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BRICS Seminar

FHIR Data Element Mapping Tool

UMLS in BRICS Data Dictionary - why UMLS?

UMLS Tool Update

AI Search on Data Elements

Biomedical Research Informatics Computing System (BRICS)

October 10th, 2024

CiSTAR

cdRNS
Common Data Repository
for Nursing Science

National Ophthalmic Disease Genotyping Network
eyeGENE®

MTBI²
Uniformed
Services
University

PDBP
Parkinson's Disease
Biomarkers Program

National Institute of
Neurological
Disorders and Stroke

FITBIR
Federal Interagency Traumatic Brain Injury Research
INFORMATICS SYSTEM

Logistics

Audio/Video	Please keep your microphone muted
Recording	• Today's session will be recorded • Will be posted on the BRICS website: https://brics.cit.nih.gov/demo
Questions & Comments	• We encourage your participation today • Please use the chat for questions & comments. The chat will be monitored throughout today's demo. • There will also be time after each speaker and at the end of the demo to ask live questions.

Agenda

Time	Topic	Speaker(s)
9:05 AM-9:20 AM	FHIR Data Element Mapping Tool	Dr. Henry Ogoe
9:20 AM-9:25 AM	UMLS in BRICS Data Dictionary - why UMLS?	Dr. Olga Vovk
9:25 AM–9:35 AM	UMLS Tool – Updates	Dr. Alexandra Bokinsky
9:35 AM-9:55 AM	AI Search on Data Elements	Dr. Maria Bagonis
9:55 AM-10:00 AM	Closing Remarks	Dr. Matthew McAuliffe

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FHIR Data Element Mapping Tool

Dr. Henry Ogoe

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FITBIR Federal Interagency Traumatic Brain Injury Research Informatics System

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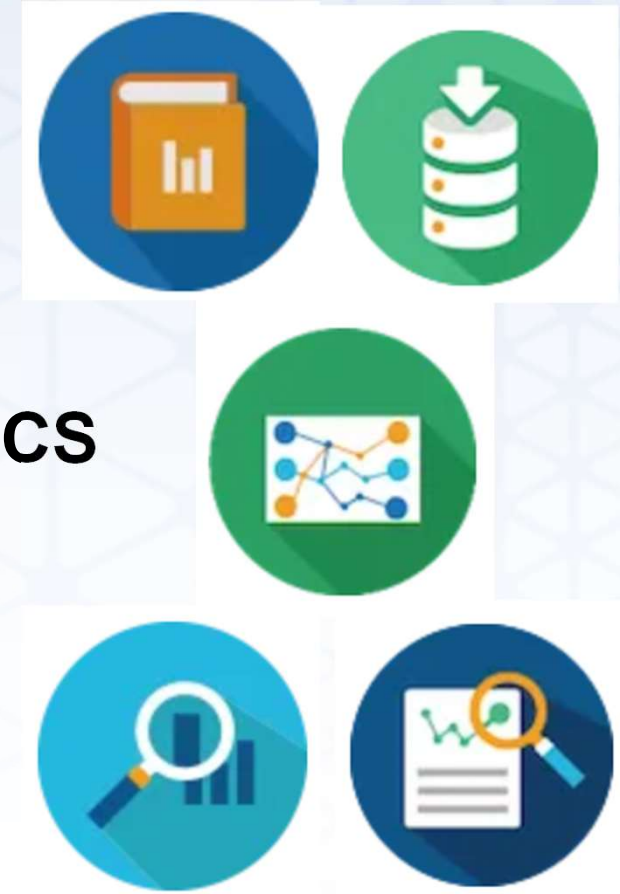
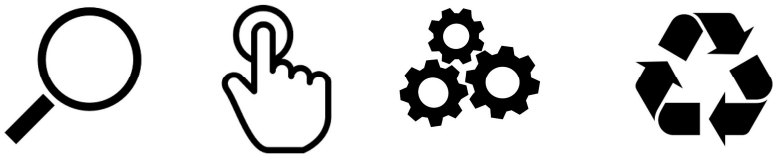
BRICS-on-FHIR

FHIR

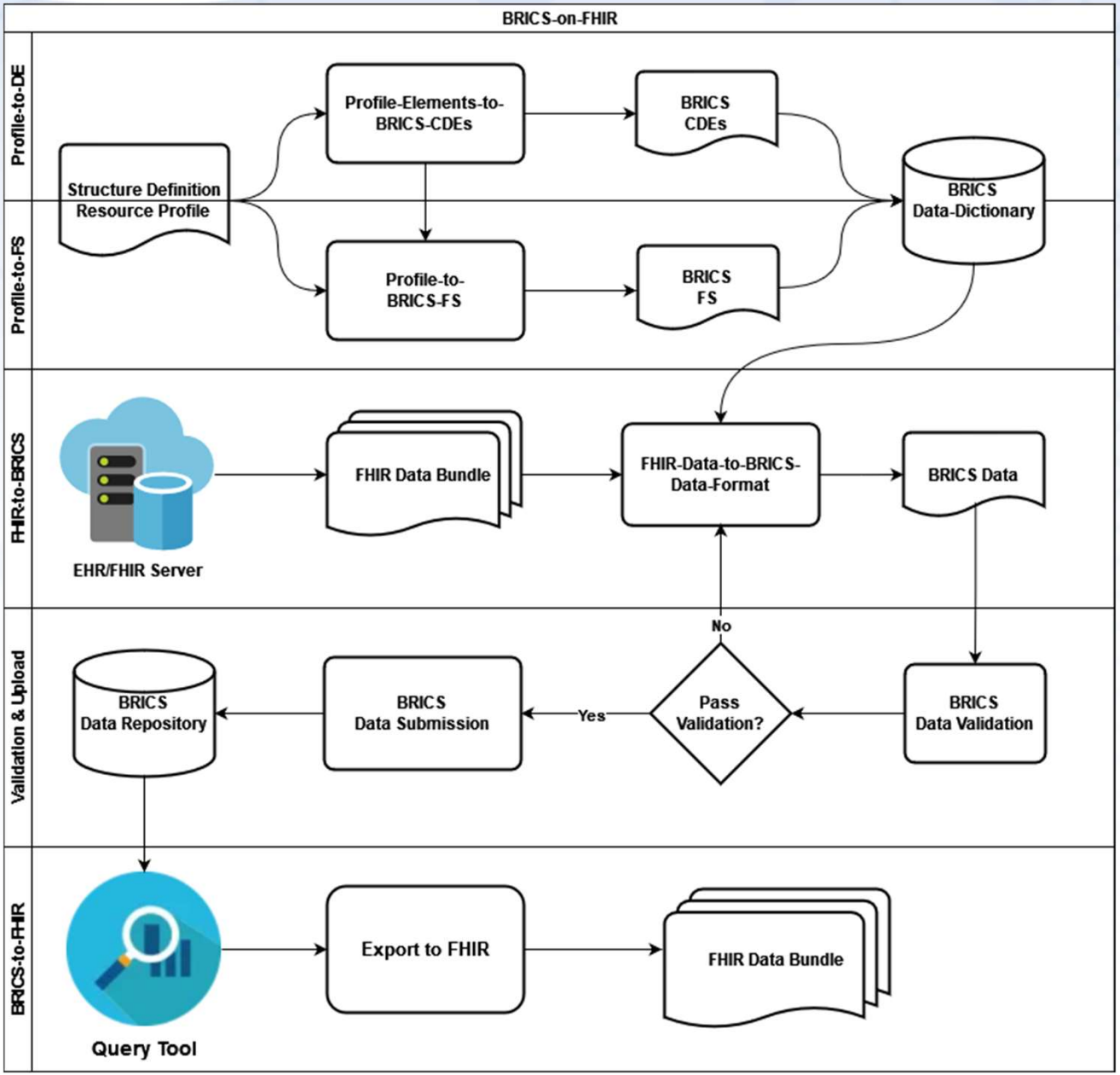
BRICS



Findable Accessible Interoperable Reusable



BRICS-on-FHIR: The Big Picture



US Core Profile – to – BRICS Form Structure

US Core Patient

BRICS Patient


```
{
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  "id" : "us-core-patient",
  "text" : {
    "status" : "extensions",
    "div" : "<div xmlns=\"http://www.w3.org/1999/xhtml\"><table bor
  },
  "url" : "http://hl7.org/fhir/us/core/StructureDefinition/us-core-
  "version" : "6.1.0",
  "name" : "USCorePatientProfile",
  "title" : "US Core Patient Profile",
  "status" : "active",
  "experimental" : false,
  "date" : "2022-09-30",
  "publisher" : "HL7 International - Cross-Group Projects",
  "contact" : [{
    "name" : "HL7 International - Cross-Group Projects",
    "telecom" : [{
      "system" : "url",
      "value" : "http://www.hl7.org/Special/committees/cgp"
    },
    {
      "system" : "email",
      "value" : "cgp@lists.HL7.org"
    }]
  }],
  "description" : "The US Core Patient Profile meets the U.S. Core
  "jurisdiction" : [{
    "coding" : [{
      "system" : "urn:iso:std:iso:3166",
      "code" : "US"
    }]
  }],
  "copyright" : "Used by permission of HL7 International, all right
  "fhirVersion" : "4.0.1"
```

```
{
    "shortName": "Patient",
    "title": "US Core Patient Profile",
    "description": "The US Core Patient Profile meets the U.S. Core
    "standardization": "Standard",
    "organization": "NIH/CIT/OSCS/BRICS",
    "required": false,
    "isCopyrighted": false,
    "diseases": "General (For all diseases)",
    "formType": "Clinical Assessment",
    "isCat": false,
    "repeatableGroups": [
        {
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            "repeatability": "EXACTLY 1",
            "dataElements": [
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                    "name": "GUID",
                    "requiredType": "REQUIRED"
                }
            ]
        },
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                    "requiredType": "RECOMMENDED"
                },
                {
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                    "requiredType": "RECOMMENDED"
                },
```

Demo

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Questions?

