



RECOMMENDATIONS FOR ELECTRONIC PHENOTYPING

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DISCLOSURES

- In the past year, I have been a consultant or sponsored researcher on clinical decision support for ONC, Hitachi, and McKesson InterQual

BACKGROUND

- Associate CMIO
 - Clinical decision support, quality measurement
 - Interoperable Applications and Services
- Standards development and implementation
 - Co-Chair, HL7 CDS Work Group
 - Founder, OpenCDS (www.opencds.org)
 - Coordinator, ONC HeD and CQF initiatives
 - Member, Health IT Advisory Committee

QUESTION 1

- How can eMERGE improve upon the current labor-intensive phenotyping toward fully-automated phenotyping methods to increase phenotyping efficiency and validity using EMRs?

RECOMMENDATIONS (BIG PICTURE)

- Learn from, and synergize with, related efforts
 - Electronic phenotyping is a common problem encountered in other areas, and beyond genomics
 - There are many related efforts in data standardization, clinical decision support (CDS), & electronic clinical quality measurement (eCQM)
 - Synergies = more resources, more adoption

USE HL7 CLINICAL QUALITY LANGUAGE (CQL)

- Developed by ONC/CMS CQF initiative
- CMS has announced it will start using CQL for eCQMs
- Consequently, EHR vendor support is likely to start emerging

CQL EXAMPLE FOR ECQM (DIABETES FOOT EXAM)

```
using QUICK
valueset "Diabetes": '2.16.840.1.113883.3.464.1003.103.12.1001'
valueset "Sensory Exam of Foot": '2.16.840.1.113883.3.464.1003.103.12.1014'
parameter MeasurementPeriod default interval[Date(2013, 1, 1), Date(2014, 1, 1))
context Patient

define IsAdult:
  AgeAt (start of MeasurementPeriod) >= 18 and AgeAt (start of MeasurementPeriod) < 75

define HasDiabetes: exists
  ([Condition: "Diabetes"] C where C.effectiveTime overlaps MeasurementPeriod)

define InDenominator: IsAdult and HasDiabetes

define InNumerator: exists
  ([Procedure: "Sensory Exam of Foot"] P where P.performanceTime during
  MeasurementPeriod )
```

BUILD ON FHIR US CORE PROFILE

- EHR vendors are focusing on these profiles (<http://www.hl7.org/fhir/us/core/>)
- While far from perfect, offers a baseline of fairly wide support

