Effect of (OHDSI) Vocabulary Mapping on Phenotype Cohorts

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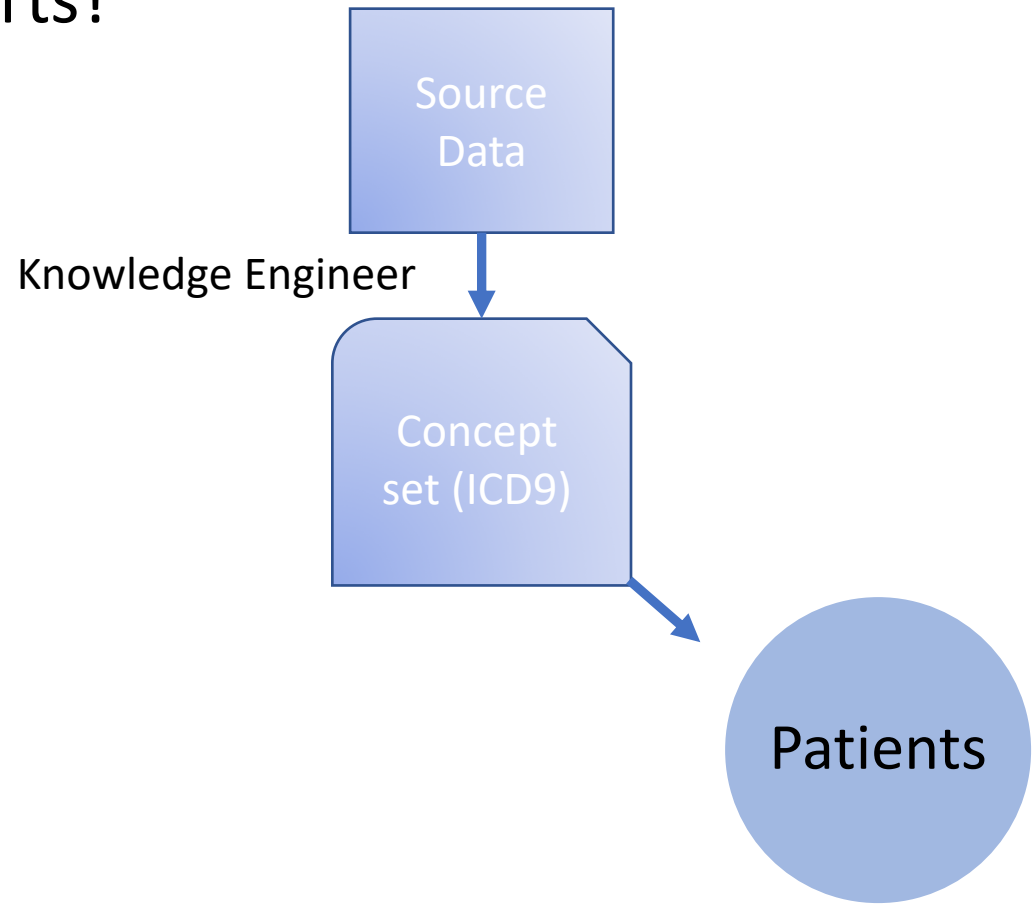
Intro

- Reasons to map:
  - International (with only 5% of the world population, the US is limited in what observational research it can do)
  - join future data (ICD10-CM plus problem lists plus NLP)
  - shift away from billing codes as the primary means of specifying patients

Question

To what extent does OHDSI preserve patient membership in phenotype cohorts?