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BRICS Data Dictionary: mapping CDE semantics to Unified Medical Language System (UMLS) concepts

How it helps make BRICS data FAIR

Dr. Olga Vovk

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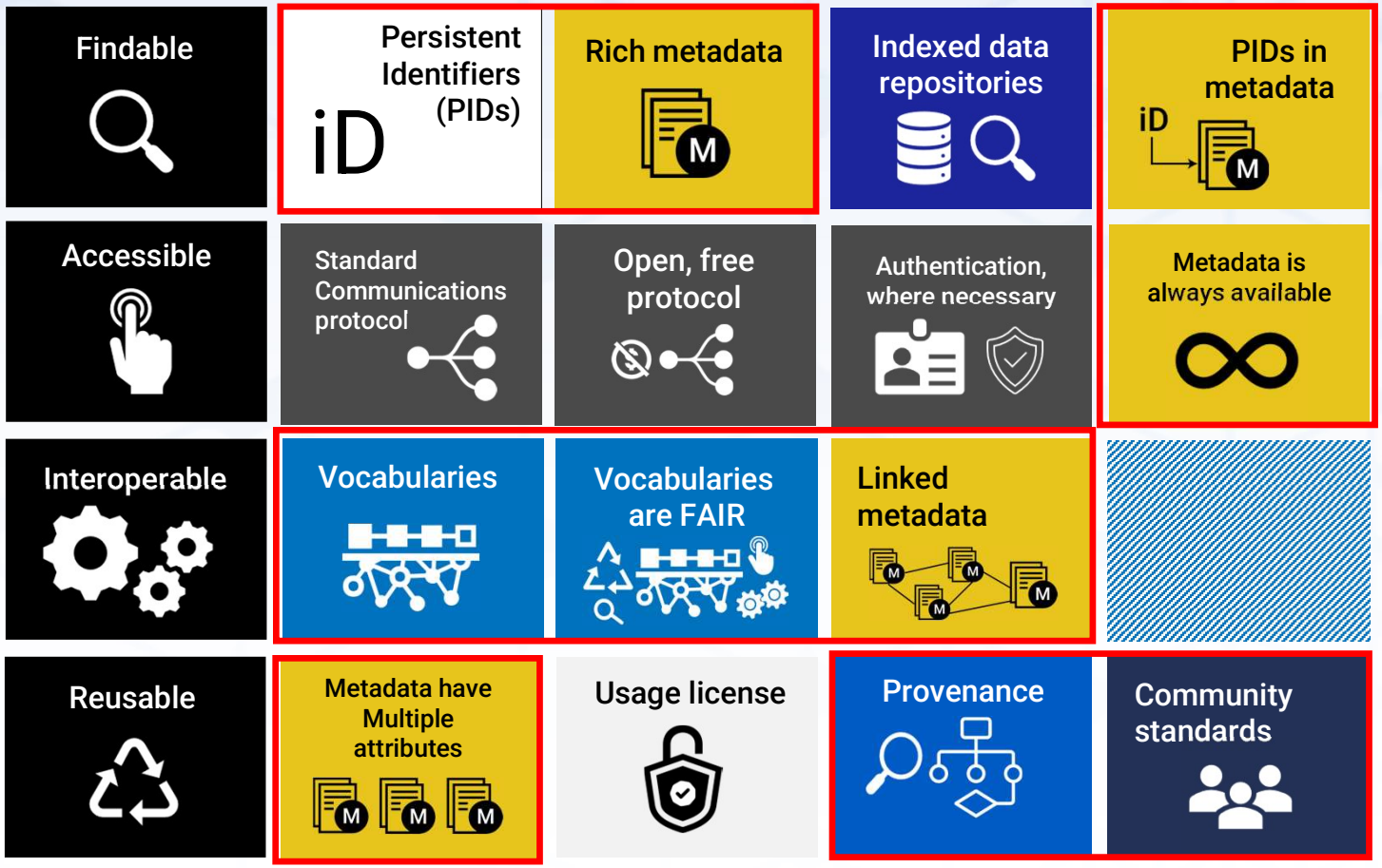
BRICS data dictionary is built on FAIR principles:

Findability, Accessibility, Interoperability, and Reusability (FAIR) principles



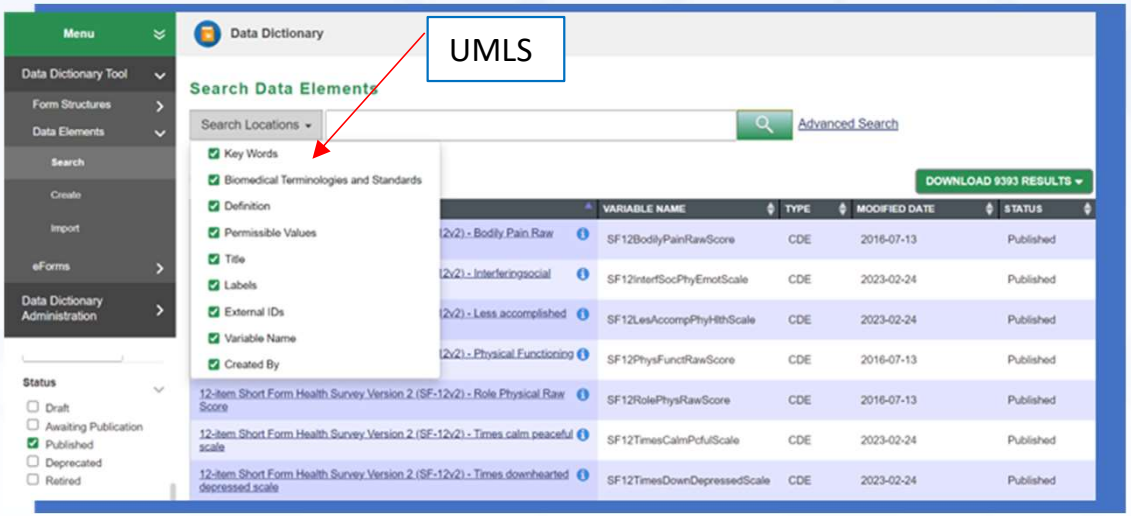
- Uses common data elements (CDEs) to collect data;
- Incorporates standard controlled vocabularies;

- Allows effectively find, query, and report data;
- Supports data exchange between independent informatics systems.

Advancing FAIR Data Principles

- The count of CDEs in BRICS DDs is big (~9,5 K published CDEs in FITBIR only) and growing.
- We support different common data models and standards.
- That affects how we manage data dictionaries, including searching, curating, creating, and the most important - re-using of CDEs.
- That affects data analysis and data discovery.

Issues common for all large CDE repositories



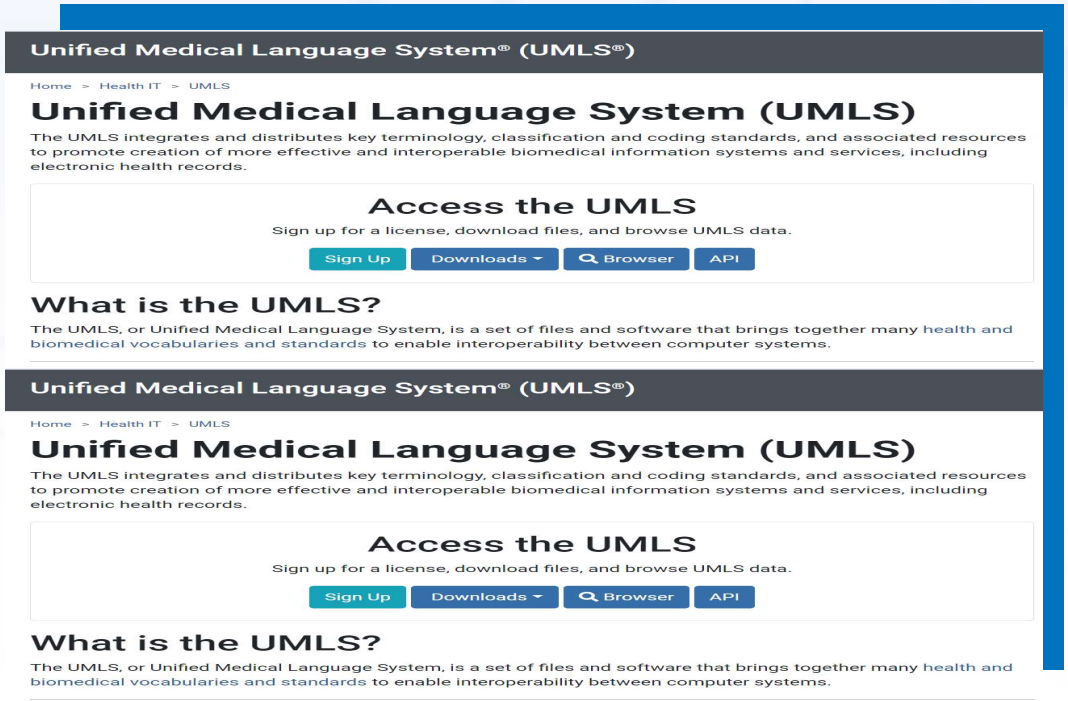
The most recent initiative we took to make BRICS to comply with the latest NIH Policy for Data Management and Sharing:

- is adding the option of organizing CDEs based on their semantics;
- by mapping DE semantics to UMLS concepts/CUIs.