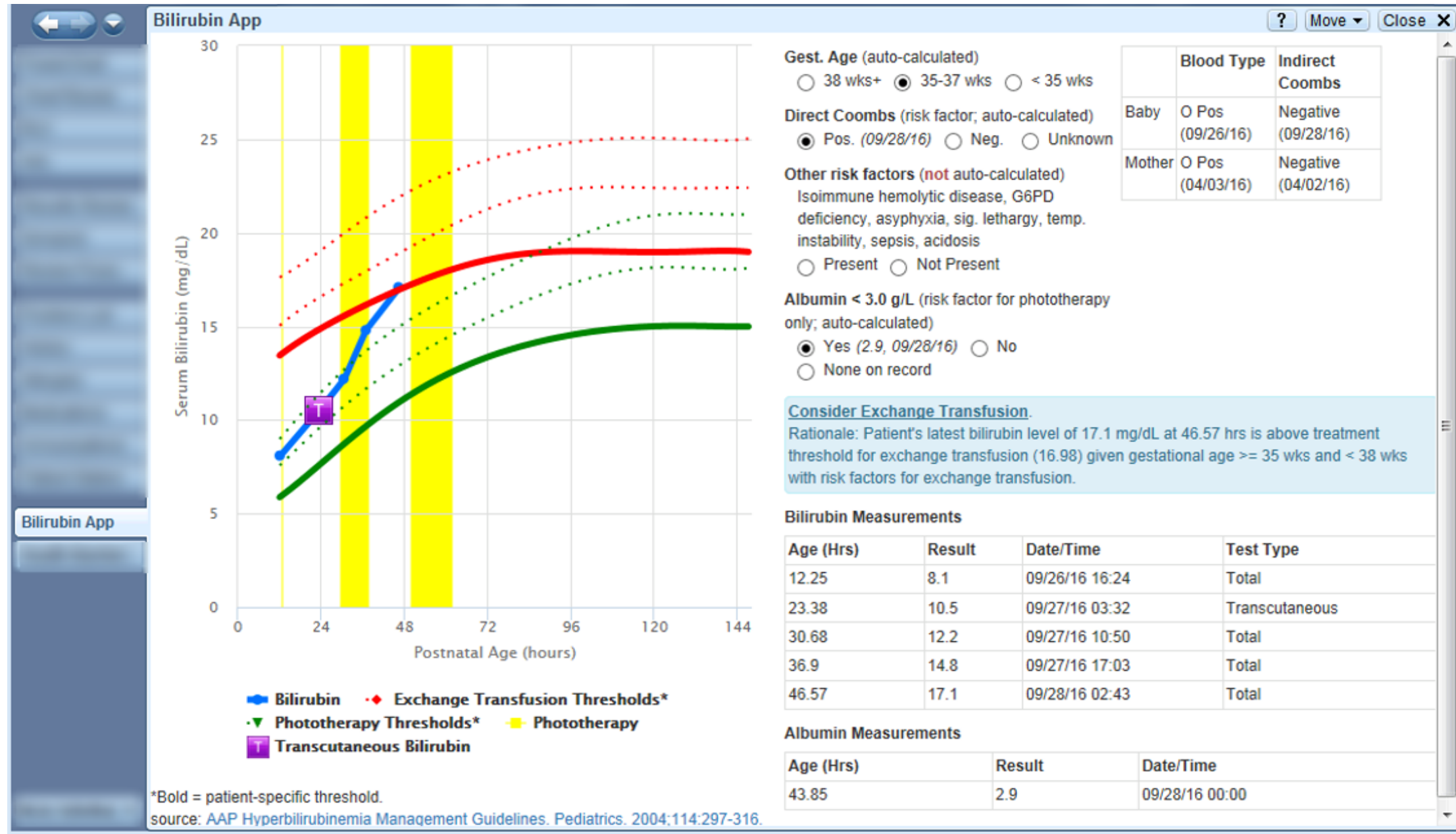
FHIR US CORE CONDITION PROFILE

Name	Flags	Card.	Type	Description & Constraints
Condition	I	0..*		US Core Condition Profile us-core-1: A code in Condition.category SHOULD be from U set.
id	Σ	0..1	id	Logical id of this artifact
meta	Σ	0..1	Meta	Metadata about the resource
implicitRules	?! Σ	0..1	uri	A set of rules under which this content was created
language		0..1	code	Language of the resource content Binding: Common Languages (extensible)
text	I	0..1	Narrative	Text summary of the resource, for human interpretation
contained		0..*	Resource	Contained, inline Resources
extension		0..*	Extension	Additional Content defined by implementations
modifierExtension	?!	0..*	Extension	Extensions that cannot be ignored
identifier	Σ	0..*	Identifier	External Ids for this condition
clinicalStatus	?! S Σ I	0..1	code	active recurrence inactive remission resolved Binding: Condition Clinical Status Codes (required)
verificationStatus	?! S Σ I	1..1	code	provisional differential confirmed refuted entered-in-er Binding: ConditionVerificationStatus (required)
category	S	1..*	CodeableConcept	problem-list-item encounter-diagnosis Binding: US Core Condition Category Codes (preferred)
severity		0..1	CodeableConcept	Subjective severity of condition Binding: Condition/Diagnosis Severity (preferred)
code	S Σ	1..1	CodeableConcept	Identification of the condition, problem or diagnosis Binding: Problem Value Set (extensible)
bodySite	Σ	0..*	CodeableConcept	Anatomical location, if relevant Binding: SNOMED CT Body Structures (example)

