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Centralized Interactive Phenomics Resource (CIPHER): A Platform for Electronic Health Data-Based Phenomics Science

Systems Demonstration - Clinical Research Informatics

S56

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VA Boston Healthcare System

#AMIA2023



Disclosure



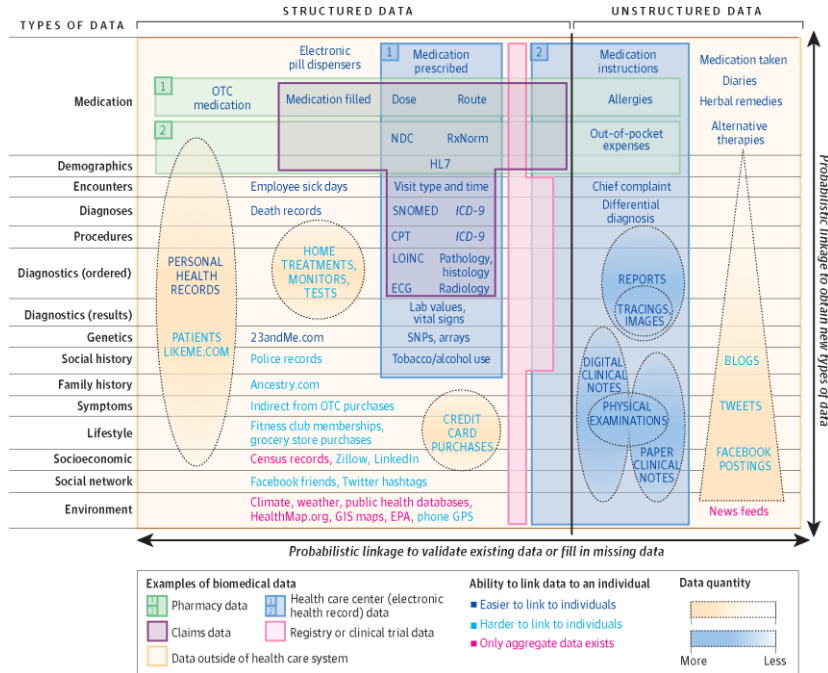
I and my spouse/partner have no relevant relationships with commercial interests to disclose.

Learning Objectives

After participating in this session, the learner should be better able to leverage the Centralized Interactive Phenomics Resource (CIPHER) platform for electronic health records (EHR) based phenotyping by

- searching the library for phenotype definitions,
- contributing phenotypes, and
- browsing connected data visualization tools.