Why map CDE semantics to UMLS?

UMLS as a terminology source

The Unified Medical Language System (UMLS) Metathesaurus, supported by NLM is a large biomedical thesaurus that is organized by concept, or meaning. It links synonymous names from over **200 different terminologies and vocabularies**.



- The Unified Medical Language System (UMLS) has been a critical tool in biomedical and health informatics for more than 30 years.
- The UMLS brings together vocabularies and standards in the biomedical field **to facilitate interoperability among different computer systems, projects**, and **applications**.

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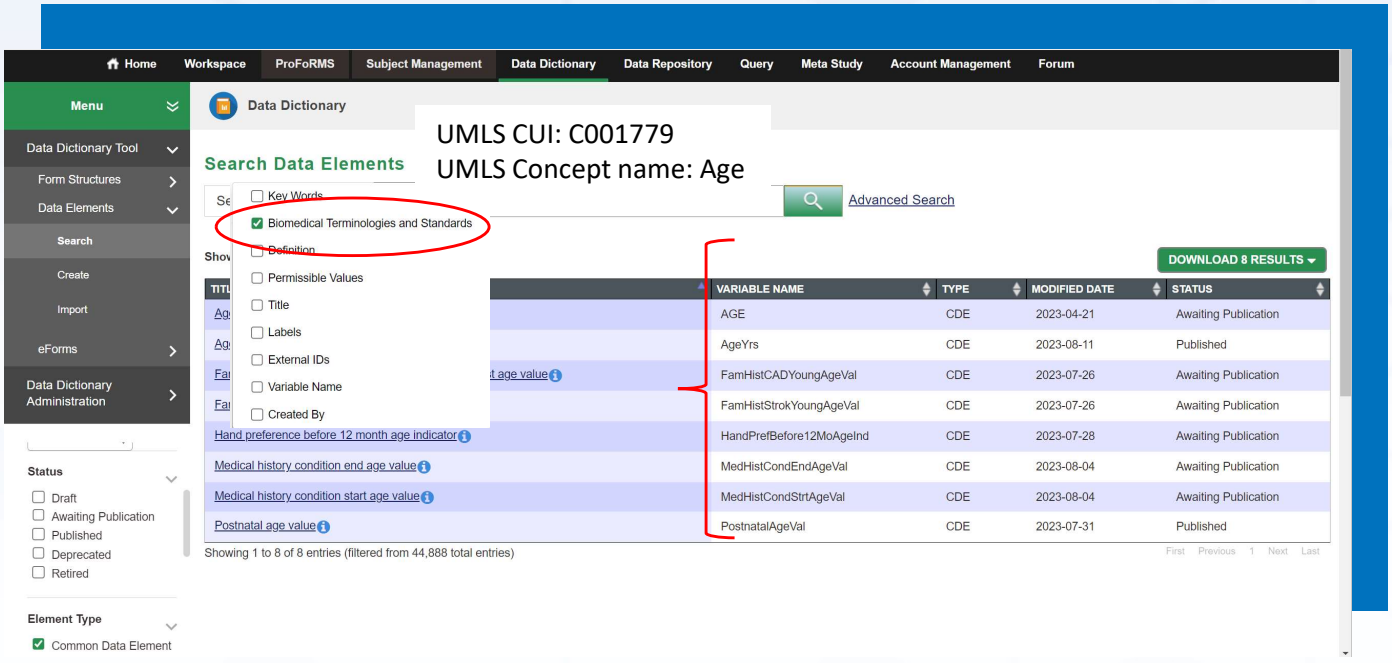
Annotating CDE semantics with UMLS concepts

- *Added UMLS coding per CDE, UDE and Permissible Values (3k DEs across BRICS)*
- *Added the ability to search for CDEs/UDEs by concept name and/or concept identifier*



- Supports programmatic analysis of the data.
- Makes data stored in a BRICS repositories:
  - more AI and machine learning ready;
  - in-line with both FAIR principles and the NIH Policy for Data Management and Sharing;
  - consistent with NLM's DE efforts supported by the NIH CDE Governance group.

We developed Data Element to UMLS Mapping Tool and mapped (across BRICS repositories) ~3K CDEs to UMLS concepts

- Supports data

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UMLS Tool Update

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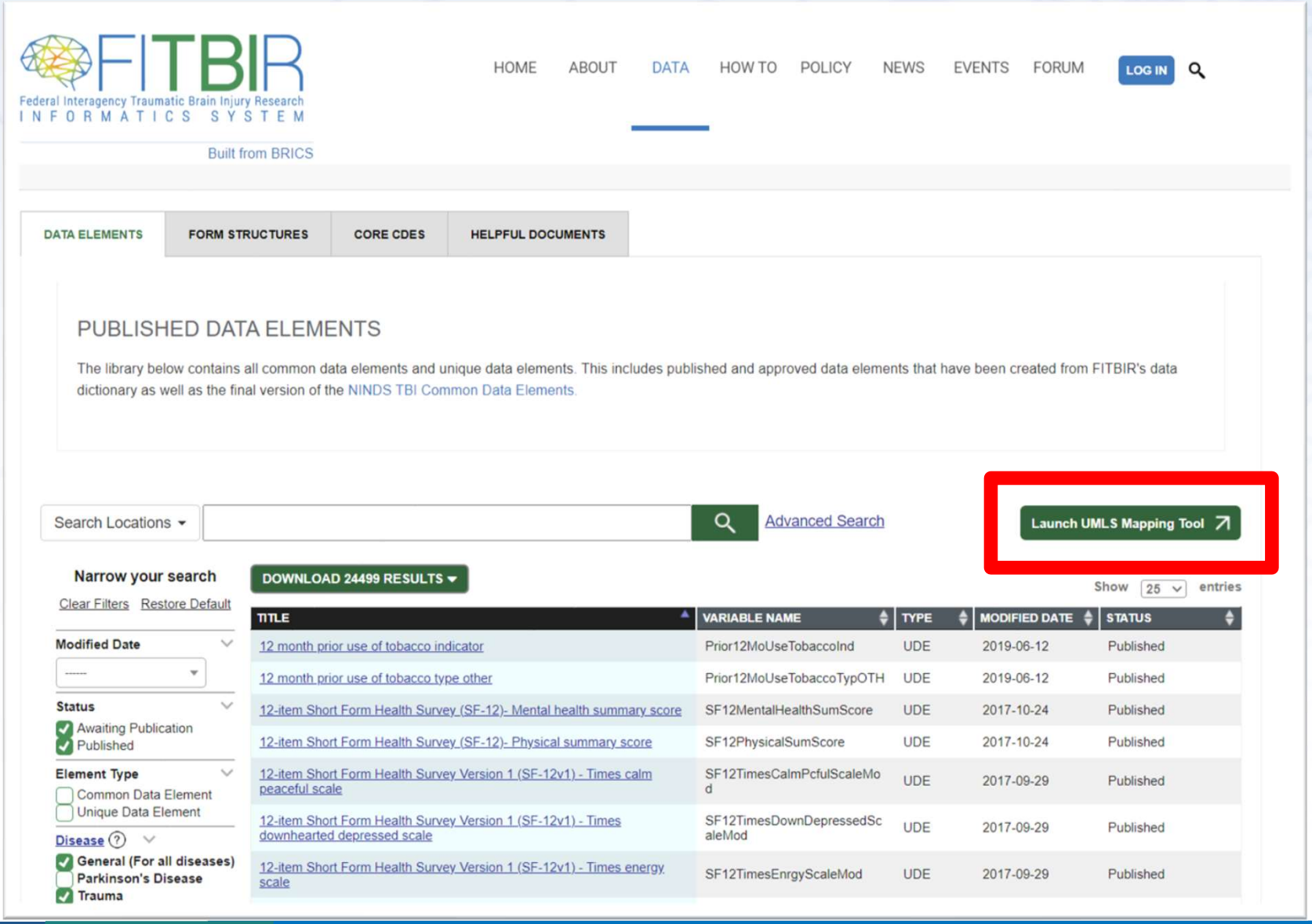
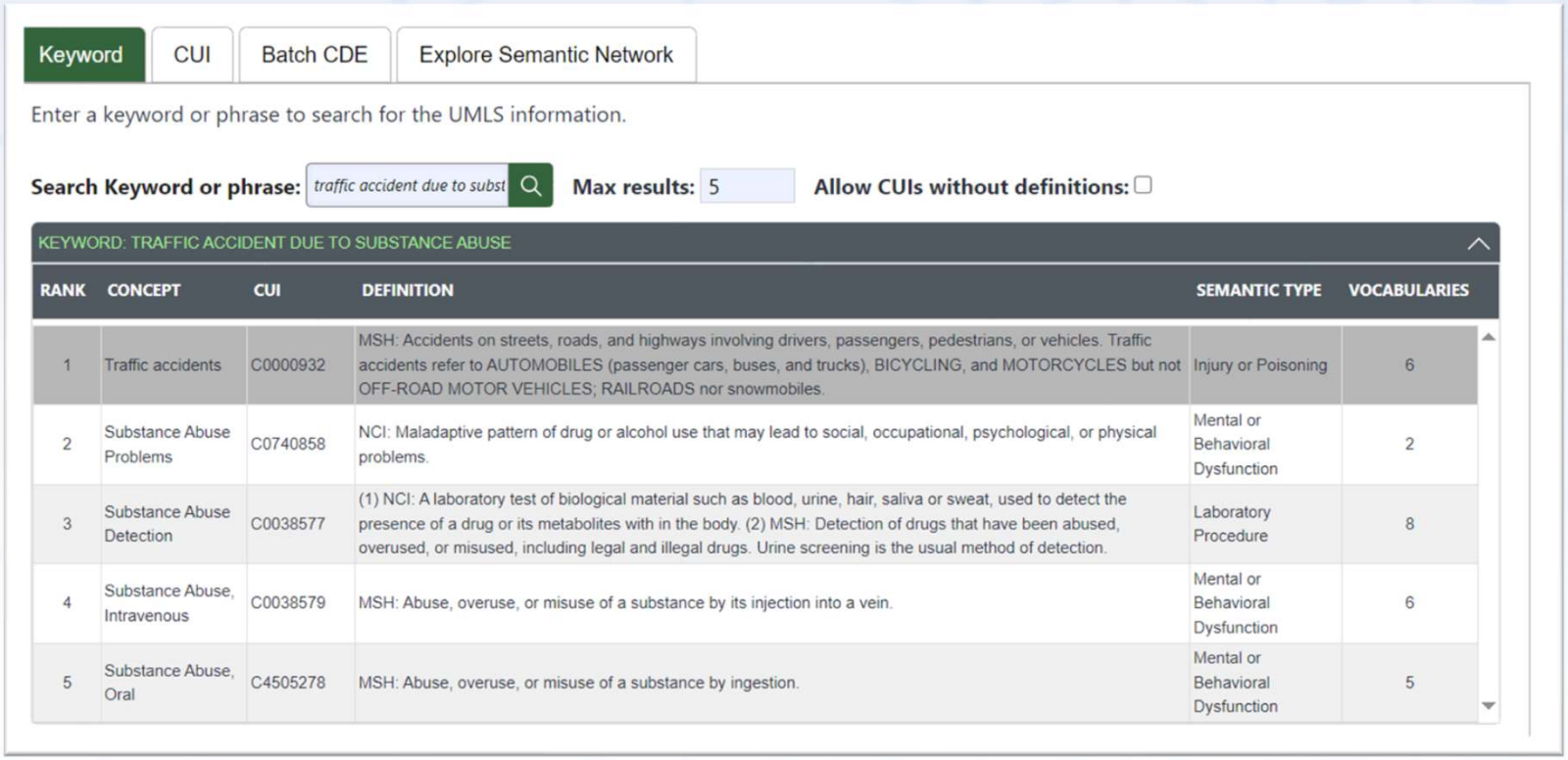
Data Element to UMLS Mapping Tool

