Qualitative evaluation of three phenotype information models to find methotrexate liver injury

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Introduction The fundamental basis for EHR driven phenotyping is to represent a query or algorithm, such that it may be executed against a repository of clinical data. Hence, the overarching goal of this project is to formally evaluate existing phenotype algorithm representation models, identify gaps, and where applicable, propose and implement extensions. To achieve this objective, we evaluated the National Quality Forum (NQF) Quality Data Model (QDM)[1] and HL7 Health Quality Measure Format (HQMF)[2] for the Measure Authoring Tool (MAT)[3], star schema and ontology for Informatics for Integrating Biology and the Bedside (i2b2)[4], and temporal abstraction ontology (TAO)[5] for Eureka! Clinical Analytics (Eureka)[6].

Materials and Methods

Phenotype algorithm: The "Methotrexate (MTX) drug-induced liver injury" phenotype algorithm shown in Figure 1 was selected for this evaluation task because it was a relatively simple algorithm that includes the complexity common to many more complicated phenotypes, such as, 1) multiple criteria (inclusion and exclusion), 2) multiple data types (diagnosis, medication, laboratory, etc.), and 3) temporal relationships.

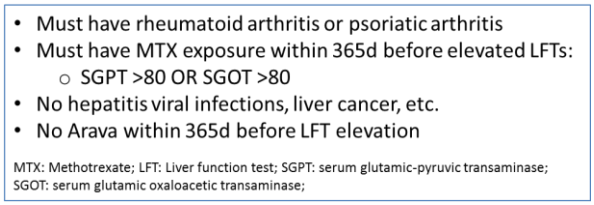

Figure 1. MTX drug-induced liver injury algorithm

Information model comparison: Three well-used phenotype authoring tools (MAT, i2b2 and Eureka) were explored in this study to identify gaps among the information models behind them by representing the MTX algorithm in each. QDM (used by MAT) is an information model describing clinical concepts in a standardized format. The star schema (used by i2b2) is designed to capture arbitrary data types and allow for rapidly developed analysis queries by incorporating with the i2b2 ontology (also called metadata) that is integral for querying the data. TAO (used by Eureka) provides a model for specifying phenotypes in terms of categories of codes, thresholds in value and slope, and both frequencies and sequences of both events and observations. During the implementation process, a list of major features for capturing semantics of phenotype algorithm has been identified and compared these three information models. The feature matrix for comparison is shown in Table 1.

Discussion In our evaluation, we have identified several strengths and limitations for the information models using in the three authoring tools. All of them have the capability to represent phenotyping algorithms for both machine and human consumption, and have their own advantages compared to the other two. For MAT, representation of terminology value sets within the QDM as code lists external to the measure definition, which are assigned a globally unique identifier, allows for their re-use. In addition, QDM requires strict definitions of value sets – code sets, code set version, a code and description for each entry. For i2b2, the query can be easily transformed and implemented against the backend clinical relational database. For Eureka, it includes well-defined temporal pattern types that cover a majority of temporal events occurring in phenotype algorithms. Phenotypes are composed from discrete building blocks (e.g., rheumatoid arthritis diagnosis codes, LFT value thresholds) that may be reused in other phenotypes. Additionally, we identified several limitations, including lack of support for specifying natural language processing (NLP) constructs and significant challenges in the ability to transform HQMF elements and components into executable queries (e.g., into SQL). These will be extensions proposed for future studies.

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Table 1. Feature Matrix of information models comparison

	QDM + HQMF (MAT)	Star Schema + Ontology (i2b2)	TAO (Eureka)
Data type (Diagnosis, Medication, Lab)	Yes	Yes	Yes
Comparison operators (equal to, greater than, less than, greater than or equal to, less than or equal to)	Yes	Yes	Yes
Logic operators (And, Or, Not)	Yes	Yes	Yes (*And* and *Or* supported in phenotype editor, *Not* supported by querying phenotypes in i2b2)
Arithmetic operators (Addition, Subtraction, Multiplication, Division, Modulo Reduction)	No	Yes	No
Aggregate Functions (MIN , MAX , SUM , AVG , COUNT)	Yes	No	No
Counting rules (e.g. multiple diagnosis codes or criteria to specify one phenotype)	Yes, but without clearly defined rules	Yes, but not for multiple criteria	Yes
Temporal Constraint	Yes	Yes (Temporal sequence of events)	Yes (Four types of temporal relations included, Category, Sequence, Frequency, Value threshold)
Complex Projection	No	No	No
Standardized terminologies	Yes Integrating VSAC	Yes (Standard terms being used)	Incomplete (Local terms being used)
Compatible to other formats	Yes (Translator being used from HQMF to i2b2[7])	Yes (Translator being used from HQMF to i2b2[7])	Yes
Logic sharing (whether partial components or entire algorithm) between algorithms	Yes	Yes	Yes
NLP support (ad-hoc or real-time)	No	No	No
Translatable into SQL	Yes (translated from HQMF -> i2b2 -> SQL	Yes	Yes

Reference

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