

BRICS Seminar

FHIR Data Element Mapping Tool UMLS in BRICS Data Dictionary - why UMLS? UMLS Tool Update Al Search on Data Elements

Biomedical Research Informatics Computing System (BRICS) October 10th, 2024













NATIONAL INSTITUTE OF NEUROLOGICAL DISORDERS AND STROKE





Logistics

Audio/Video	Please keep your microphone muted						
Recording	 Today's session will be recorded Will be posted on the BRICS website: <u>https://brics.cit.nih.gov/demo</u> 						
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 <u>after each speaker</u> and at the <u>end</u> of the demo to ask live questions. 						









Agenda

Time	Торіс	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









Biomedical Research Informatics Computing System (BRICS)

FHIR Data Element Mapping Tool

Dr. Henry Ogoe











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BRICS-on-FHIR





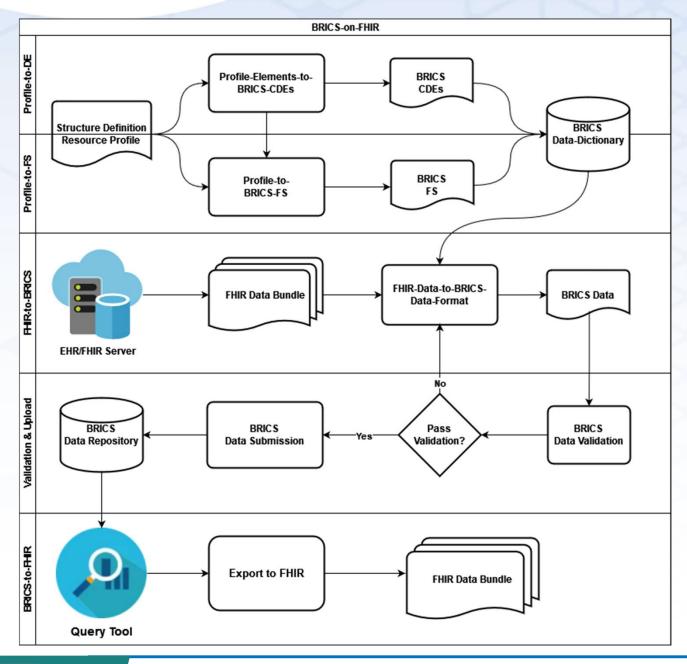
NIH



Learn more about BRICS at: https://brics.cit.nih.gov



BRICS-on-FHIR: The Big Picture



NIH National Institutes of Health Center for Information Technology



Learn more about BRICS at: https://brics.cit.nih.gov



US Core Profile – to – BRICS Form Structure

US Core Patient

```
"resourceType" : "StructureDefinition",
"id" : "us-core-patient",
 "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",
    "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
```

BRICS Patient

```
"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": [
        "name": "Main",
        "repeatability": "EXACTLY 1",
        "dataElements": [
                 "name": "GUID",
                 "requiredType": "REQUIRED"
        "name": "Patient 01",
        "repeatability": "LESSTHAN 2",
        "dataElements": [
                 "name": "PatientId",
                 "requiredType": "RECOMMENDED"
                 "name": "PatientMeta",
                 "requiredType": "RECOMMENDED"
```











Questions?

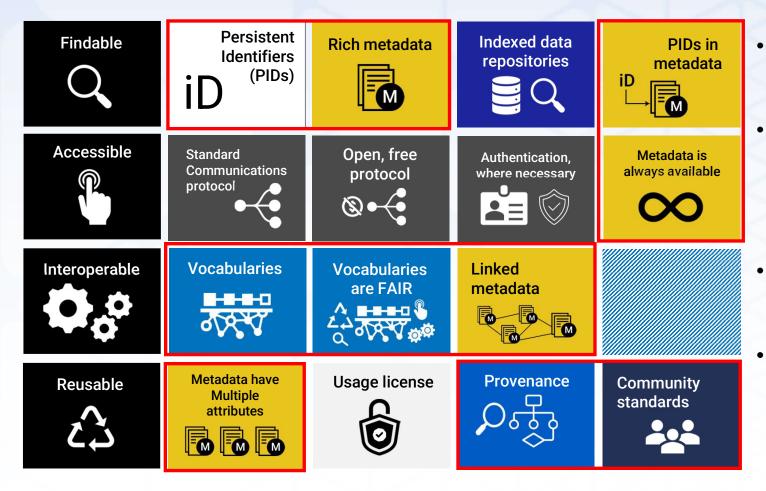


BRICS Data Dictionary: mapping CDE semantics to Unified Medical Language System (UMLS) concepts How it helps make BRICS data FAIR Dr. Olga Vovk



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.