FITBIR Data Dictionary: Launch the UMLS Mapping Tool (New!)



Data Element to UMLS Mapping Tool

Search phrase: “traffic accident due to substance abuse”



Keyword | CUI | Batch CDE | Explore Semantic Network

Enter a keyword or phrase to search for the UMLS information.

Search Keyword or phrase: traffic accident due to subst | Max results: 5 | Allow CUIs without definitions:

KEYWORD: TRAFFIC ACCIDENT DUE TO SUBSTANCE ABUSE

RANK	CONCEPT	CUI	DEFINITION	SEMANTIC TYPE	VOCABULARIES
1	Traffic accidents	C0000932	MSH: Accidents on streets, roads, and highways involving drivers, passengers, pedestrians, or vehicles. Traffic accidents refer to AUTOMOBILES (passenger cars, buses, and trucks), BICYCLING, and MOTORCYCLES but not OFF-ROAD MOTOR VEHICLES; RAILROADS nor snowmobiles.	Injury or Poisoning	6
2	Substance Abuse Problems	C0740858	NCI: Maladaptive pattern of drug or alcohol use that may lead to social, occupational, psychological, or physical problems.	Mental or Behavioral Dysfunction	2
3	Substance Abuse Detection	C0038577	(1) NCI: A laboratory test of biological material such as blood, urine, hair, saliva or sweat, used to detect the presence of a drug or its metabolites with in the body. (2) MSH: Detection of drugs that have been abused, overused, or misused, including legal and illegal drugs. Urine screening is the usual method of detection.	Laboratory Procedure	8
4	Substance Abuse, Intravenous	C0038579	MSH: Abuse, overuse, or misuse of a substance by its injection into a vein.	Mental or Behavioral Dysfunction	6
5	Substance Abuse, Oral	C4505278	MSH: Abuse, overuse, or misuse of a substance by ingestion.	Mental or Behavioral Dysfunction	5

Highlighted result:

Traffic accidents
C0000932

National Library of Medicine Unified Medical Language System (UMLS)

- **Metathesaurus**: contains over one million biomedical concepts from over 100 source vocabularies
- **Semantic Network**: defines 133 broad categories and fifty-four relationships between categories for labeling the biomedical domain

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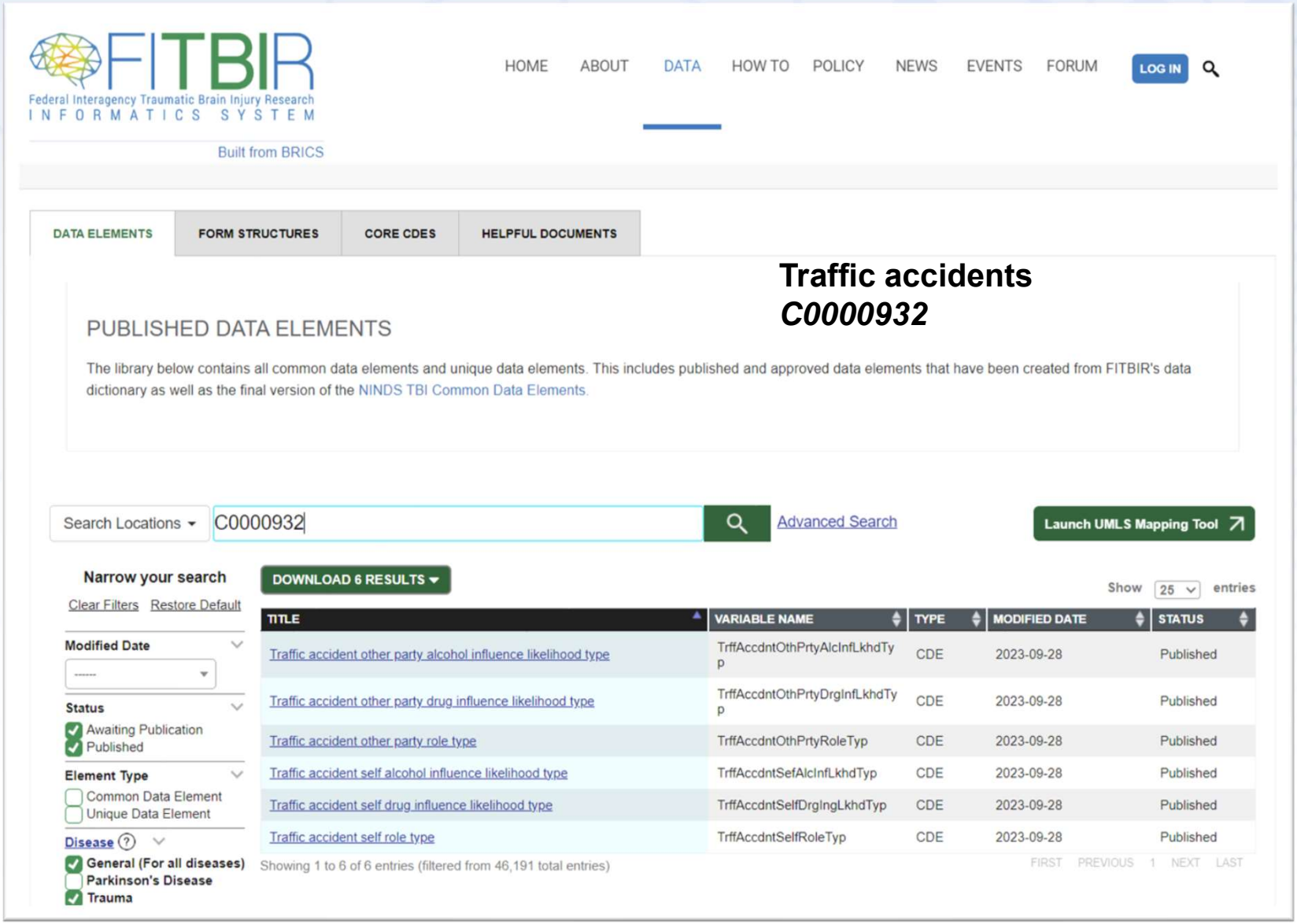
Data Element to UMLS Mapping Tool