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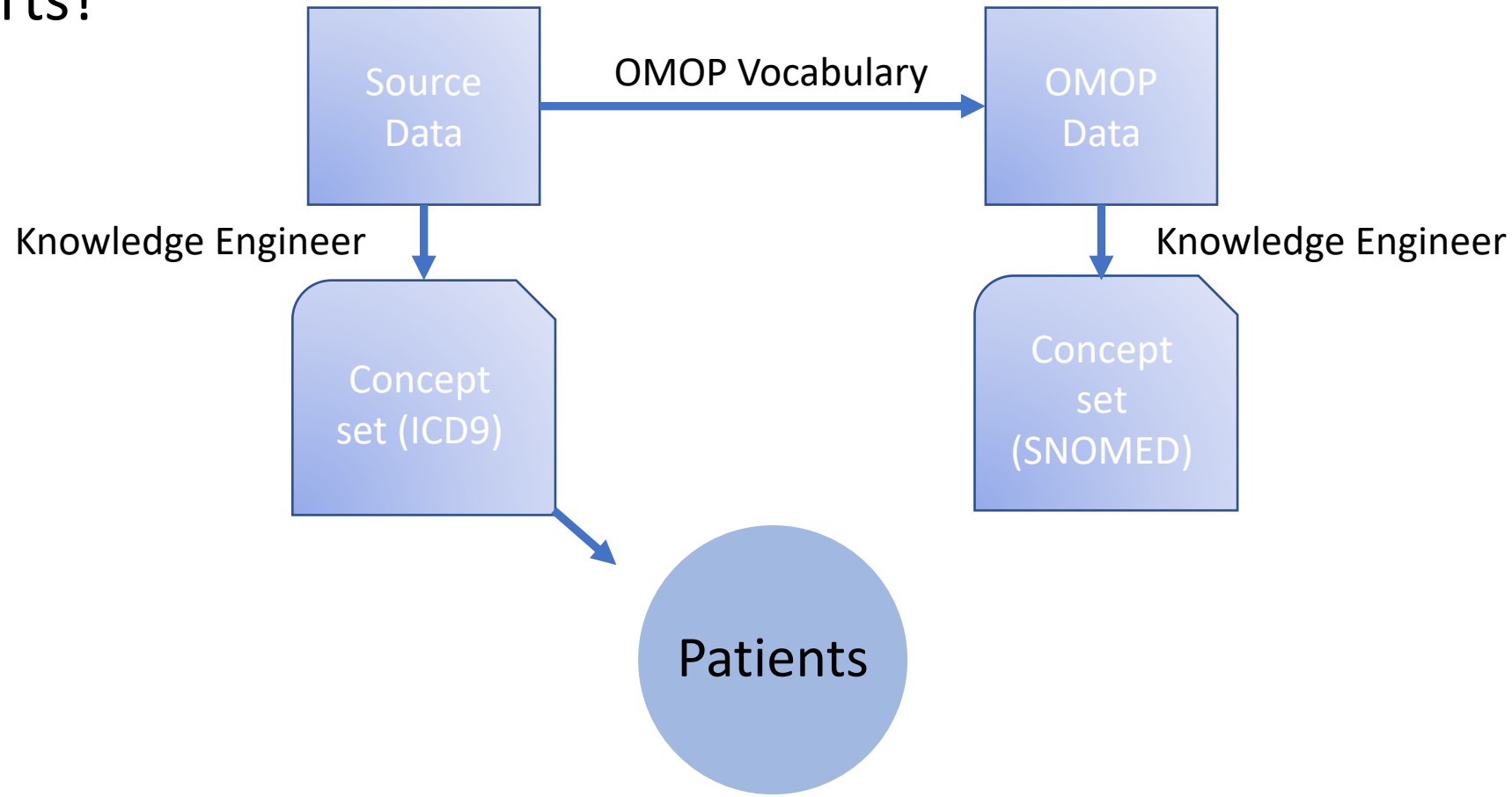

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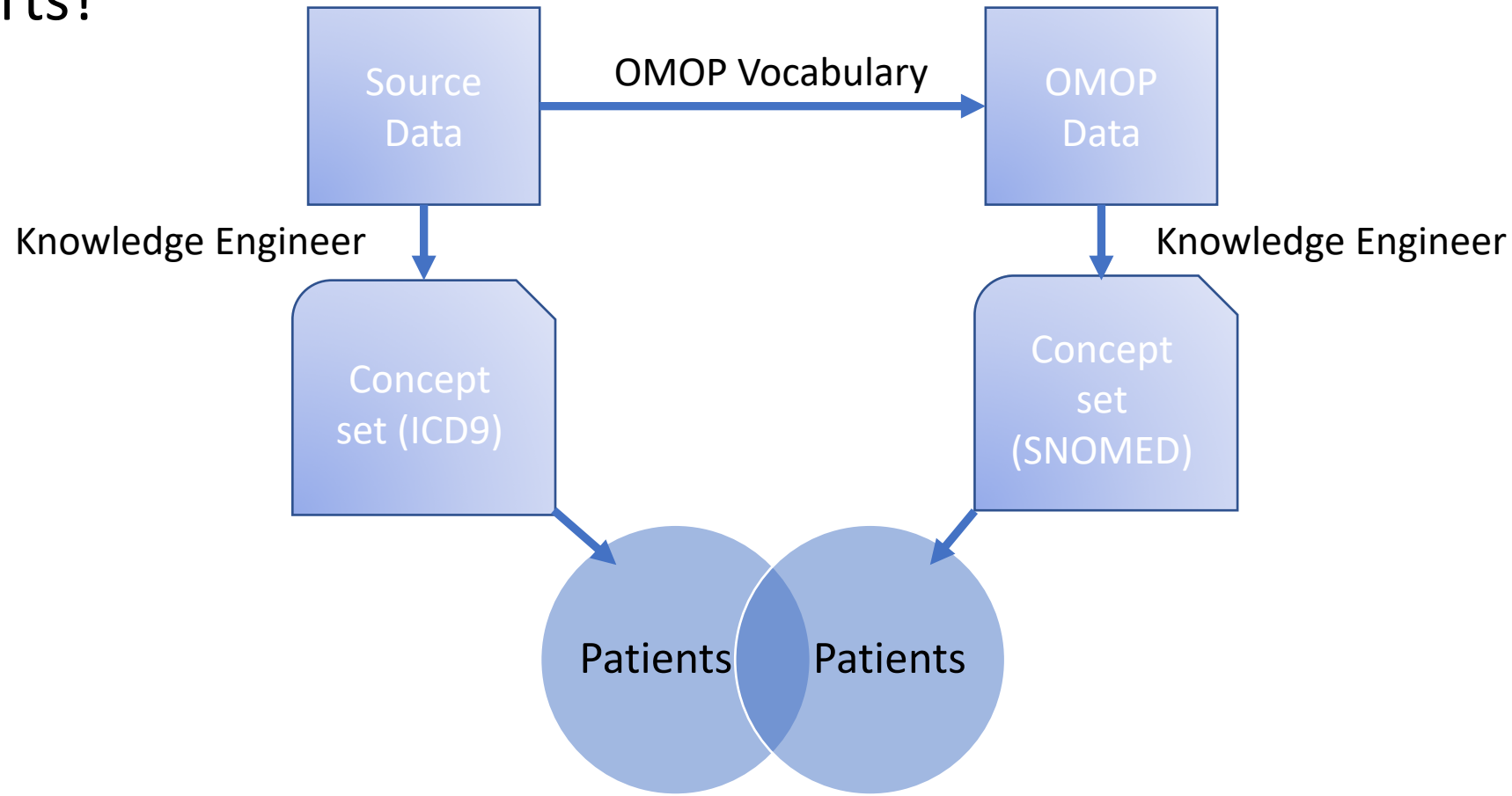

