NIH Collaboratory
Health Care Systems Research Collaboratory
Rethinking Clinical Trials ™

Suggestions for Identifying Phenotype Definitions Used in Published Research

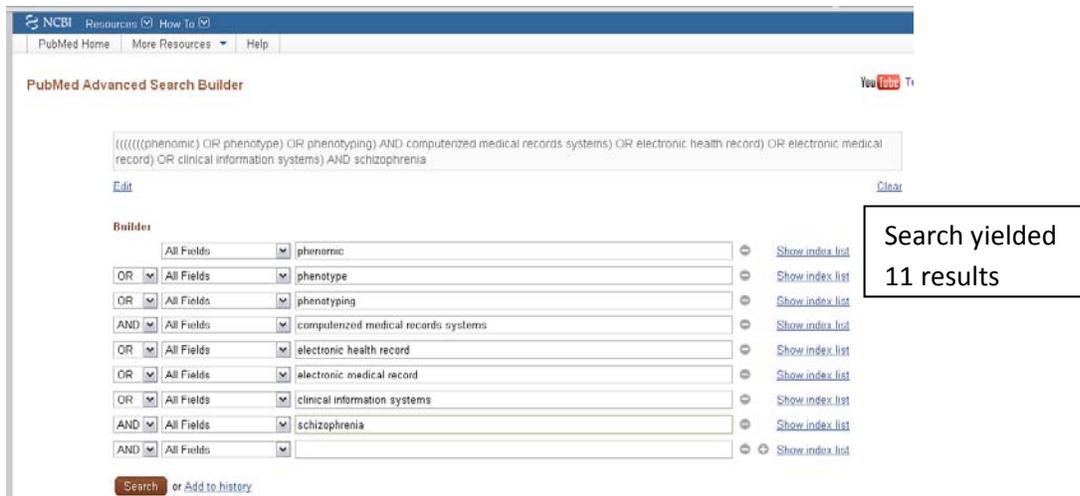*From the NIH Collaboratory Phenotypes, Data Standards, and Data Quality Core*

Similar to other areas of scientific inquiry, a researcher considering the secondary use of Electronic Health Record (EHR) data for a given purpose would do well to first investigate whether others have done work in the same area, and if so, what methods were used and what the results were. Searching the peer-reviewed literature for publications about "phenotyping" efforts is not as straightforward as searching for more clearly defined and understood concepts, however. In order to discover as many relevant publications as possible, a search strategy is needed that incorporates as many core concepts as possible, while at the same time without including so many that the results set becomes unwieldy.

The Phenotypes, Data Standards, and Data Quality Core of the NIH Health Care Systems Research Collaboratory developed this "how-to" guideline to aid those conducting a literature search for publications related to utilizing EHR data for the purpose of characterizing patients, populations, or cohorts based on existing data within EHRs.

Literature Search in PubMed

1) Recent literature search for true EHR phenotype content:

"phenomic" OR "phenotype" OR "phenotyping" AND EHR (OR alternate terms) AND condition (OR alternate terms)

Alternate terms for inclusion

Parent category for "Electronic Health Record" in MeSH is "Medical Records Systems, Computerized"

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Alternate terms for "Medical Records Systems, Computerized" in MeSH:

- Automated Medical Record Systems
- Automated Medical Records System
- Medical Record System, Automated
- Medical Record Systems, Automated
- Medical Records System, Automated
- Medical Records System, Computerized
- Computerized Medical Records Systems
- Computerized Patient Medical Records
- Medical Records Systems, Automated
- Automated Medical Records Systems
- Computerized Medical Record System
- Computerized Medical Record Systems
- Computerized Medical Records System
- Medical Record System, Computerized
- Medical Record Systems, Computerized
- Automated Medical Record System
- Medical Records, Computerized
- Computerized Medical Records
- Medical Record, Computerized
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- Record, Computerized Medical
- Records, Computerized Medical

Alternate terms for "Electronic Health Record" in MeSH:

- Electronic Medical Record
- Medical Record, Electronic
- Medical Records, Electronic
- Record, Electronic Medical
- Records, Electronic Medical
- Electronic Medical Records
- Health Record, Electronic
- Health Records, Electronic
- Record, Electronic Health
- Records, Electronic Health

Conduct MeSH search on domain area term(s) to identify alternate terms for inclusion in search:
Condition = "schizophrenia"

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Alternate terms for "schizophrenia":

- Schizophrenias
- Schizophrenic Disorders
- Disorder, Schizophrenic
- Disorders, Schizophrenic
- Schizophrenic Disorder
- Dementia Praecox

2) Literature search for computable definitions not labeled as phenotypes

Although not labeled at phenotypes per se, many major and well-recognized clinical trials and registries have used EHR data to identify cohorts, and these sources should be considered for many conditions. A search with disease name + "registry" or "pragmatic trial" might be fruitful.

Clinical and expert guidance can be important for identification of these pivotal trials; another potential technique might be to limit results to high-impact journals via a PubMed search.

Note: This is a dynamic field, and new guidelines and experience with phenotype development are happening continuously. One major forum for these discussions is the *Journal of the American Medical Informatics Association* at http://jamia.bmj.com/. We recommend that anyone using computable phenotypes consult this journal regularly for new updates.

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