Search Data Dictionary by UMLS CUI: C0000932



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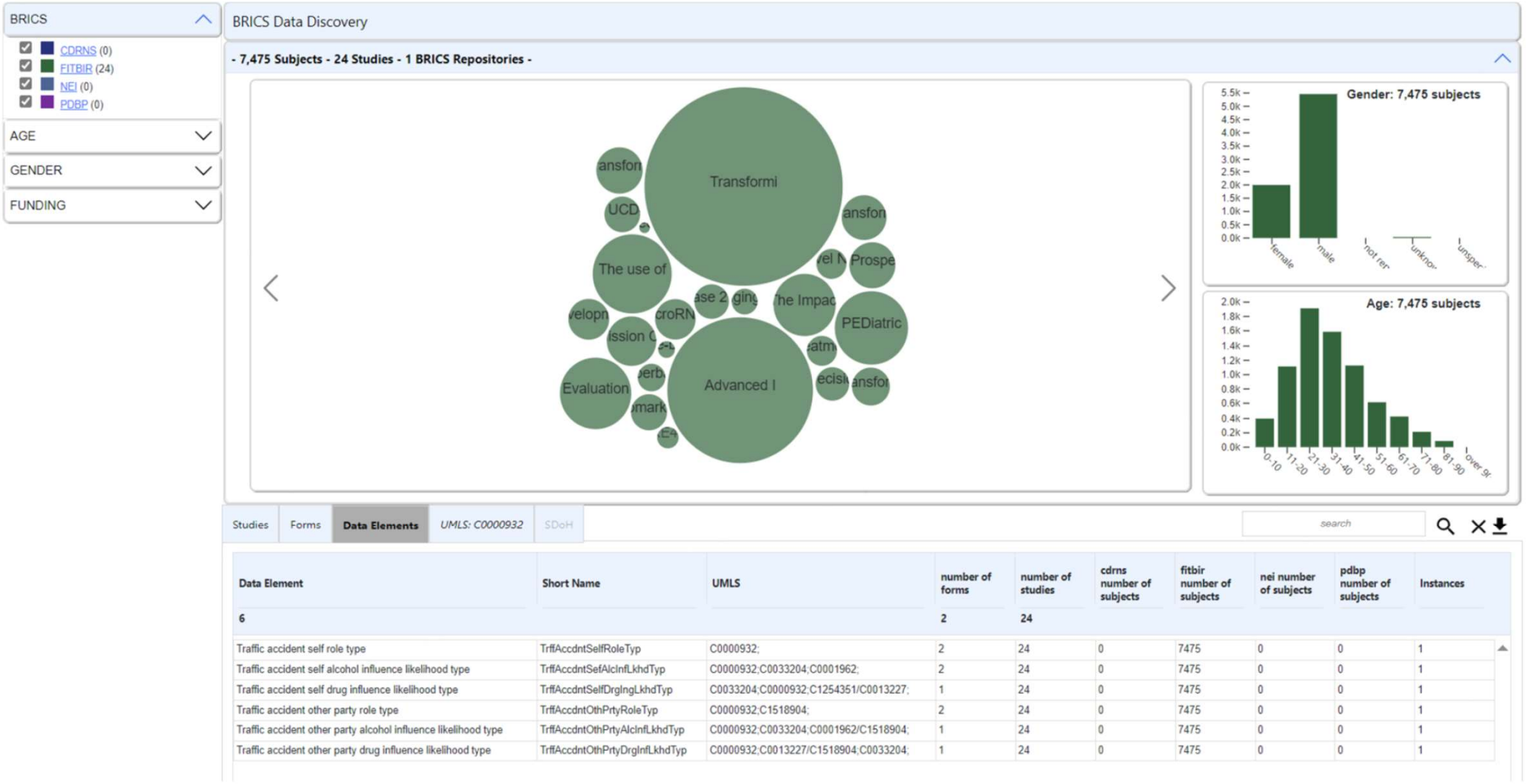
Data Element to UMLS Mapping Tool

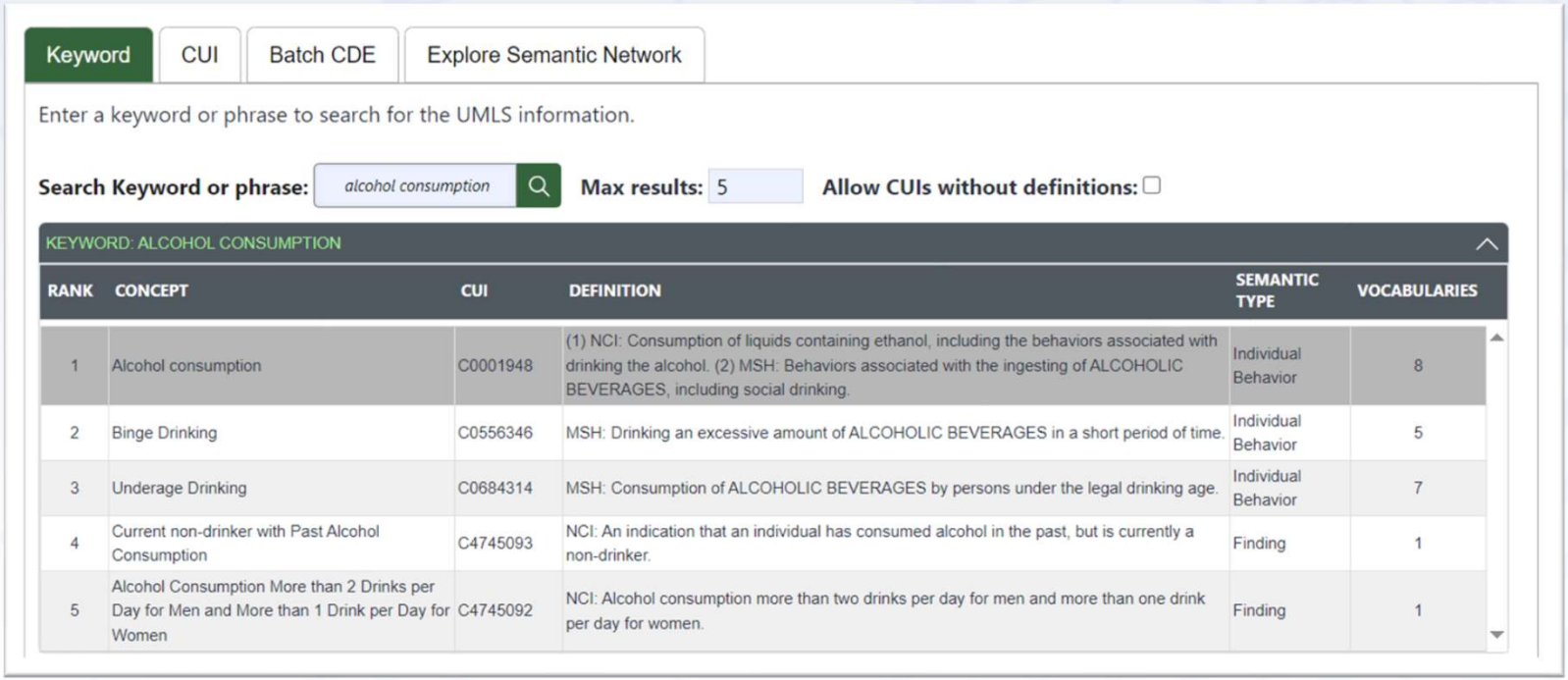
BRICS Data Discovery: Search by UMLS CUI: C0000932



Data Element	Short Name	UMLS	number of forms	number of studies	cdrns number of subjects	fitbir number of subjects	nei number of subjects	pdbp number of subjects	Instances
6			2	24					
Traffic accident self role type	TrffAccdntSelfRoleTyp	C0000932;	2	24	0	7475	0	0	1
Traffic accident self alcohol influence likelihood type	TrffAccdntSefAlcInfLkhdTyp	C0000932;C0033204;C0001962;	2	24	0	7475	0	0	1
Traffic accident self drug influence likelihood type	TrffAccdntSelfDrgIngLkhdTyp	C0033204;C0000932;C1254351/C0013227;	1	24	0	7475	0	0	1
Traffic accident other party role type	TrffAccdntOthPrtyRoleTyp	C0000932;C1518904;	2	24	0	7475	0	0	1
Traffic accident other party alcohol influence likelihood type	TrffAccdntOthPrtyAlcInfLkhdTyp	C0000932;C0033204;C0001962/C1518904;	1	24	0	7475	0	0	1
Traffic accident other party drug influence likelihood type	TrffAccdntOthPrtyDrgInfLkhdTyp	C0000932;C0013227/C1518904;C0033204;	1	24	0	7475	0	0	1

Data Element to UMLS Mapping Tool

Search phrase: “alcohol consumption”



