Phenotype ideas

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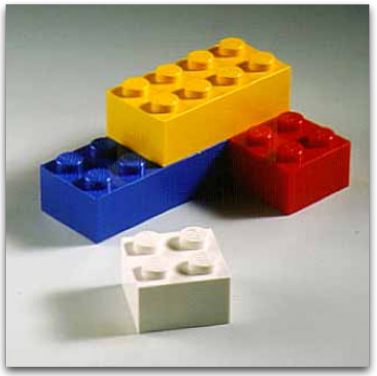
Extending the Lessons Learned

- To expand the 'EMR phenotype workforce' beyond eMERGE, what do the lessons learned say about what skills and technologies are needed for an institution (say, a 'typical CTSA awardee' with a research data warehouse containing EMR-derived data) to be able to join the PheKB club and be both a user and producer of new phenotype specifications?
- Stated otherwise, what would be needed to scale up to dozens or hundreds of collaborating institutions?

Phenotype Lego Blocks



- Can complex phenotypes be decomposed into component sub-units that can be concatenated in (arbitrary?) combinations like modular software, representing things such as physiologic states (e.g., fever, weight loss, renal insufficiency, hypoxia, etc.) as well as the deceptively simple ones (e.g., gender, age)?
- A modular approach to assembly of new phenotypes would potentially have a large leveraging effect on institutions not having to re-invent every new clinically-relevant phenotype from scratch.

The *eMERGE* Network
electronic Medical Records & Genomics
A consortium of biorepositories linked to electronic medical records data for conducting genomic studies

Making Decision Support Real

- Is eMERGE ready and able to commit to a prototype consortium-wide closed-loop clinical decision support demonstration project which would employ a standardized and automated decision support 'package‘:
  - all of the needed components for "IF {patient-specific complex clinical phenotype==true} THEN do this" and measure effects of the decision support intervention in actual healthcare operations across all consortium members.
  - If not now, when?

The scope of decision support

- "Rule based systems" do not mean providers must follow rules. Rules in informatics context = computerized approach to identification of characteristics.
- Examples of interventions
  - Educational prompts: here is additional general information to consider in this setting.
  - Data gathering prompts: given what is known about this {genotype|phenotype|genotype+phenotype}, it would be helpful to get this additional observation or testing.
  - Guidance that improves certainty of diagnosis given data currently available.
  - Guidance for best-evidence-based therapy selection.
  - Information relevant to prevention and/or prognosis.