Issues common for all large CDE repositories

Data Dictionary UMLS Data Dictionary Too Search Data Element Advanced Search Search Locations . Key Words Biomedical Terminologies and Standards Definition Permissible Values (2v2) - Bodily Pain Raw () SF 12Bodily Pain RawScore CDE 2016-07-13 Published Title eForm 2v2) - Interferingsocial 0 SF12InterfSocPhyEmotScale CDE 2023-02-24 Published Z Labels Data Dictionar (2v2) - Less accomplished () SF12LesAccompPhyHithScale External IDs 2023-02-24 CDE Published Variable Name (2v2) - Physical Euroctioning () SF12PhysFunctRawScore CDE 2016-07-13 Published Created By Status 12-item Short Form Health Survey Version 2 (SF-12v2) - Role Physical Raw () SF 12RolePhysRawScore CDE 2016-07-13 Published C Draft Awaiting Publication 12-Item Short Form Health Survey Version 2 (SF-12v2) - Times calm peaceful () SF12TimesCalmPcfulScal CDE 2023-02-24 Published Published Decrecate 12-Item Short Form Health Survey Version 2 (SF-12v2) - Times downhearted 0 SF12TimesDownDepres 2023-02-24 Published

That affects data analysis and data discovery.

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

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



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Why map CDE semantics to UMLS?

UMLS as a terminology source

<u>The Unified Medical Language System (UMLS) Metathesaurus</u>, 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**.

Unified Medical Lan	guage System [®] (UMLS [®])
Home > Health IT > UMLS	
Unified Medie	cal Language System (UMLS)
	utes key terminology, classification and coding standards, and associated resources ective and interoperable biomedical information systems and services, including
	Access the UMLS
Sigr	n up for a license, download files, and browse UMLS data.
	Sign Up Downloads - Q Browser API
What is the UM	ILS?
	anguage System, is a set of files and software that brings together many health and andards to enable interoperability between computer systems.
Unified Medical Lan	guage System® (UMLS®)
Home > Health IT > UMLS	
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Unified Media The UMLS Integrates and distribute to promote creation of more effected electronic health records.	Access the UMLS data.

- 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.









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

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Awaiting Publication Published	1	Postnat	tal age value			F	PostnatalAge	Val	CDE	2023-07-31	Published

Element Type

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

- 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 <u>NIH CDE</u> <u>Governance group</u>.







Biomedical Research Informatics Computing System (BRICS)

UMLS Tool Update

Dr. Alexandra Bokinsky











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Data Element to UMLS Mapping Tool

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

	Injury Research Y S T E M		HOME	ABOUT	DATA	HOW TO	POLICY	NEWS	EVENTS	FORUM	LOG IN	۹
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Search phrase: "traffic accident due to substance abuse"

Keywo	ord CUI	Batch CI	DE Explore Semantic Network		
nter a	keyword or ph	rase to sea	rch for the UMLS information.		
earch	Keyword or p	hrase: traff	ic accident due to subst Q Max results: 5 Allow CUIs without definitions:		
KEYWC	RD: TRAFFIC ACCI	DENT DUE TO	D 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 OFF-ROAD MOTOR VEHICLES; RAILROADS nor snowmobiles.	not 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









Search Data Dictionary by UMLS CUI: C0000932

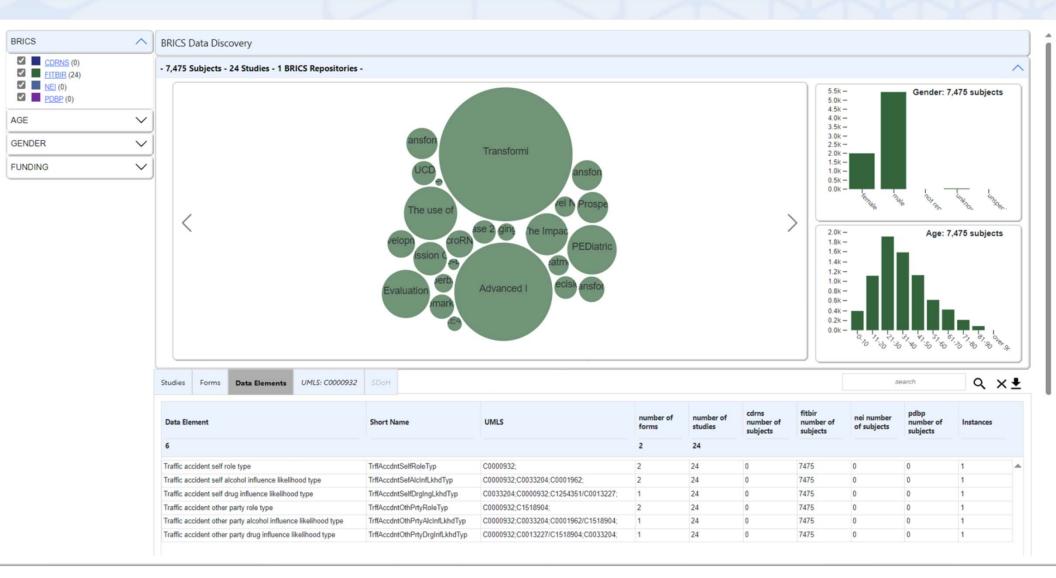
Interagency Traumati		R ry Research S T E M		HOME	ABOUT	DATA	ноw то	POLICY	NEWS	EVENTS	FORUM	LOG IN	٩
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BRICS Data Discovery: Search by UMLS CUI: C0000932