RANK	CONCEPT	CUI	DEFINITION	SEMANTIC TYPE	VOCABULARIES
1	Alcohol consumption	C0001948	(1) NCI: Consumption of liquids containing ethanol, including the behaviors associated with drinking the alcohol. (2) MSH: Behaviors associated with the ingesting of ALCOHOLIC BEVERAGES, including social drinking.	Individual Behavior	8
2	Binge Drinking	C0556346	MSH: Drinking an excessive amount of ALCOHOLIC BEVERAGES in a short period of time.	Individual Behavior	5
3	Underage Drinking	C0684314	MSH: Consumption of ALCOHOLIC BEVERAGES by persons under the legal drinking age.	Individual Behavior	7
4	Current non-drinker with Past Alcohol Consumption	C4745093	NCI: An indication that an individual has consumed alcohol in the past, but is currently a non-drinker.	Finding	1
5	Alcohol Consumption More than 2 Drinks per Day for Men and More than 1 Drink per Day for Women	C4745092	NCI: Alcohol consumption more than two drinks per day for men and more than one drink per day for women.	Finding	1

Highlighted result:

Alcohol consumption
C0001948

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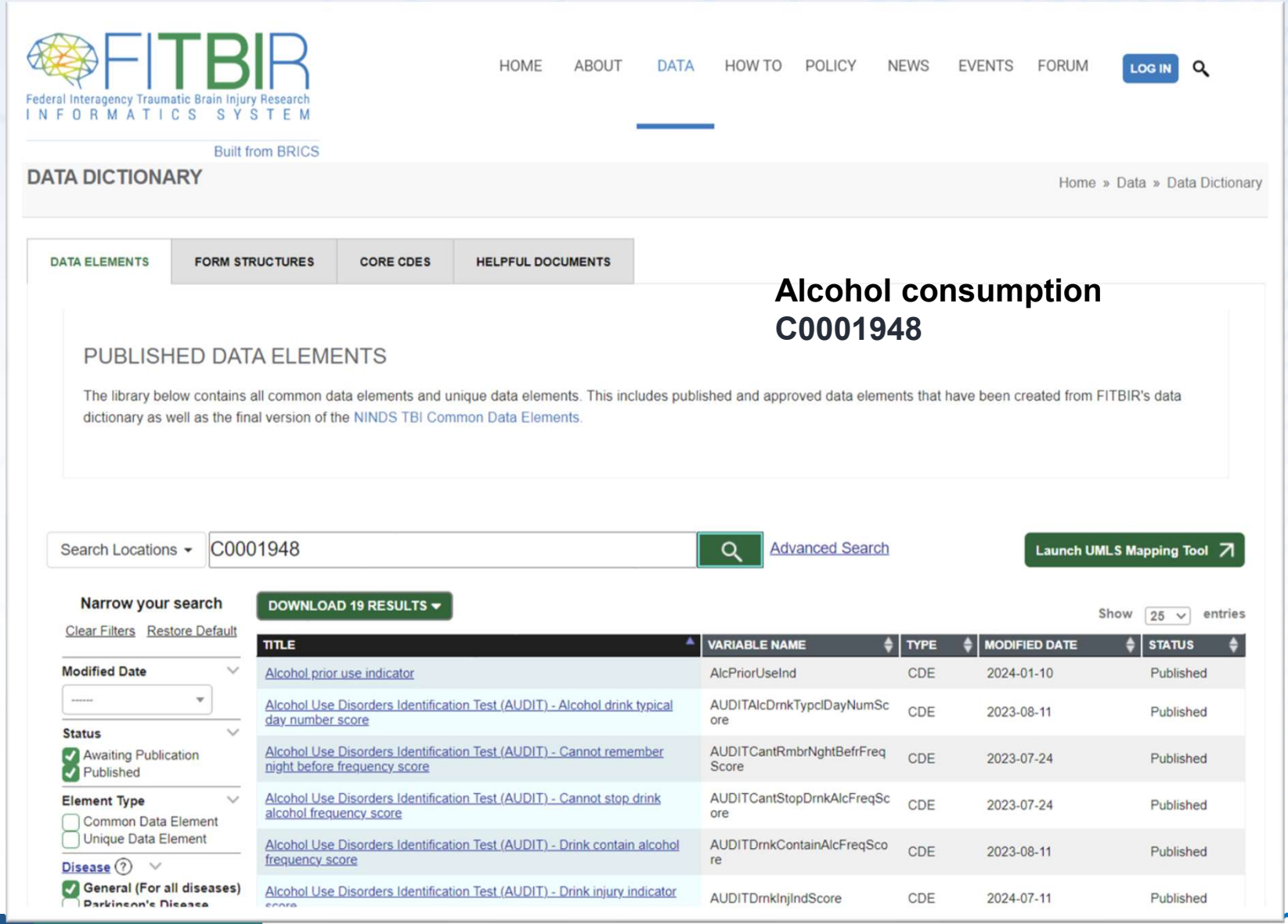
Data Element to UMLS Mapping Tool

Search Data Dictionary by UMLS CUI: C0001948



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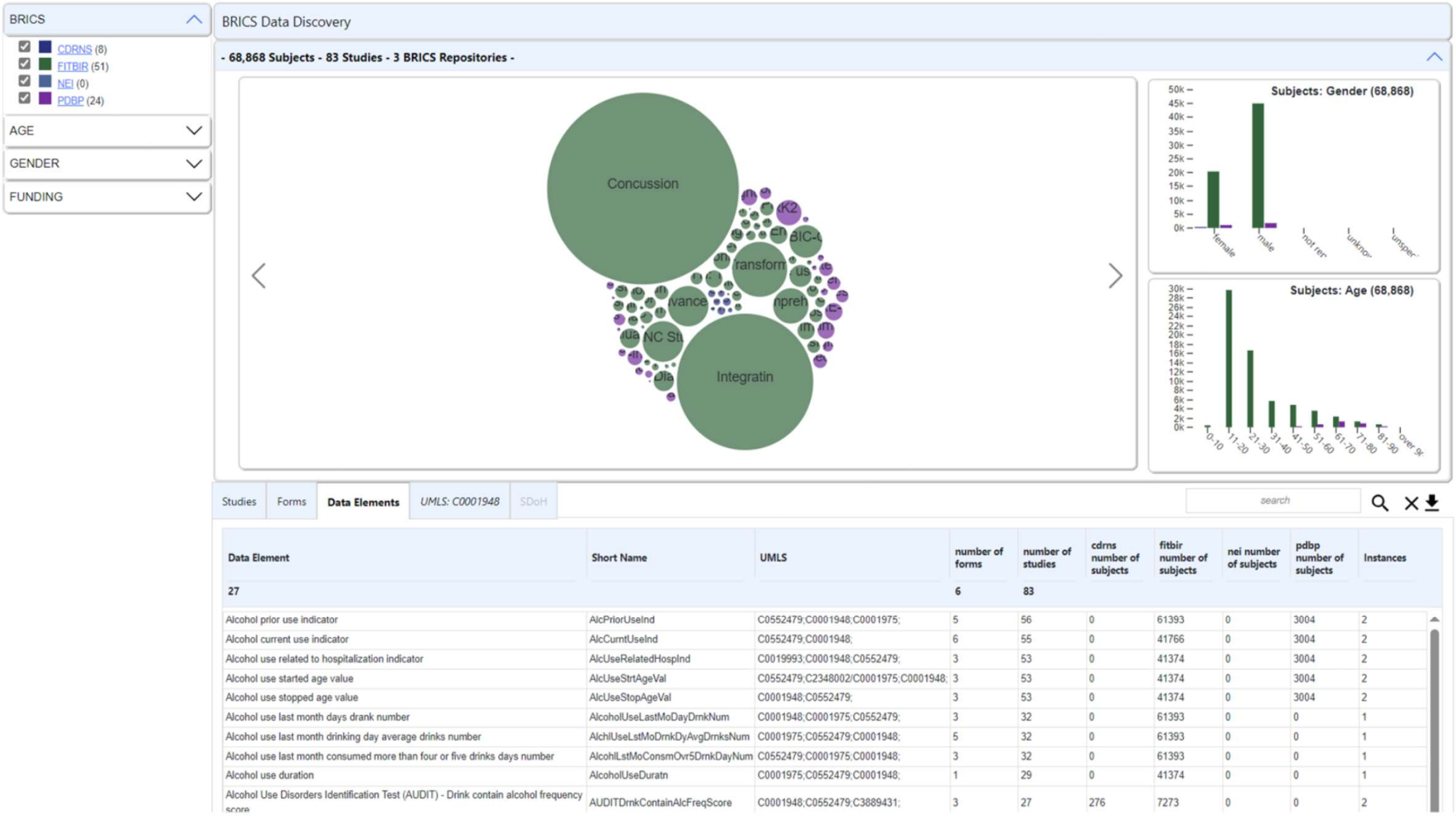
Data Element to UMLS Mapping Tool