EHRs are a rich resource for clinical research and healthcare operations



Heart failure



Diabetes

- Code curation
- Machine learning
- Other approaches



PTSD

Weber JAMA 2014

Phenotype development has its challenges



Institutional
knowledge



Computing
resources

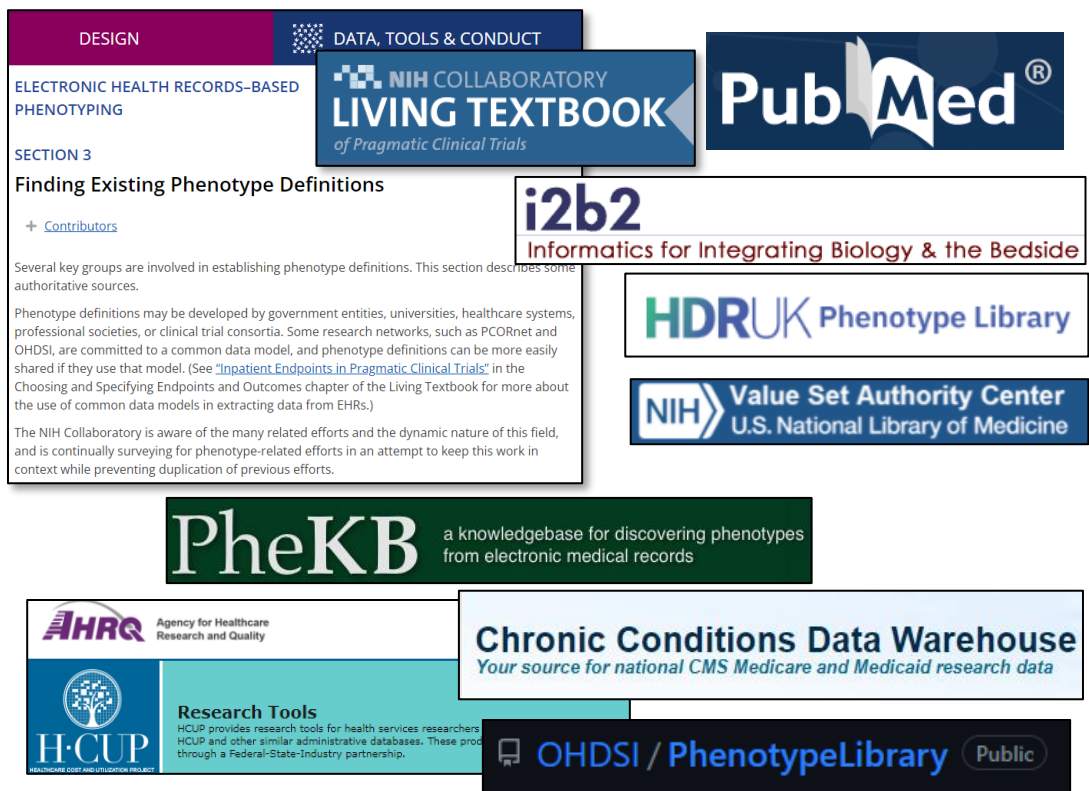


Clinical
expertise



System specific
data variability

Phenotype libraries allow reuse of existing definitions

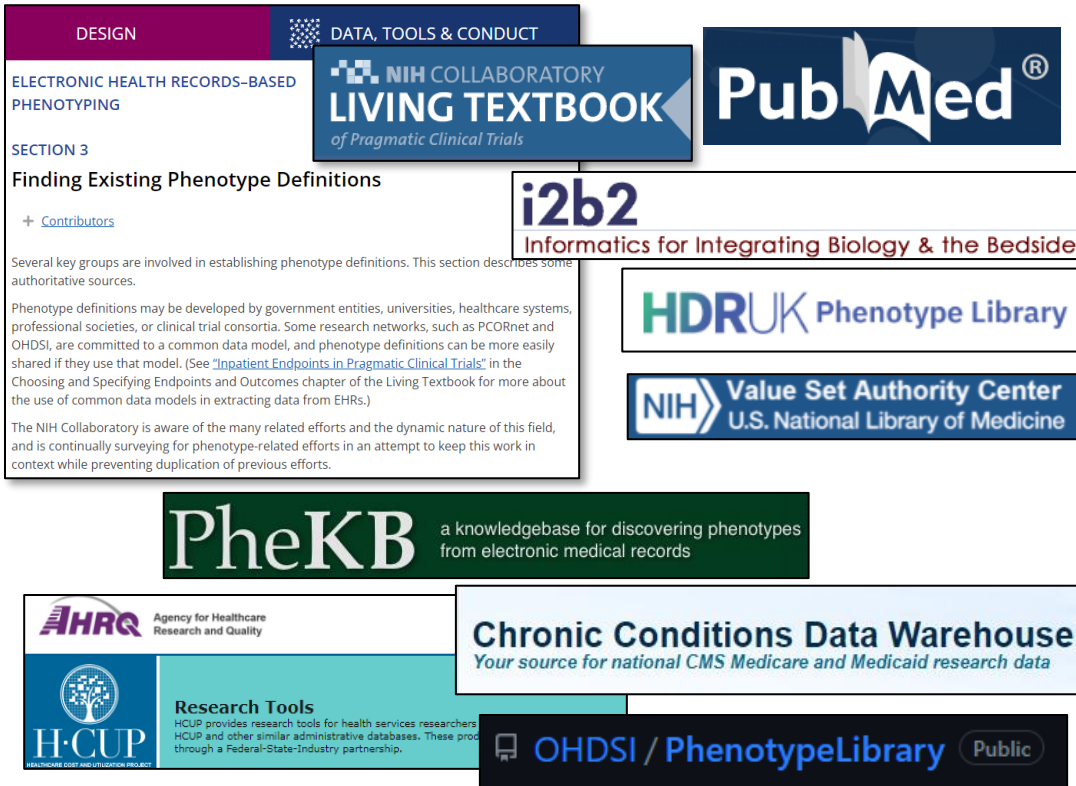


The screenshot shows a webpage with a purple header 'DESIGN' and a blue header 'DATA, TOOLS & CONDUCT'. The main content area is titled 'ELECTRONIC HEALTH RECORDS-BASED PHENOTYPING' and 'SECTION 3 Finding Existing Phenotype Definitions'. It includes a '+ Contributors' link and a paragraph of text. Below the text are several logos and boxes: 'NIH COLLABORATORY LIVING TEXTBOOK of Pragmatic Clinical Trials', 'PubMed', 'i2b2 Informatics for Integrating Biology & the Bedside', 'HDRUK Phenotype Library', 'NIH Value Set Authority Center U.S. National Library of Medicine', 'PheKB a knowledgebase for discovering phenotypes from electronic medical records', 'Agency for Healthcare Research and Quality', 'Chronic Conditions Data Warehouse Your source for national CMS Medicare and Medicaid research data', 'H-CUP Research Tools HCUP provides research tools for health services researchers HCUP and other similar administrative databases. These products are developed through a Federal-State-Industry partnership.', and 'OHDSI / PhenotypeLibrary Public'.

Benefits

- Reuse existing definitions
- Evaluate portability
- Save time and expedite reusability

There are opportunities for improvement and enhancement



Challenges

- Targeted user community
- Focused scope
- Metadata captured
- User features and capabilities

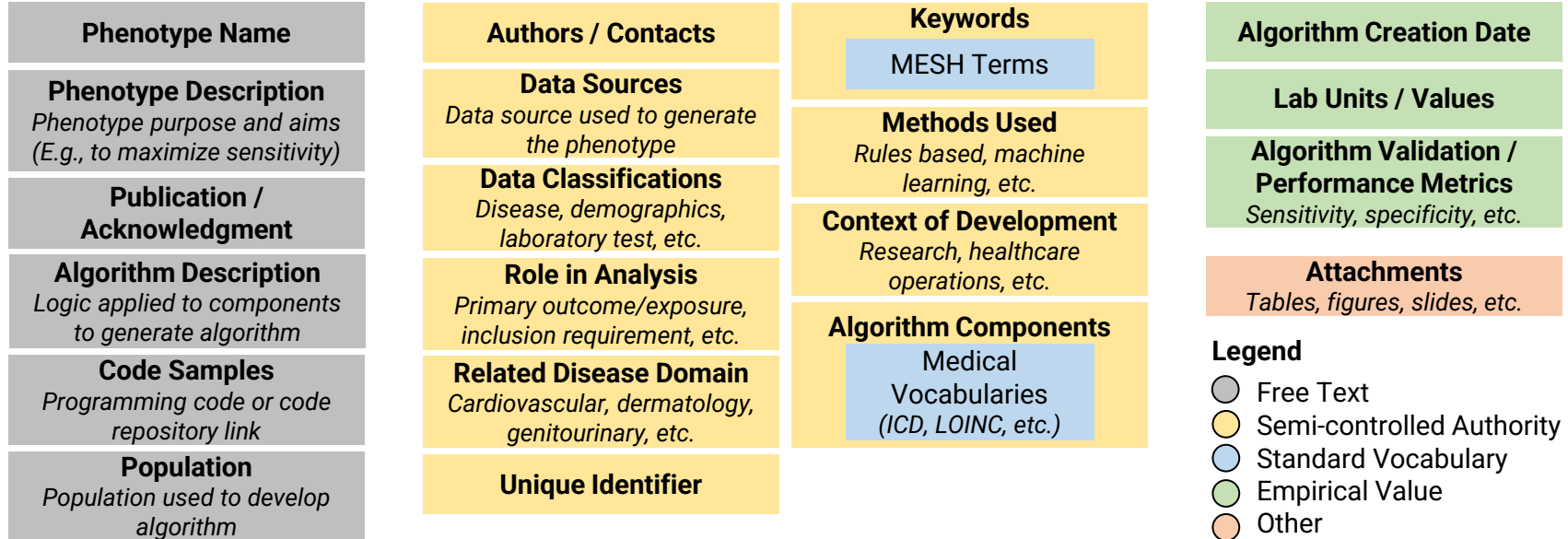
CIPHER: A public phenotype library developed in the VA by phenomics experts



Development Aims

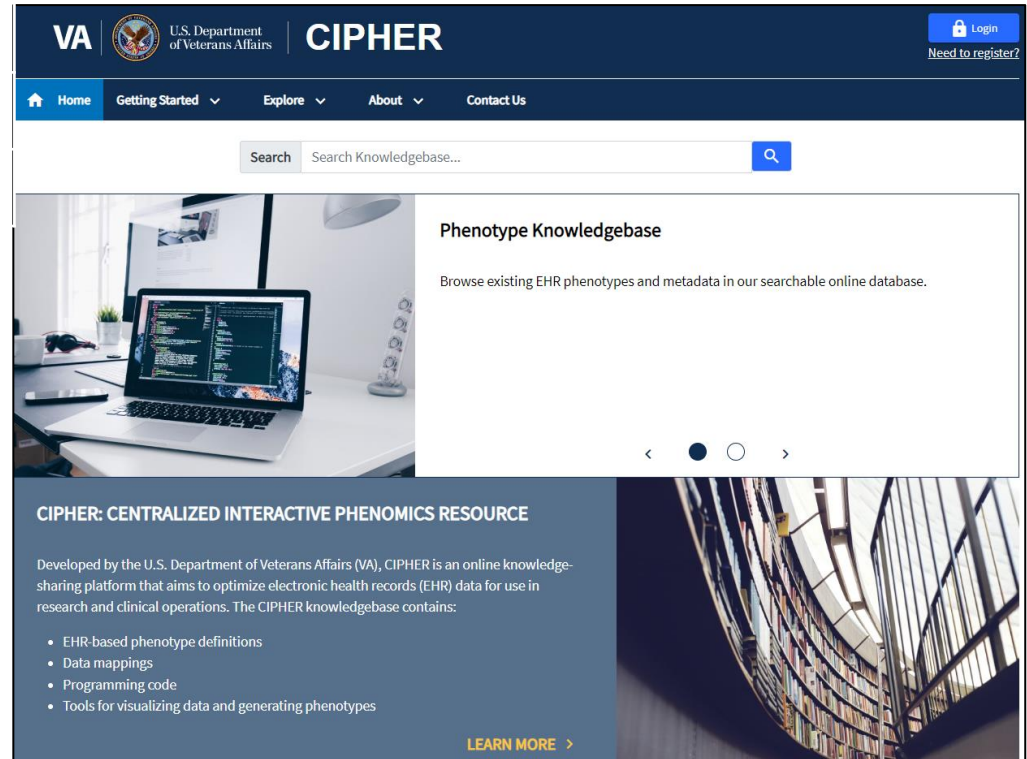
- CIPHER metadata standard
- Scale content management
- Navigation and search
- Integrated visualization tools
- Health system agnostic

