PSQ Core Focus Areas

• **Phenotypes**
  – Support and discussion for individual groups
  – General guidance (Living Text, web page, journals and conferences)
  – Grow a library or collection of phenotype definitions

• **Data Standards**
  – Propose “standard” phenotype definitions and approaches to EHR data
  – How to get these used and reported in studies (the “Table 1 project”)

• **Data Quality**
  – [white paper](#) (Meredith Zozus, lead), part of online Living Text
  – Watching PCORI Data Quality Group, and DQ Common Data Model
  – The use of population-level data is essential to explore, measure, and report “data quality” so that the results can be appropriately interpreted.
  – Need adequate data and methods to detect the likely and genuine variation between populations at different trial sites and/or intervention groups.
Challenge - Presenting Baseline Characteristics for Clinical Trial Reporting

Multiple possible phenotypes:

**SUPRME-DM Phenotype**

**Definition:**
Adult Durham Population patients who meet **ONE OR MORE** of the following criteria during a DukeMed encounter between 2007-2011:

- One or more instances of the specified ICD-9-CM diagnosis codes (see table 7) on an **inpatient** encounter
- OR 2 or more instances of the specified ICD-9-CM diagnosis codes (see table 7) on **outpatient** encounters on separate days
- OR 1 or more instances of active stand-alone medication (see table 8) reported during outpatient medication reconciliation
- OR 1 or more Oral Glucose Tolerance Test (OGTT) 2-hour 75g result >= 200 mg/dl where there is NO DIAGNOSIS CODE on the same encounter indicating pregnancy (V22, V23)
- OR 2 or more hemoglobin A1c results >= 6.5% on 2 different days within 730 days span
- OR 2 or more fasting glucose results >= 126 mg/dl on 2 different days within 730 days span
- OR 2 or more random glucose results >= 200 mg/dl on 2 different days within 730 days span
- OR within a 750 days span on 2 different days:
  - Fasting glucose results >= 126 mg/dl
  - AND Random glucose results >= 200 mg/dl
- OR within a 750 days span (can be same day):
  - Hemoglobin A1c results >= 6.5%
  - AND Fasting glucose results >= 126 mg/dl

**Abnormal Lab Results**

**Definition:**
Laboratory results

Adult Durham Population patients who meet **ONE OR MORE** of the following criteria during a DukeMed encounter between 2007-2011:

- One or more instances of hemoglobin A1c results >= 6.5%
- OR one or more fasting glucose results >= 126 mg/dl within 365 days span
- OR one or more random glucose results >= 200 mg/dl within 365 days span

**Abnormal HbA1c (NCY A1c Registry Definition)**

**Definition:**
Adult Durham Population patients who meet **ONE OR MORE** of the following criteria during a DukeMed encounter between 2007-2011:

- One or more instances of hemoglobin A1c results >= 6.5%
Collaboratory Approaches

Current approach:

• Support search and evaluation of existing phenotypes
• Facilitate documentation
• Share definitions
• Share implementation information and results
• Link to other resources
• Promote as “best available”
• Work toward standards
Collaboratory Approaches

Current approach:
• Support search and evaluation of existing phenotypes
• Facilitate documentation
• Share definitions
• Share implementation information and results
• Link to other resources
• Promote as “best available”
• Work toward standards

Supporting activities:
• List of authoritative sources
• Create documentation “templates”:
  • Logic
  • Supporting evidence
  • Implementation guidance
• Develop methods for scientific validation studies
• Data quality recommendations for any pragmatic trials using EHR data
Presentation for phenotypes (in progress)

Phenotype Definitions Used

**Populations:**
- Patients with chronic pain
- Patients with imaging studies for lower back pain
- Patients who are candidates for CRC screening

**Confounders or Risks:**
- Diabetes
- Hypertension

**Outcomes:**
- Mortality
- Suicide attempt

Selection and planning:
- Justification and guidance for use in Pragmatic Trials

Implementation:
- Human readable phenotype, collaboration, versioning, public dissemination

Recommendations on Collaboratory website

NIH Collaboratory: Rethinking Clinical Trials

Type 2 Diabetes Mellitus Phenotype Definitions

From the NIH Collaboratory Phenotypes, Data Standards, and Data Quality Core

Available at: https://www.nihcollaboratory.org/Pages/phenotype-repository.aspx

Background: The Phenotypes, Data Standards, and Data Quality Core of the NIH Health Care Systems Research Collaboratory is developing a series of recommendations for the collection/query of data from electronic health records (EHRs) and/or ancillary systems for person characteristics and clinical features to support standardized reporting of baseline characteristics of research populations in intervention and observational studies.

Purpose of this document: This document represents our synthesis of existing phenotype definitions that have been used in diabetes research and population health activities. Using guidelines for the evaluation of existing phenotypes, our informatics and EHR phenotyping experience, and specialized clinical/research expertise, we suggest a suite of phenotype definitions, each appropriate for a particular purpose. The following is our recommendation, complete with a justification and supporting information and resources, for explicit EHR-derived phenotype definitions for diabetes. However, neither the Collaboratory nor the NIH has formally endorsed these definitions or their use in the data collection or reporting of this condition at this time (see disclaimer).

Audience: This document and supporting information is directed to clinical researchers and research sponsors who are making decisions about the data to use for studies. These documents should provide specifications and guidance that will assist researchers in making informed and deliberate choices about EHR data to use in research studies. The supporting...

In the future...

Standard code lists (VSAC) or executable code
Collaboratory Approaches

Current approach:
• Support search and evaluation of existing phenotypes
• Facilitate documentation
• Share definitions
• Share information
• Link to other resources
• Promote as “best available”
• **Work toward standards**

Advocate:
• Evaluate and compare
• Facilitate use
• Explore incentives

Research sponsors
SDOs
Policy makers

Work toward standards
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