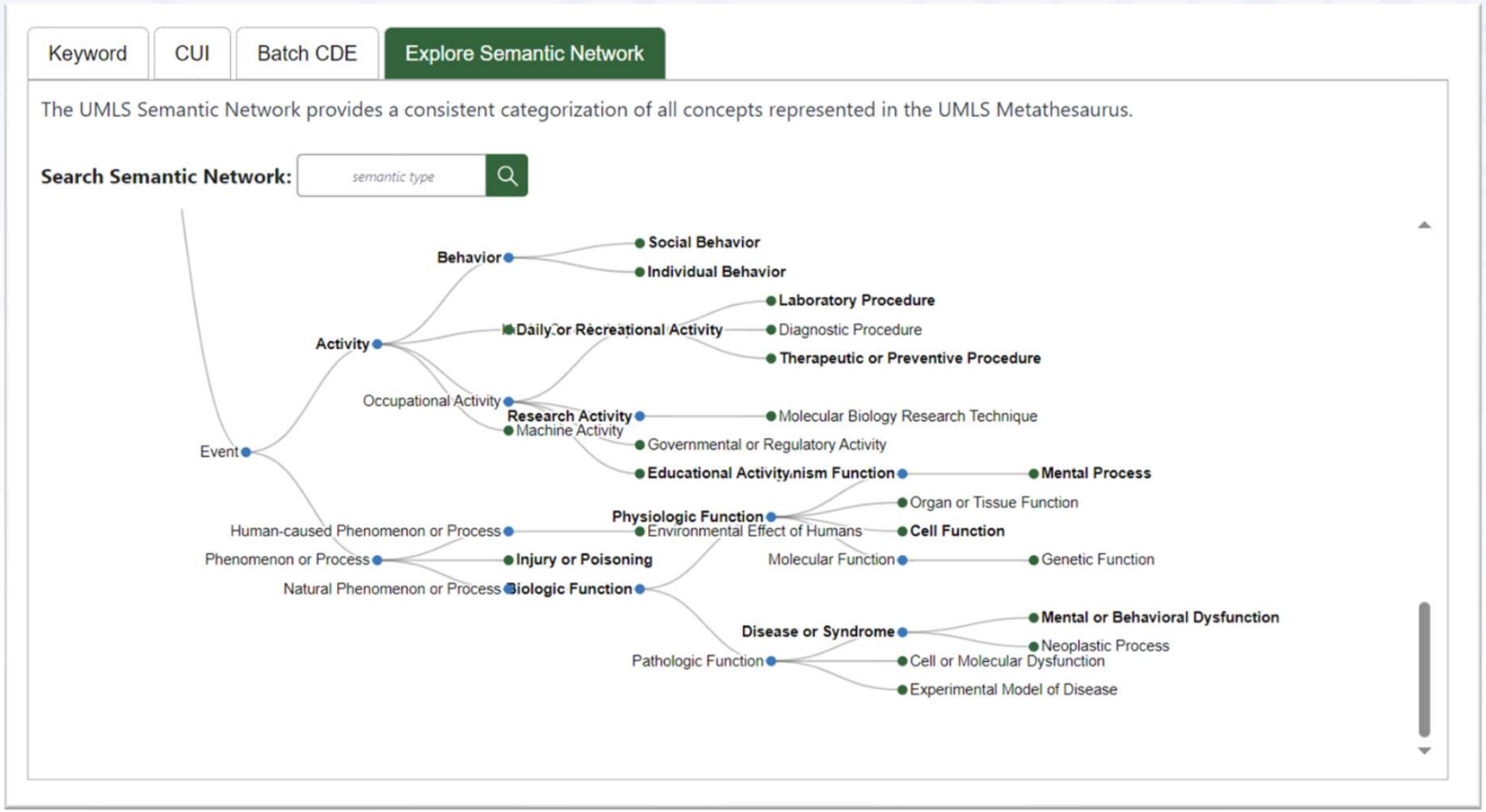
BRICS Data Discovery: Search UMLS by CUI: C0001948



Data Element	Short Name	UMLS	number of forms	number of studies	cdrns number of subjects	fitbir number of subjects	nei number of subjects	pdbp number of subjects	Instances
27			6	83					
Alcohol prior use indicator	AlcPriorUseInd	C0552479;C0001948;C0001975;	5	56	0	61393	0	3004	2
Alcohol current use indicator	AlcCurntUseInd	C0552479;C0001948;	6	55	0	41766	0	3004	2
Alcohol use related to hospitalization indicator	AlcUseRelatedHospInd	C0019993;C0001948;C0552479;	3	53	0	41374	0	3004	2
Alcohol use started age value	AlcUseStrtAgeVal	C0552479;C2348002/C0001975;C0001948;	3	53	0	41374	0	3004	2
Alcohol use stopped age value	AlcUseStopAgeVal	C0001948;C0552479;	3	53	0	41374	0	3004	2
Alcohol use last month days drank number	AlcoholUseLastMoDayDrnkNum	C0001948;C0001975;C0552479;	3	32	0	61393	0	0	1
Alcohol use last month drinking day average drinks number	AlchlUseLstMoDrnkDyAvgDrnksNum	C0001975;C0552479;C0001948;	5	32	0	61393	0	0	1
Alcohol use last month consumed more than four or five drinks days number	AlcohlLstMoConsmOvr5DrnkDayNum	C0552479;C0001975;C0001948;	3	32	0	61393	0	0	1
Alcohol use duration	AlcoholUseDuratn	C0001975;C0552479;C0001948;	1	29	0	41374	0	0	1
Alcohol Use Disorders Identification Test (AUDIT) - Drink contain alcohol frequency score	AUDITDrnkContainAlcFreqScore	C0001948;C0552479;C3889431;	3	27	276	7273	0	0	2

Data Element to UMLS Mapping Tool: What's New?

- Keyword (phrase) search
- Concept Unique Identifier (CUI) search
- Batch Data Element (.csv file)
  - **User selected search category**
- **Interactive Visualization of the UMLS Semantic Network**



Demo

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Questions?

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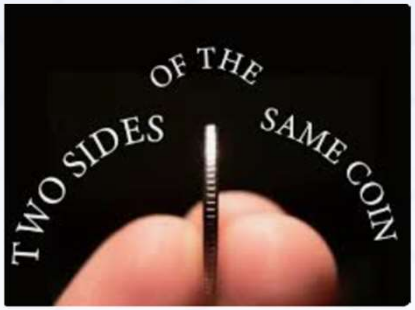
Avoiding the Data Element (DE) Dictionary Explosion: *Reducing Unintentional Redundancy*

BRICs Tool Development: Approaches to Facilitate Data Dictionary Searches

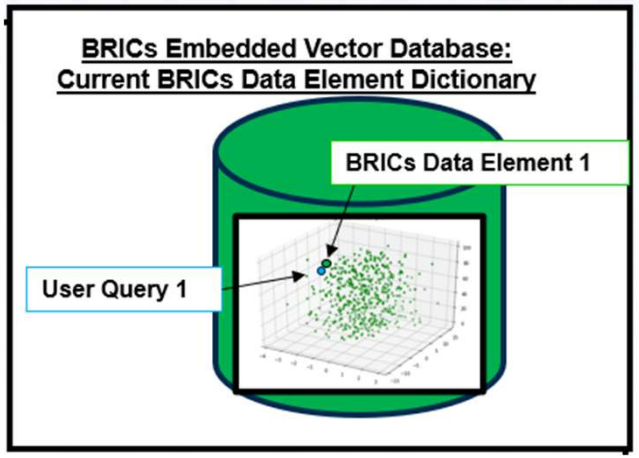


UMLS Code Assignments to Group Common Data Elements
(Standardized Concept Assignments to DEs Via Manual Curation)

UMLS Concept1:

BRICs Data Element 1
BRICs Data Element 2
BRICs Data Element 3…
Etc

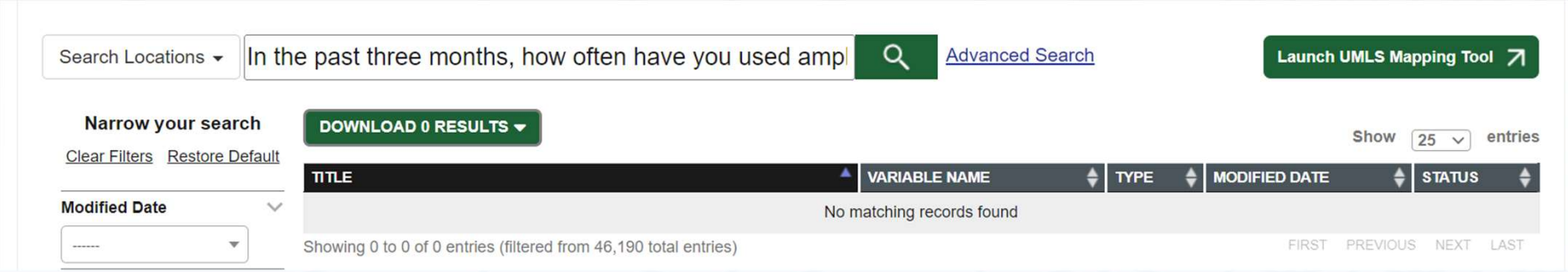
Semantic Similarity Searches to Group Common Data Elements
Pre-Trained Large Language Models transform DE text such that DEs with similar meaning cluster in embedding space