CIPHER's phenotype metadata standard



CIPHER website components

1. Phenotype knowledgebase
2. Phenotype collection workflow
3. Data visualization tools



The screenshot shows the CIPHER website homepage. At the top, there is a dark blue header with the VA logo, the U.S. Department of Veterans Affairs name, and the CIPHER logo. A navigation menu includes Home, Getting Started, Explore, About, and Contact Us. A search bar is located below the navigation. The main content area features a large image of a laptop displaying data on a desk. To the right of the image, the text reads "Phenotype Knowledgebase" and "Browse existing EHR phenotypes and metadata in our searchable online database." Below this, there is a section titled "CIPHER: CENTRALIZED INTERACTIVE PHENOMICS RESOURCE" with a description of the platform and a list of features: EHR-based phenotype definitions, Data mappings, Programming code, and Tools for visualizing data and generating phenotypes. A "LEARN MORE" link is at the bottom right of this section.

VA U.S. Department of Veterans Affairs CIPHER

Home Getting Started Explore About Contact Us

Search Search Knowledgebase...

Phenotype Knowledgebase

Browse existing EHR phenotypes and metadata in our searchable online database.

CIPHER: CENTRALIZED INTERACTIVE PHENOMICS RESOURCE

Developed by the U.S. Department of Veterans Affairs (VA), CIPHER is an online knowledge-sharing platform that aims to optimize electronic health records (EHR) data for use in research and clinical operations. The CIPHER knowledgebase contains:

- EHR-based phenotype definitions
- Data mappings
- Programming code
- Tools for visualizing data and generating phenotypes

LEARN MORE >

Searchable database of phenotype articles

1. Phenotype knowledgebase
2. Phenotype collection workflow
3. Data visualization tools

- ✓ CIPHER standard
- ✓ Unique phenotype article identifier
- ✓ Complex searching
- ✓ Change control

The screenshot displays the CIPHER web application interface. At the top, there is a navigation bar with the VA logo, U.S. Department of Veterans Affairs, and the CIPHER logo. A user profile icon labeled 'JH' is in the top right. Below the navigation bar, there are tabs for 'Home', 'Getting Started', 'Explore', 'About', 'Contact Us', and 'Admin'. The main content area is divided into a left sidebar and a main search results area. The sidebar contains a 'Clear Filters' button and a 'Collapse' button, followed by a list of filter categories: Data Classification, Related Disease Domain, Data Sources Used, Algorithm components, Role of phenotype in analysis, Date algorithm created, Author, Method used, and Publication. The 'Author' filter is expanded, showing 'MVP Cognitive Decline and Dementia During Aging Working Group (3)' and 'Million Veteran Program (MVP) (3)'. The 'Publication' filter is set to 'Yes'. The main search area has a search bar with 'dementia' entered. Below the search bar, it says 'Search results for: dementia'. There are sorting options ('Sort by Date algorithm created (descending)'), a dropdown for 'Items per page: 10', and pagination controls ('1-3 of 3'). Two search results are visible, both titled 'Alzheimer's Disease (MVP Cog Working Group)'. The first result lists authors Logue MW, Miller MW, Sherva R, Zhang R, Harrington KM, Fonda JR, Merritt VC, Panizzon MS, Hauger RL, Wolf E J, Neale Z, and Gaziano JM, and includes the text 'Alzheimer's disease and related dementias among aging veterans: Examining gene-by-environment interactions with post-traumatic stress disorder and traumatic brain injury, Alzheimers Dement. 2022 Dec 22. doi: 10.1002/alz.12870. Epub ahead of print. PMID: 36546606., MVP Cognitive Decline and Dementia During Aging Working Group, Million Veteran Program (MVP)'. The second result is titled 'Alzheimer's Disease, Non-specific Dementias (MVP Cog Working Group)' and lists the same authors and text.

Standardized collection of phenotype metadata

1. Phenotype knowledgebase
2. Phenotype collection workflow
3. Data visualization tools

- ✓ Validation against standard vocabularies
- ✓ CIPHER review
- ✓ Populate into knowledgebase

VA U.S. Department of Veterans Affairs CIPHER JH

Add Algorithm Component

② Enter Algorithm Component Codes

ICD-9 Diagnostic Codes
How would like to add the information?

Search for a code by its name or description

Perform a wildcard search by using % or *

Enter codes directly. You may provide multiple codes separated by a comma. No other special characters are allowed.

434.%

- ✓ 434.0 - CEREBRAL THROMBOSIS
- 434.00 - CEREBRAL THROMBOSIS W/O MENTION OF CEREBRAL INFARCTION
- ✓ 434.01 - CEREBRAL THROMBOSIS W/ CEREBRAL INFARCTION
- 434.10 - CEREBRAL EMBOLISM W/O MENTION OF CEREBRAL INFARCTION
- ✓ 434.1 - CEREBRAL EMBOLISM
- ✓ 434.11 - CEREBRAL EMBOLISM W/ CEREBRAL INFARCTION

