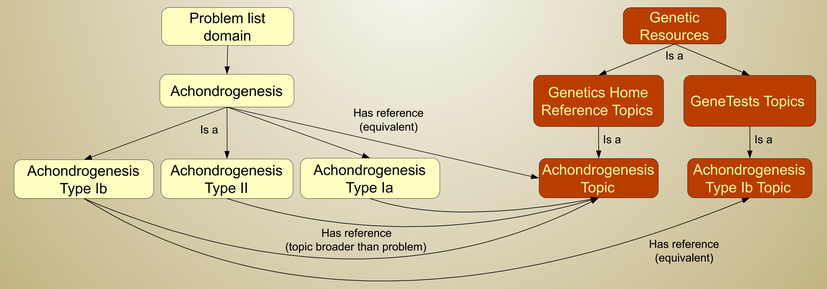

Figure 2 – Example of "equivalent" and "broader than" semantic links.

RESULTS

Four different cases of semantic links were found:

- Equivalent: topic corresponded to only one problem (e.g., "Congenital Hypothyroidism").
- Topic broader than problem: e.g., "Dystrophinopathies" topic to "Duchenne Muscular Dystrophy" and "Becker Muscular Dystrophy" problems.
- Topic narrower than problem: e.g., "Neurofibromatosis 1" and "Neurofibromatosis 2" topics and the problem "Neurofibromatosis." Links were not created in these cases.
- No match: topic had no corresponding problem concept –> solution was to add equivalent problem list concept to problem list domain matching the topic condition (e.g., "Aceruloplasminemia").

DISCUSSION

Advantages of the tested method (as opposed to simple keyword search):

- Precise retrieval, users don't have to select from a search result set
- GHR and GeneTests are offered to users in the infobutton page only when the condition is covered by at least one of these resources.

Disadvantage:

- Requires labor intensive vocabulary mapping. Nevertheless, semantic links between problems and GHR / GeneTests topics could be shared among institutions.

Potential solutions for "topic narrower than problem" cases:

- Create more specific problem concept that is equivalent to topic
- GHR / GeneTests to create an overview document that covers all condition subtypes.

CONCLUSION

The new infobutton links to genetic resources were made available to HELP2 users in January of 2006.

More recently, new links were created for 14 out of 31 conditions that are part of the new Utah newborn screening program.

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