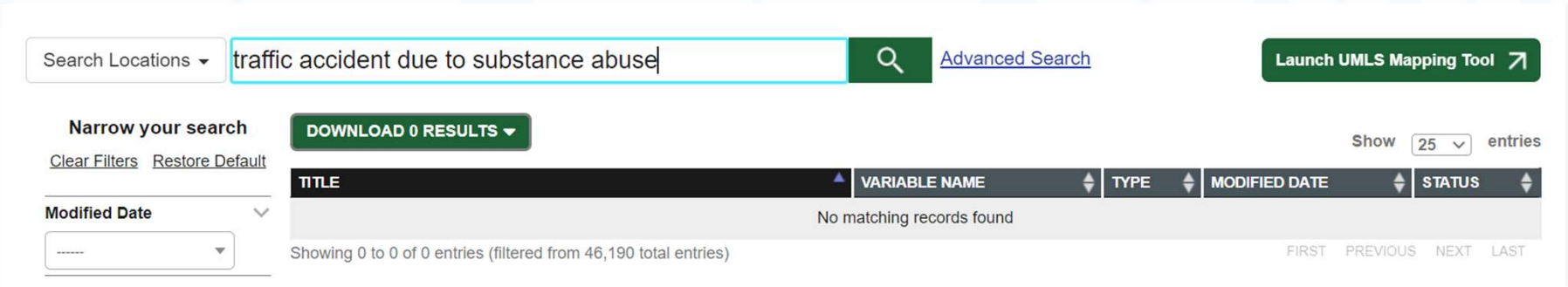
Limitations of Traditional Searches (Partial Matches Etc)

Perform poorly when:

I) Searching for matches to **complex text** (such as a Behavioral Question)



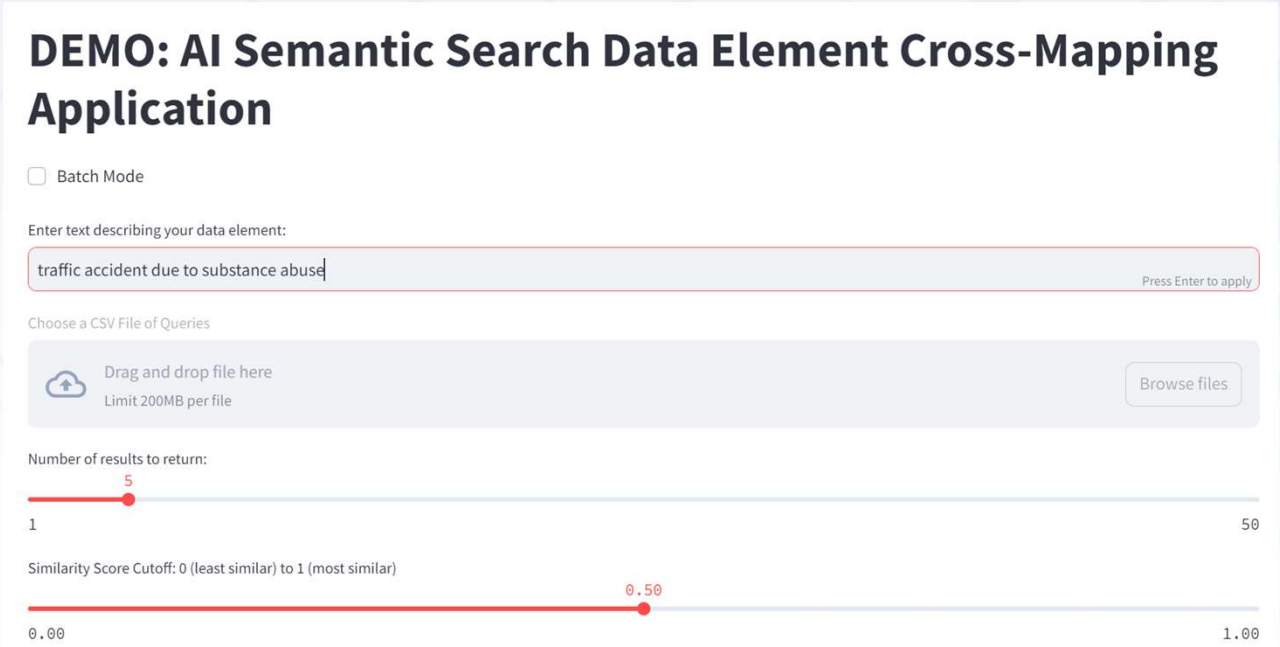
II) Searching for **semantically similar common data elements that aren't spelled the same, but are clearly related in their meaning**



https://fitbir.nih.gov/dictionary/dictionary/searchDataElementAction!list.action

Solution: Semantic Similarity Searches Using Pre-Trained Large Language (LLM) Models:

- **Completed an Internal Proof of Concept 'AI' Tool for Searching the FITBIR Dictionary (Currently NIH Access Only) – Demo Today**



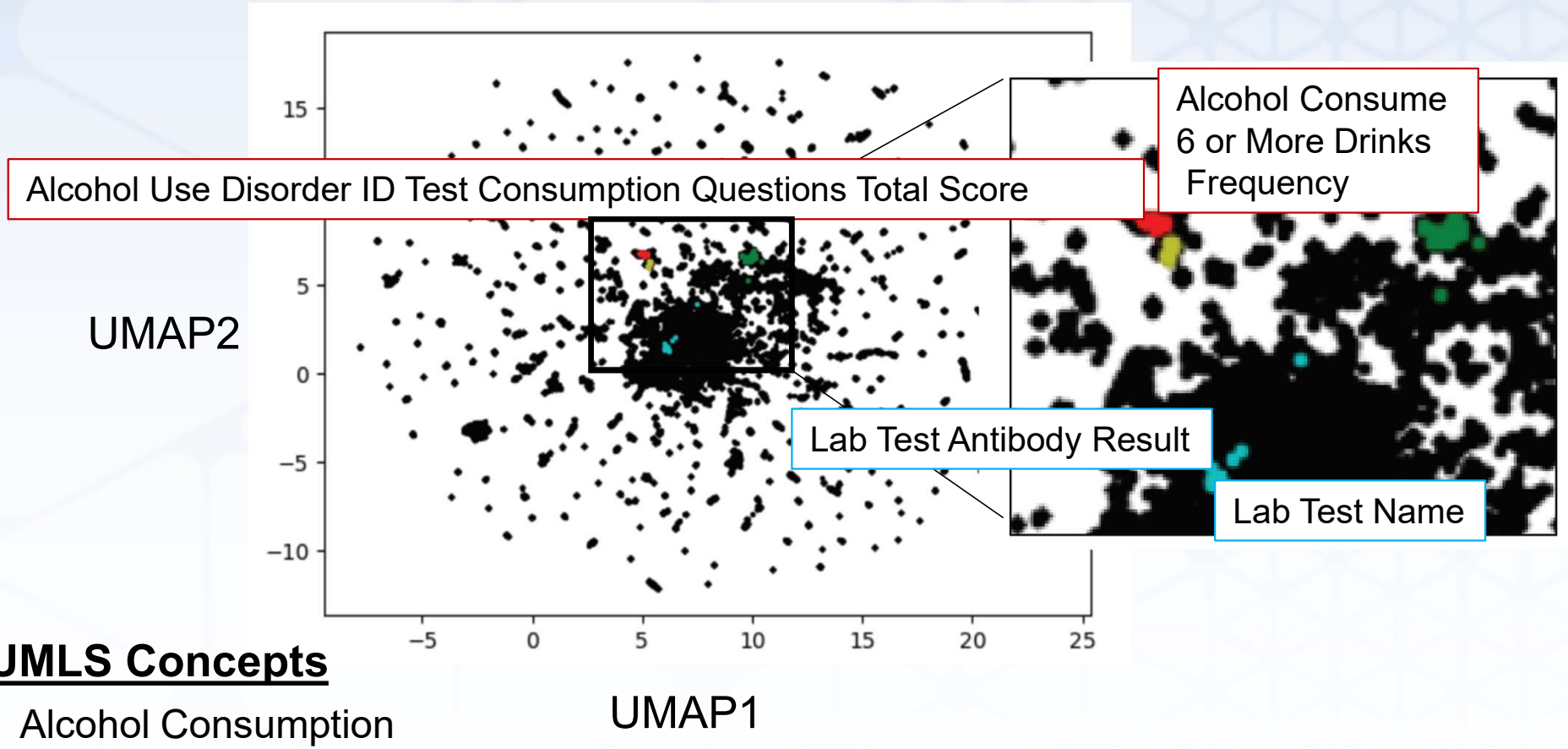
http://spencer.cit.nih.gov:8501/

- ***In Progress: productionizing AI tool for release/integration on the public FITBIR data dictionary website (Coming Soon)***

Demo

Human Vetted UMLS DE Concept Labels Can Be Used to Confirm LLM Performance

LLM Embedding Space: FITBIR Data Dictionary (Vector = 768)



UMLS Concepts

- Alcohol Consumption
- Illicit Drugs
- Laboratory Procedures
- Headache

Future Goals: Make Common Data Elements Easy to Search/Harmonize

So We Can Focus on Scientific Discovery!

- **Complete Testing and Validation Internally of Demo**
- **Integrate additional filters on:**
  - **Form (Collection of Common Data Elements)**
  - **Permissible Values**
  - **UMLS Concepts**

  (Note: semantic searches can be integrated with traditional search methods to get the best of both worlds)
- **Optimize data element cross-mapping work-flow: Example- Integrate AI Semantic Search into Previous Data Mapping and Transformation Tool**
- **Public Release of FITBIR AI Semantic Search Data Element Cross-Mapping Application –** *other instances possibly to follow*
- **Potentially Facilitate other Data Dictionary Harmonization Efforts (preclinical vs clinical TBI etc.)**
- **Development of Additional AI tools to Facilitate Efficient Data Discovery/Compilation Across Multiple Studies.**

Questions/Comments/Collaborations: Contact maria.bagonis@nih.gov

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Questions?

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Thank you for joining in!

Will see you all on November 14th, 2024!