433.01 433.11 433.21 433.31

433.81 433.91 434.01 434.11

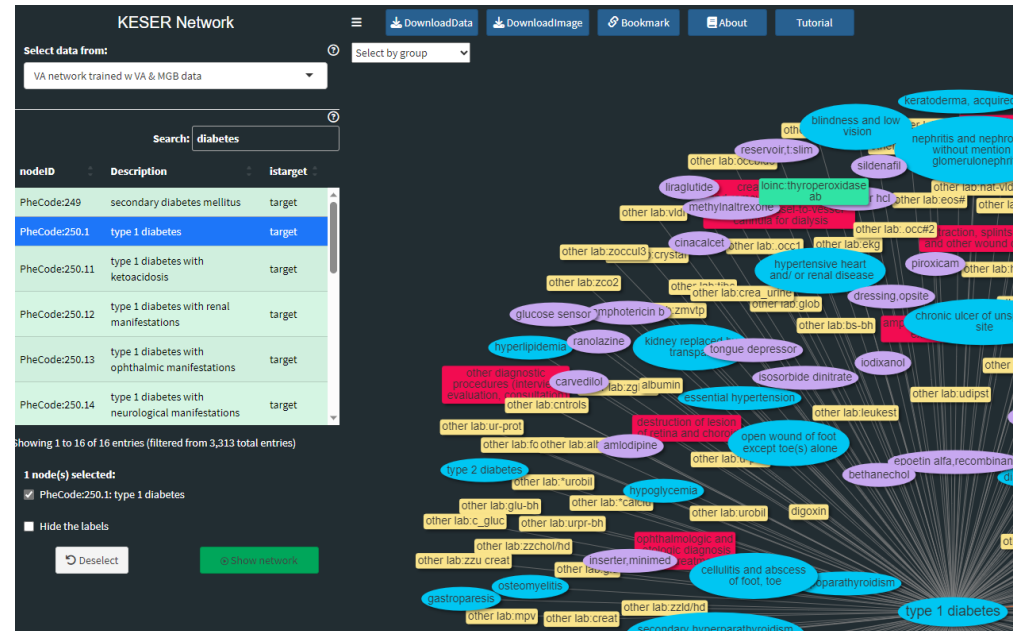
434.91 434.0 434.1 434.9

OK Cancel

Tools connected to phenotype definition knowledgebase

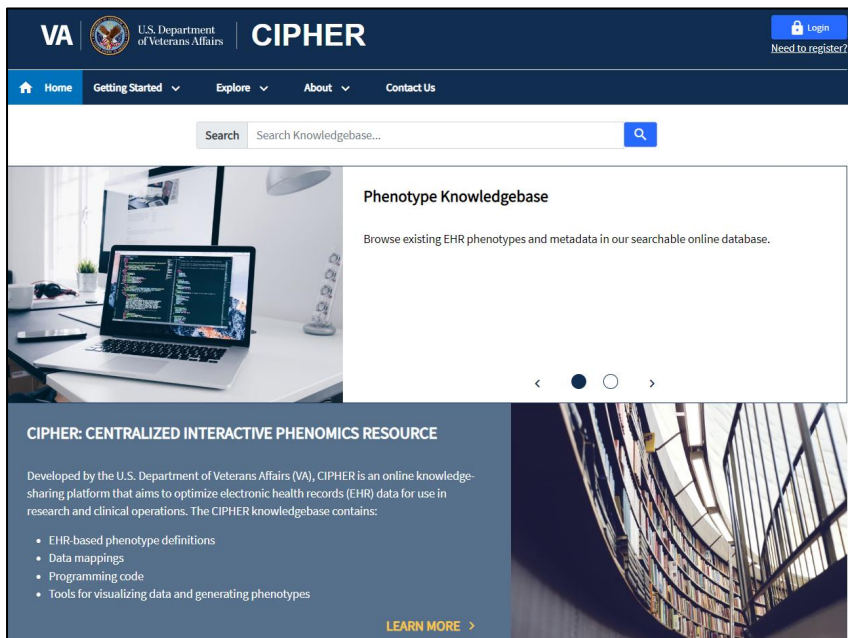
1. Phenotype knowledgebase
2. Phenotype collection workflow
3. Data visualization tools

- ✓ *ICD Hierarchy*: View code mappings
- ✓ *KESER*: Aid phenotype development
- ✓ *Link to knowledgebase*



Hong NPJ Digit Med 2021

CIPHER demonstration



The screenshot shows the CIPHER website interface. At the top, there is a navigation bar with the VA logo, "U.S. Department of Veterans Affairs", and "CIPHER". A search bar is located below the navigation bar. The main content area features a "Phenotype Knowledgebase" section with a description: "Browse existing EHR phenotypes and metadata in our searchable online database." Below this, there is a section titled "CIPHER: CENTRALIZED INTERACTIVE PHENOMICS RESOURCE" with a brief description and a list of features:

- EHR-based phenotype definitions
- Data mappings
- Programming code
- Tools for visualizing data and generating phenotypes

A "LEARN MORE >" link is visible at the bottom right of the main content area.



<https://phenomics.va.ornl.gov/>

CIPHER's user base continues to grow

Current Users



MILLION
VETERAN
PROGRAM

VA



U.S. Department
of Veterans Affairs



HARVARD
MEDICAL SCHOOL



Mass General Brigham

Boston
MCRC



Total Phenotypes

4,200+

Published

3,900+

Future Collaborations



CENTERS FOR DISEASE
CONTROL AND PREVENTION



National Institutes of Health
Turning Discovery Into Health



Agency for Healthcare
Research and Quality



CHARGE
CONSORTIUM
COHORTS FOR HEART & AGING RESEARCH
IN GENOMIC EPIDEMIOLOGY



CENTERS FOR MEDICARE & MEDICAID SERVICES



U.S. Department of Defense



Strengths and limitations of CIPHER

Strengths

- Standard metadata collection
- Healthcare system agnostic
- Validation of standard vocabularies
- Integrated data visualization tools
- Structure is designed for expansion

Limitations

- Licensed data elements may have restrictions (UMLS)
- No linkage to patient level data

Future directions and next steps

Future directions

- Expand database content
- Integrate additional tools
- Expand partners and contributors
- Collect user feedback

How you can use CIPHER

- Browse phenotypes and tools
- Contribute phenotypes and tools
- Share user feedback and ideas

Acknowledgements

- Kelly Cho
- Anne Ho
- Francesca Fontin
- Ashley Galloway
- Jeff Gosian
- Monika Maripuri
- Michael Murray
- Rahul Sangar
- Joanne Sordillo
- Vidisha Tanukonda
- Edward Zielinski
- Suma Muralidhar
- Mike Gaziano
- Rachel Ramoni
- Katherine Liao
- Tianxi Cai
- David Gagnon
- Stacey Whitbourne
- SuChun Cheng
- Vidul Panickan
- Andy Zimolzak
- John Russo
- Laura Davies
- David Heise
- Keith Connatser
- Chad Steed
- Anuj Kapadia

VA



U.S. Department
of Veterans Affairs



Mass General Brigham

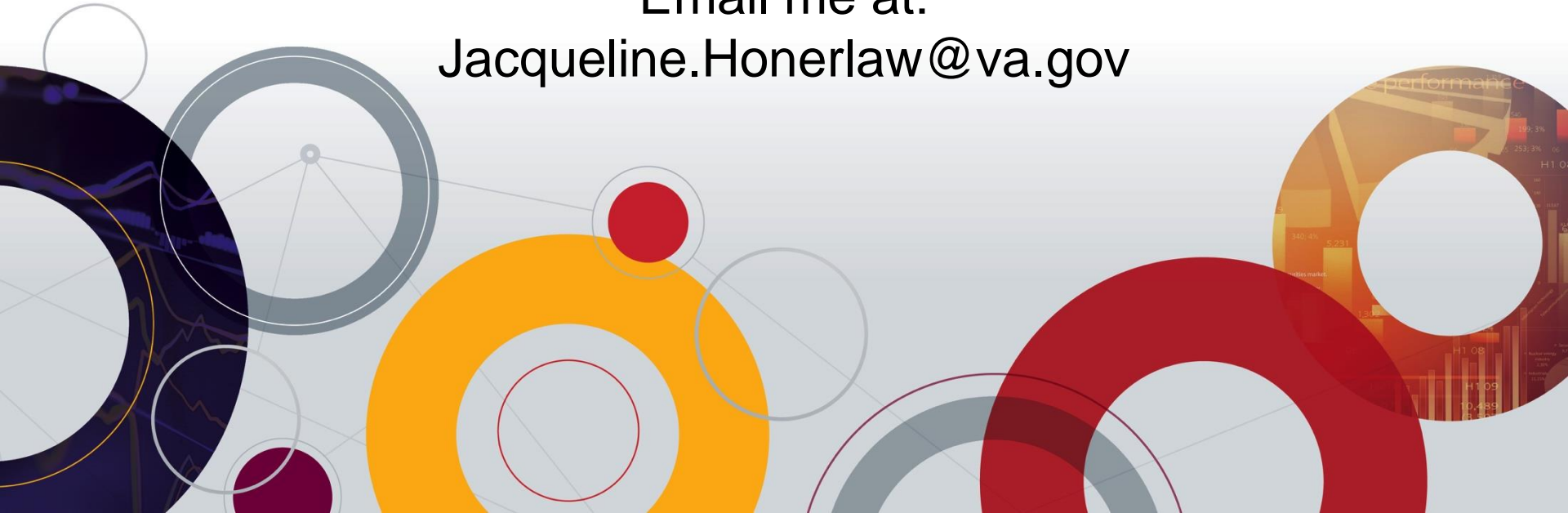




<https://phenomics.va.ornl.gov/>

Thank you!