SUPPORT/WORK WITH CIMI AND HSPC

- HL7 Clinical Information Modeling Initiative (CIMI)
 - Detailed clinical models for true interoperability
 - Tooling to generate and leverage FHIR profiles
 - E-phenotyping efforts will likely fail without this layer
- Healthcare Services Platform Consortium (HSPC)
 - Includes effort to build and share interoperable FHIR interfaces prior to native EHR vendor support

EXAMPLE APP USING CUSTOM FHIR EXTENSIONS



QUESTION 2

- How might machine-learning and other advanced computational tools be used to improve electronic phenotyping in the eMERGE network?

RECOMMENDATIONS

- Focus on basic approaches easiest to scale (rule-based processing of structured data)
- Make judicious use of natural language processing (NLP)
- Synergize with other related efforts
 - E.g., NCI U24 grant (PI: Kawamoto and Del Fiol) to identify and manage patients at risk of early breast and colorectal cancer

QUESTION 3

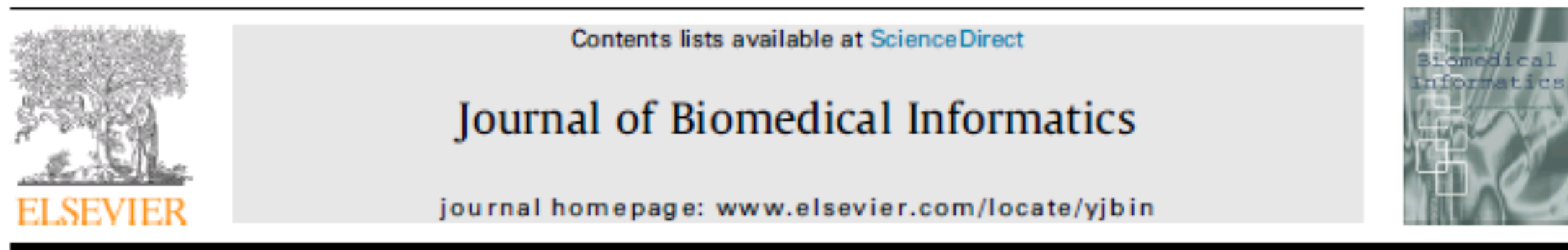
- How can eMERGE assess phenotype comparability across diverse patient populations and diverse healthcare settings (e.g. academic and county hospitals, community clinics and other national healthcare systems)?

FOCUS ON AREAS WITH GOLD STANDARDS

- Establishing gold standard phenotypes can be very costly
- Leverage areas where significant effort has gone into establishing gold standards
 - E.g., ACS National Surgical Quality Improvement Program (NSQIP) registry

SEEK MANUAL PHENOTYPING EFFICIENCIES

- Leverage NLP-facilitated phenotyping
- Study alternate approaches to manual phenotype validation



Single-reviewer electronic phenotyping validation in operational settings: Comparison of strategies and recommendations



Polina Kukhareva^{a,*}, Catherine Staes^a, Kevin W. Noonan^b, Heather L. Mueller^b, Phillip Warner^a, David E. Shields^a, Howard Weeks^{b,c}, Kensaku Kawamoto^a

LEVERAGE INCREASING EHR CONSOLIDATION

- Develop phenotyping and phenotype validation approaches optimized for most frequently used EHR systems
- Significant reach even with a relatively small number of EHR platforms

SUMMARY OF RECOMMENDATIONS

- Learn from, and synergize with, related efforts
- Focus on basic approaches easiest to scale
- Make judicious use of NLP
- Leverage areas where significant effort has gone into establishing gold standards
- Seek manual phenotyping efficiencies
- Leverage increasing EHR consolidation

THANK YOU!

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