Search phrase: "alcohol consumption"

Keyw	ord CUI Batch CDE Ex	plo <mark>re Sem</mark> a	antic Network		
Enter a	keyword or phrase to search for the	UMLS info	rmation.		
Search	Keyword or phrase: alcohol consum	ption Q	Max results: 5 Allow CUIs without definitions:		
KEYWO	ORD: 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

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







Search Data Dictionary by UMLS CUI: C0001948

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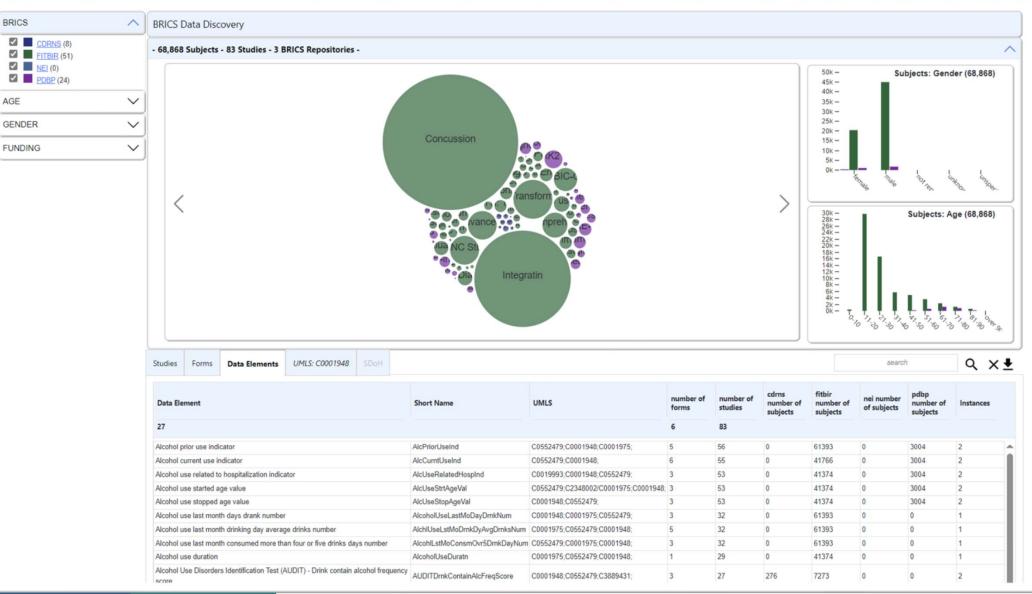


Center for Information Technology





BRICS Data Discovery: Search UMLS by CUI: C0001948









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

Keyword	CUI	Batch CDE	Explore Semantic Network	
The UMLS Se	emantic N	letwork provides	a consistent categorization of all concepts represented in the UMLS Metathesaurus.	
Search Sema	antic Net	twork: seme	antic type Q	
			Social Behavior	
			Behavior Individual Behavior	
			Laboratory Procedure	
			Daily or Recreational Activity	
		Activity	Therapeutic or Preventive Procedure	
	Even		Cupational Activity Research Activity Machine Activity Governmental or Regulatory Activity	
			Educational Activity.nism Function Mental Process	
	•	Human-caused Pheno		
	Pher	nomenon or Process	Injury or Poisoning Molecular Function Genetic Function	
		Natural Pheno	menon or Process Siologic Function	
			Disease or Syndrome Mental or Behavioral Dysfunction Neoplastic Process Pathologic Function Cell or Molecular Dysfunction	
			Experimental Model of Disease	ļ











Questions?