Email me at:
Jacqueline.Honerlaw@va.gov



Search for a phenotype definition

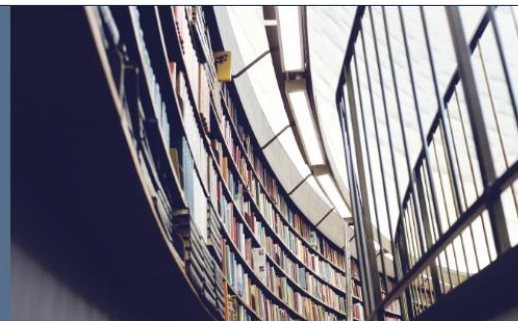
The screenshot shows the top navigation bar of the CIPHER website. It includes the VA logo, the U.S. Department of Veterans Affairs name, and the CIPHER logo. There are links for 'Login' and 'Need to register?'. Below the navigation bar is a search bar with the placeholder text 'Search Knowledgebase...'. The main content area features a large image of a laptop displaying code on a desk. To the right of the image is the heading 'Phenotype Knowledgebase' and the text 'Browse existing EHR phenotypes and metadata in our searchable online database...'. Below this text are navigation arrows.

This inset screenshot shows a dropdown menu from the 'Explore' navigation item. The menu options are 'Visualization Tools', 'About Visualization Tools', and 'Browse Knowledgebase'. The 'Browse Knowledgebase' option is highlighted with a red rectangular border.

CIPHER: CENTRALIZED INTERACTIVE PHENOMICS RESOURCE

Developed by the U.S. Department of Veterans Affairs (VA), CIPHER is an online knowledge-sharing platform that aims to optimize electronic health records (EHR) data for use in research and clinical operations. The CIPHER knowledgebase contains:

- EHR-based phenotype definitions
- Data mappings
- Programming code
- Tools for visualizing data and generating phenotypes



Search for a phenotype definition

The screenshot displays the CIPHER web application interface. At the top, there is a navigation bar with the VA logo, U.S. Department of Veterans Affairs, and the CIPHER logo. A 'Login' button and a link to 'Need to register?' are also present. Below the navigation bar, there are tabs for 'Home', 'Getting Started', 'Explore', 'About', and 'Contact Us'. The 'Explore' tab is active.

The main content area is divided into a left sidebar and a main search results area. The sidebar contains a list of filters, including 'Data Classification', 'Related Disease Domain', 'Data Sources Used', 'Algorithm components', 'Role of phenotype in analysis', 'Date algorithm created', 'Author', 'Method used', 'Publication', and 'Algorithm code'. A 'Validated' filter is selected, and its value is set to 'Yes'. The search bar at the top of the main area contains the text 'dementia' and a search icon. Below the search bar, the results are displayed, including a link to 'Alzheimer's Disease, Non-specific Dementias (MVP Cog Working Group)' and another link to 'Alzheimer's Disease, Related Dementias (MVP Cog Working Group)'. The search results area also includes a 'Sort by' dropdown menu set to 'Search relevance', an 'Items per page' dropdown menu set to '10', and a page indicator '1 - 3 of 3'.

Review definition: Basic information

Home Getting Started ▾ Explore ▾ About ▾ Contact Us

Basic Information and Contact

General Phenotype

Alzheimer's Disease, Non-specific Dementias (MVP Cog Working Group)

Abbreviations and Keywords

AD+

Publication

[Logue MW, Miller MW, Sherva R, Zhang R, Harrington KM, Fonda JR, Merritt VC, Panizzon Million Veteran Program. Alzheimer's disease and related dementias among aging veterans: interactions with post-traumatic stress disorder and traumatic brain injury. Alzheimers J. Epub ahead of print. PMID: 36546606.](#)

Data Classification(s)

Diseases

Related Disease

Mental/ Behavioral Health Neurology

Author(s)

MVP Cognitive Decline and Dementia During Aging Working Group Million Veteran Program (MVP)

Contact

MVP Cognitive Decline and Dementia During Aging Working Group mark.logue@va.gov

Acknowledgement

Mark W. Logue, Richard Hauger, Victoria Merritt, and Matthew Panizzon. Phenotypes developed by the MVP Cognitive Decline and Dementia During Aging Working Group, with support from VA Grants BX004192 (MWL), BX005749 (MWL), I01 CX001727 (RH), and IK2 CX001952 (VM).

Review definition: Algorithm overview

Home Getting Started ▾ Explore ▾ About ▾ Contact Us

Algorithm Overview

Source of Phenotype Data

CDW (Corporate Data Warehouse)

Context of Development

Research

Role of Phenotype in Analysis

Primary Outcome/Exposure

Phenotype Description

The Million Veteran Program (MVP) Cognitive Decline and Dementia During Aging Working Group created a scale of nested ICD-code based dementia phenotypes, from the most restrictive (AD) to the least restrictive (all cause dementia) - see Table 1 below. These phenotypes were developed for use in genetic studies of dementia, and allow the user to select an appropriate level of specificity for their project.

Population

Million Veteran Program (MVP) enrollees

Date Algorithm Created

01/01/2020

Review definition: Algorithm components

Home Getting Started ▾ Explore ▾ About ▾ Contact Us

Algorithm Components

Method Used

Rules-Based (i.e., only structured data were used)

Algorithm Description

To quality as a case requires the presence of two or more ICD codes

Algorithm Components

ICD-9 Diagnostic Codes (8)

290.0, 290.20, 290.21, 290.3, 294.20, 294.21, 294.8, 331.0

ICD-10 Diagnostic Codes (5)

F03.90, F03.91, G30.1, G30.8, G30.9

(1) Programming Code Other

```
/******  
MVP Phenotype Code for Dementia, ADRD, ADplus and Stricter AD  
Project Name: Alzheimer's Phenotyping in MVP
```


Review definition: Validation & additional information

Validation

Validated

Yes

Description of Validation

We have performed a chart review of n=39 “difficult cases”. That is, MVP participants with either a) subjects with low to moderate AD case probability according to the Multimodal AD (Liao et al. Am Med Inform Assoc, 2019). These were reviewed in tandem by Drs. Hauger and [redacted] classified as “Likely not”, “Possible”, or “Likely” cases of MCI, AD, and dementia. Chart review results were compared to the classifications according to the MAP algorithm and according to our ICD-code based definitions of MCI, AD, ADRD, and dementia (Table 2). The performance of the ICD code set is shown in "Algorithm Performance Measures". The authors also evaluated the suitability of our ICD code based phenotypes in genetic studies by testing their association with the APOE E4 isoform (the strongest AD genetic risk factor) in European-descent MVP subjects (Table 3).

Performance Metric	Value
Sensitivity	0.82
Specificity	0.71
Negative Predictive Value (NPV)	0.75
Positive Predictive Value (PPV)	0.78
Area Under the ROC Curve (AUC)	N/A
Kappa	

Additional Information

Attachments

Type	Description
Alzheimer's Disease, Non-specific Dementias	Additional Information - ICD codes, Performance of MAP

Contributing phenotypes

Contributing Phenotypes

We have an easy-to-use process and resources for contributing your phenotype to CIPHER.

You may start by visiting our [How to Use CIPHER](#) page, which gives an overview of phenotyping, how phenotypes are used, benefits of contributing your phenotype to CIPHER, and general tips for using the CIPHER website.

Next steps to contribute a phenotype:

1. **Create a user account.** Navigate to the top right-hand corner of the screen and choose to register a new account. Please enter a valid email that you have access to. You will need this to verify your account.
2. **Complete the Phenotype Entry Form.** Open the phenotype entry form by clicking [here](#). You can also navigate to the form directly via the navigation bar at the top of the screen, under the Getting Started tab. You may save your work in progress and return to finish entering your phenotype at any time. To resume editing, please click on “My Phenotypes” under the “Getting Started” tab on the navigation bar.
 - a. **Choose the type of phenotype you would like to submit (general, lab, or medication).**
 - b. **Follow the instructions in the online wizard to complete the form.**
3. **Submit your phenotype.** Once you have entered in all your phenotype details, you can review your entry and submit for review.
 - a. **Once your phenotype is submitted it will be reviewed by the CIPHER team.** If our team has any questions regarding the submission, we will reach out to the contact listed on the entry form. Once the phenotype entry is finalized by CIPHER it will be searchable within our [Phenotype Knowledgebase](#).
 - b. You can check the status of your submission by navigating to “My Phenotypes” on the navigation bar under the Getting Started tab.
 - c. Should you have any questions about your submission in the meantime, please contact CIPHER@va.gov.

Contributing phenotypes

VA



U.S. Department
of Veterans Affairs

CIPHER

JH



Home

Getting Started ▾

Explore ▾

About ▾

Contact Us



Admin ▾

Create New Phenotype

My Phenotypes

My Assigned Phenotypes


Items per page: 10 ▾

1 - 10 of 19



Phenotype Name	Author	Last Modified	Status		
▾ Example phenotype	test	just now	Draft		
▾ Example medication phenotype	test	a moment ago	Draft		

Contributing phenotypes

Home Getting Started **▼** Explore **▼** About **▼** Contact Us  Admin **▼**

General Phenotype

Please complete all fields below. You will have the opportunity to review all information at the final step. Save and Finish Later

① Basic Information and Contact

Phenotype Name*:
What is the name of your phenotype?

Abbreviations and Keywords*:
What abbreviations or keywords can help users find your phenotype?

- Keywords will be used as search terms in the library.
- Use the search below to select keywords already stored in the CIPHER database.
- If the keyword is not yet stored, enter in your own and select "Add Entry".
- To remove a keyword please click the "x" next to the word.
- We suggest utilizing [MESH](#) to find relevant keywords.

Contributing phenotypes

Home Getting Started Explore About

Basic Information and Contact

2 Algorithm Overview

Source of Phenotype Data*:
What data sources were used to generate the phenotype?

VA Data

- CDW (Corporate Data Warehouse)
- CSDR (COVID-19 Shared Data Resource)
- DoD (Department of Defense)
- MVP (Million Veteran Program)
- OMOP (Observational Medical Outcomes Part)
- Other (VA Data Source)

Non-VA Data

- CMS (Medicare & Medicaid)
- NDI (National Death Index)
- Other

Context of Development*:
For what purpose did you develop your algorithm? (Choose all that apply.)

- Research
- Healthcare Operations
- Quality Improvement
- Clinical Care
- Other

Role of Phenotype in Analysis*:
Describe how the phenotype was used. Choose all that apply:

- Primary Outcome/Exposure
- Secondary Outcome/ Exposure
- Inclusion/Exclusion Requirement
- Comorbidity/Covariate
- Other

Date Algorithm Created*:
When was your algorithm created?

Choose a date *
11/1/2023

Date Range for Source Data:
What was the date range of the data used to create your algorithm?

Choose a date
MM/DD/YYYY - MM/DD/YYYY

Back Next

Contributing phenotypes

[Home](#) [Getting Started](#) [Explore](#) [About](#) [Contact Us](#)

3 Algorithm Components

Method Used*:
What logic was applied to the algorithm components to create this phenotype? (Select all that apply)

- Rules-Based (i.e., only structured data were used)
- Machine learning: Supervised
- Machine learning: Semi-Supervised
- Machine learning: Unsupervised
- Machine learning: Other machine learning approach
- Other

Contributing phenotypes

Algorithm Components*

Which of the following components were used in your algorithm?

- Please click the “Add Algorithm Components” button to add a collection of standard vocabularies (ICD, LOINC, etc.) and EHR specific variables used to create the algorithm.
- Each component must be added separately.

Add Algorithm Component

ICD-9 Diagnostic Codes (1) 410.00

Algorithm Programming Code

Do you have programming code to share?

Add Code Sample

Back

Next

Edit ICD-9 Diagnostic Codes Algorithm Component

- Search for a code by its name or description
- Perform a wildcard search for a specific code using % or *
- Enter codes directly. You may provide multiple codes separated by a comma (default) or change the delimiter.

410.%

- 410.0 - ACUTE MYOCARDIAL INFARCTION OF ANTEROLATERAL WALL
- 410.00 - ACUTE MYOCARDIAL INFARCTION
- 410.01 - ACUTE MYOCARDIAL INFARCTION
- 410.02 - ACUTE MYOCARDIAL INFARCTION
- 410.1 - ACUTE MYOCARDIAL INFARCTION OF OTHER ANTERIOR WALL
- 410.10 - ACUTE MYOCARDIAL INFARCTION

410.10

(1)

Visualization tools: KESER

Billing codes, or
phenotype in E
throughput phe
10 billing codes

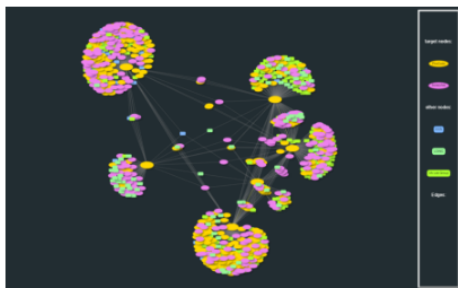
Visualization Tools

About Visualization Tools

Browse Knowledgebase

Million Veteran Program (MVP)

es (ICD) codes, are often leveraged to define a patient's
not always organized meaningfully for the purpose of high-
the use of ICD codes for research by regrouping ICD-9 and ICD-



Knowledge Extraction via Sparse Embedding Regression Network

Infer relatedness among diseases, treatment, procedures and laboratory measurements by creating a visual, interactive knowledge map.