Biomedical Research Informatics Computing System (BRICS)

Al Search for Data Dictionary

Dr. Maria Bagonis











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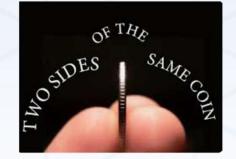




Avoiding the Data Element (DE) Dictionary Explosion: Reducing Unintentional Redundancy



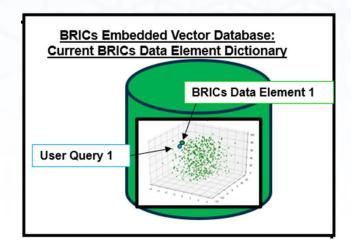
BRICs Tool Development: Approaches to Facilitate Data Dictionary Searches



UMLS Code Assignments to Group Common Data Elements

(Standardized Concept Assignments to DEs Via Manual Curation)

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



UMLS Concept1:

BRICs Data Element 1 BRICs Data Element 2 BRICs Data Element 3... Etc

National

Institutes

of Health

NIH







Limitations of Traditional Searches (Partial Matches Etc)

Perform poorly when:

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

Search Locations - In th		the	ne past three months, how often have you used amp		Q Advanced Search			Launch	ing Too	ol ⊿	
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Modified Date		\sim	No	matching	records found						
	•		Showing 0 to 0 of 0 entries (filtered from 46,190 total entries)					FIRST	PREVIOUS	NEXT	LAST

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

Search Locations - traffi	c accident due to substance abuse	Q Advanced Search	Launch UMLS Mapping Tool 7				
Narrow your search Clear Filters Restore Default	DOWNLOAD 0 RESULTS -		Show 25 v entries				
	ΠΠΕ	🔺 VARIABLE NAME 🛛 🌲 TYPE	♦ MODIFIED DATE				
Modified Date		No matching records found					
	Showing 0 to 0 of 0 entries (filtered from 46,190 total entries)		FIRST PREVIOUS NEXT LAST				

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

DEMO: AI Semantic Search Data Element Cross-Mapping Application

traffic accident due to substance abuse	Press Enter to ap
hoose a CSV File of Queries	
Drag and drop file here Limit 200MB per file	Browse files
umber of results to return: 5	
•	
milarity Score Cutoff: 0 (least similar) to 1 (most similar)	
.00	1.

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



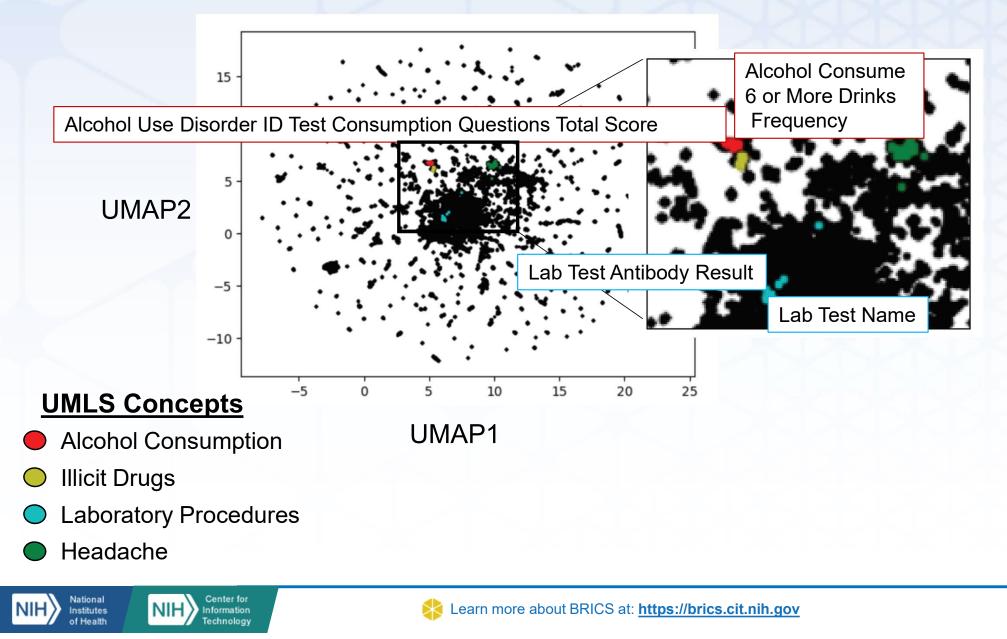






Human Vetted UMLS DE Concept Labels Can Be Used to Confirm LLM Performance

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





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 <u>Data Mapping and Transformation Tool</u>
- 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 <u>maria.bagonis@nih.gov</u>









Questions?



Thank you for joining in!

Will see you all on November 14th, 2024!