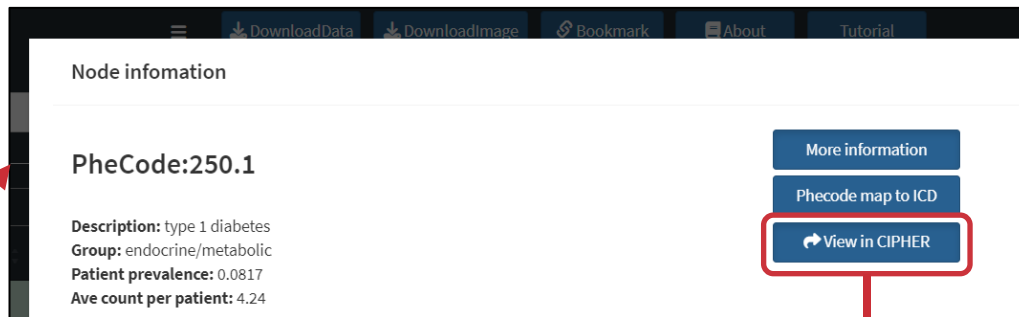
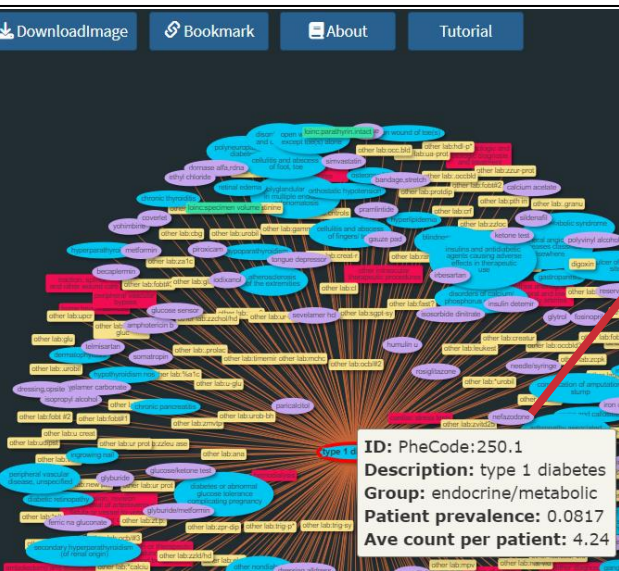
Overview

Uses

Author & Citation

The increasing availability of EHR systems has created enormous potential for translational research. However, it is difficult to know all the relevant codes related to a phenotype due to the large number of codes available. Codified concepts include all ICD diagnosis codes (rolled up to PheCodes), medications (mapped to RxNorm at ingredient level), procedures (rolled up to clinical classification system (CCS) procedure codes), as well as laboratory test results (mapped Logical Observation Identifier Names and Codes (LOINC), short names at the U.S. Department of Veterans Affairs (VA), or local lab codes).

Visualization tools: KESER

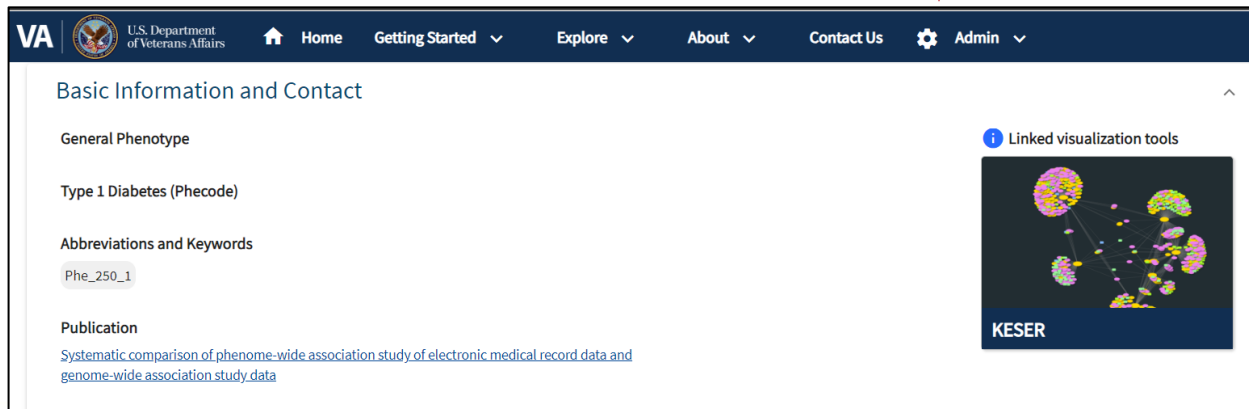


Node information

PheCode:250.1

Description: type 1 diabetes
Group: endocrine/metabolic
Patient prevalence: 0.0817
Ave count per patient: 4.24

[More information](#)
[Phecode map to ICD](#)
[View in CIPHER](#)



VA U.S. Department of Veterans Affairs

Home Getting Started Explore About Contact Us Admin

Basic Information and Contact

General Phenotype

Type 1 Diabetes (Phecode)

Abbreviations and Keywords

Phe_250_1

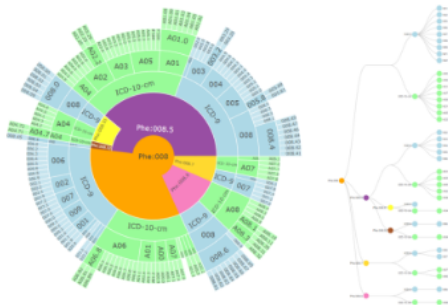
Publication

[Systematic comparison of phenome-wide association study of electronic medical record data and genome-wide association study data](#)

Linked visualization tools

KESER

Visualization tools: ICD Hierarchy



ICD Hierarchy

Visualize mappings from Phecode to ICD-9 and ICD-10 codes.

Overview

Uses

Author & Citation

Electronic health record (EHR)-based studies offer several advantages in research: they are cost efficient, allow for large scale longitudinal analyses, and provide the potential to analyze hundreds of human diseases, drug responses, and many observable clinical traits.

Billing codes, or International Classification of Diseases (ICD) codes, are often leveraged to define a patient's phenotype in EHR-based studies. However, these are not always organized meaningfully for the purpose of high-throughput phenotypic analyses. [PheCodes](#) facilitate the use of ICD codes for research by regrouping ICD-9 and ICD-10 billing codes into clinically relevant phenotypes.

Visualization tools: ICD Hierarchy

PheCode Map with ICD

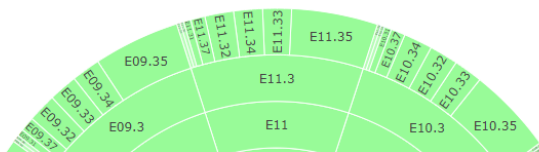
☰ About ? Help

PheCode Mapping with ICD-9 and ICD-10-cm Codes


Phecode	Phenotype	ICD version	ICD code	ICD Description
250	All	All	All	All
250.1	Type 1 diabetes	ICD-9	250.01	type I diabetes mellitus [insulin dependent type] [IDDM] [juvenile type], not stated as uncontrolled, without mention of complication
250.1	Type 1 diabetes	ICD-9	250.03	type I diabetes mellitus [juvenile type], uncontrolled, without mention of complication
250.11	Type 1 diabetes with ketoacidosis	ICD-9	250.11	type I diabetes mellitus [insulin dependent type] [IDDM] [juvenile type], not stated as uncontrolled, with ketoacidosis
250.11	Type 1 diabetes with ketoacidosis	ICD-9	250.13	type I diabetes mellitus [juvenile type], uncontrolled, with ketoacidosis
250.1	Type 1 diabetes	ICD-9	250.21	Diabetes mellitus, type I [insulin dependent type] [IDDM] [juvenile type] with hyperosmolarity, not stated as uncontrolled
250.1	Type 1 diabetes	ICD-9	250.23	Diabetes mellitus, type I [juvenile type] with hyperosmolarity, uncontrolled
250.1	Type 1 diabetes	ICD-9	250.31	Diabetes mellitus, type I [insulin dependent type] [IDDM] [juvenile type], not stated as uncontrolled
250.1	Type 1 diabetes	ICD-9	250.33	Diabetes mellitus with other coma, type I [juvenile type], uncontrolled

Previous 1 2 3 4 5 ... 17 Next

Sunburst



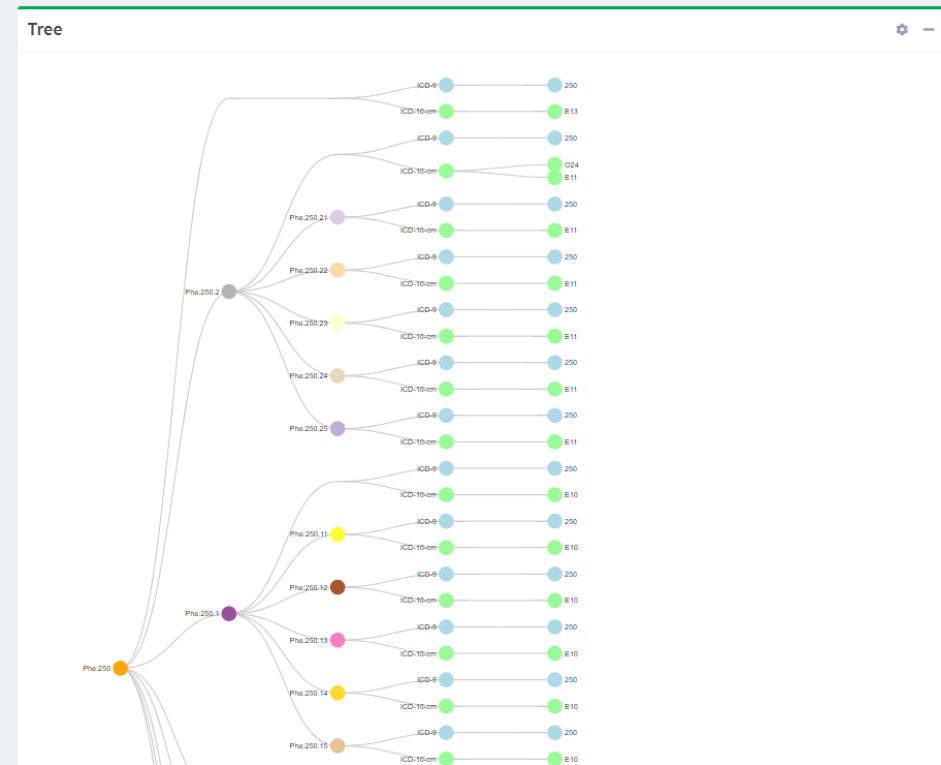
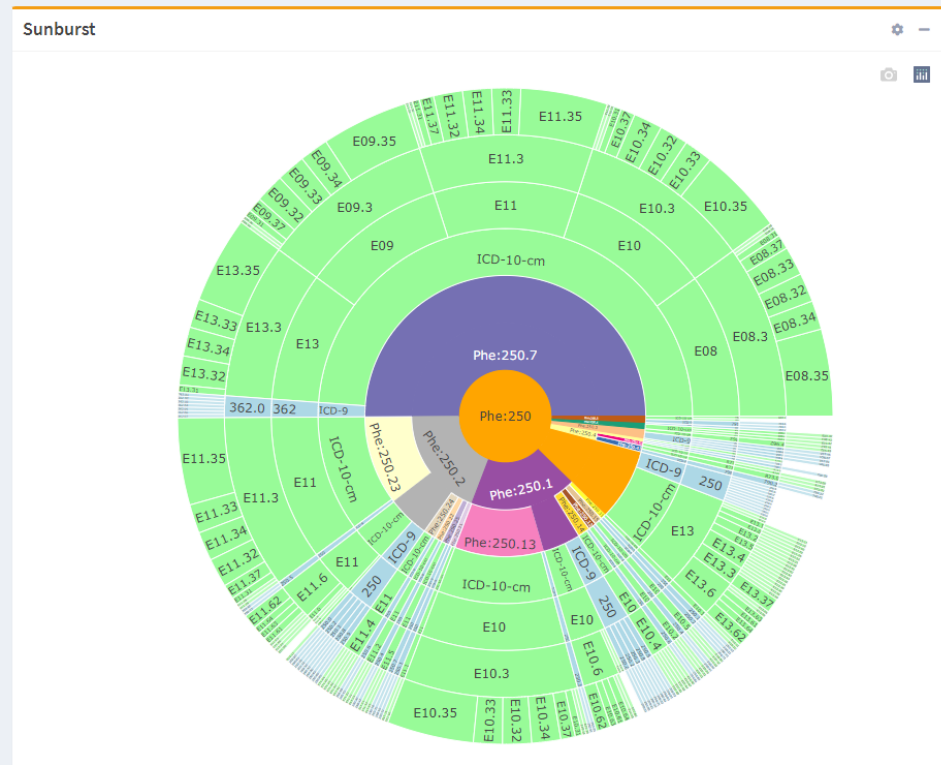
Tree



Legend

- ICD-10-cm
- ICD-9
- Phe:250
- Phe:250.1
- Phe:250.11
- Phe:250.12
- Phe:250.13
- Phe:250.14
- Phe:250.15
- Phe:250.2
- Phe:250.21
- Phe:250.22
- Phe:250.23
- Phe:250.24
- Phe:250.25
- Phe:250.3
- Phe:250.4
- Phe:250.41
- Phe:250.42
- Phe:250.5
- Phe:250.6
- Phe:250.7

Visualization tools: ICD Hierarchy



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Please use the following instructions for citing the use of CIPHER and any phenotypes utilized:

- **Citing the CIPHER Phenotype Library**
 - Please cite use of the CIPHER phenotype library by referencing the article below: Honerlaw J, Ho YL, Fontin F, Gosian J, Maripuri M, Murray M, Sangar R, Galloway A, Zimolzak AJ, Whitbourne SB, Casas JP, Ramoni RB, Gagnon DR, Cai T, Liao KP, Gaziano JM, Muralidhar S, Cho K. Framework of the Centralized Interactive Phenomics Resource (CIPHER) standard for electronic health data-based phenomics knowledgebase. J Am Med Inform Assoc. 2023 Mar 7:ocad030. doi: 10.1093/jamia/ocad030. Epub ahead of print. [PMID: 36882092](#).
- **Citing Specific Phenotypes**
 - If the phenotype has an associated publication listed on the phenotype page, please reference it.
 - If the phenotype does not include a publication, please follow the guidance in the acknowledgements section of the phenotype and/or cite the URL of the phenotype page used.

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- Increases the reproducibility of your methods and the number of citations of your publications.
- Facilitates collaboration by sharing your work products, enabling collaboration requests, and opportunity to obtain feedback from the CIPHER user community.
- Provides opportunities to have your work featured in CIPHER newsletters and communications to the CIPHER user community.
- Affords you with a sharable platform to disseminate work products and supports compliance with the FAIR Data Principles.