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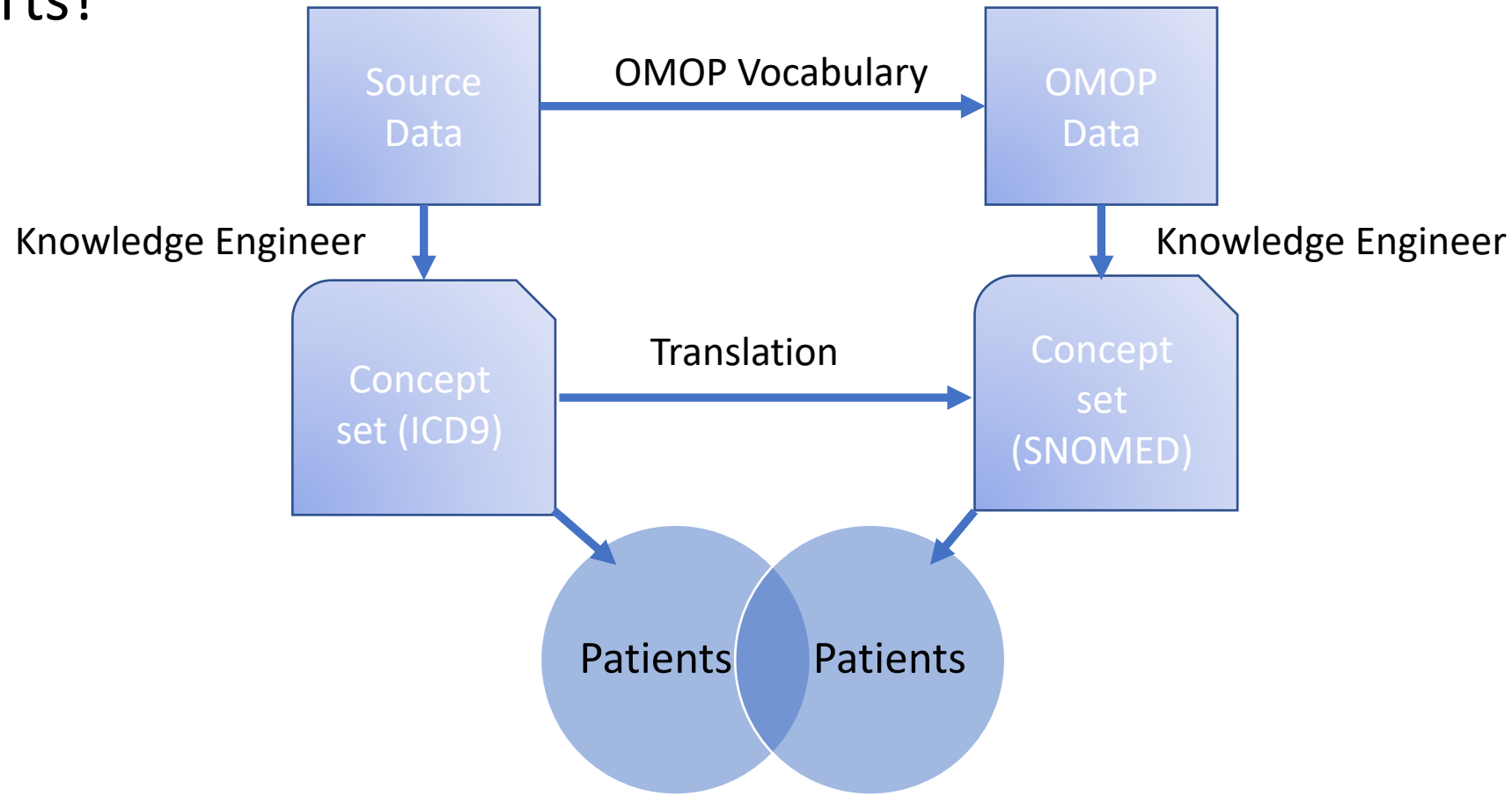
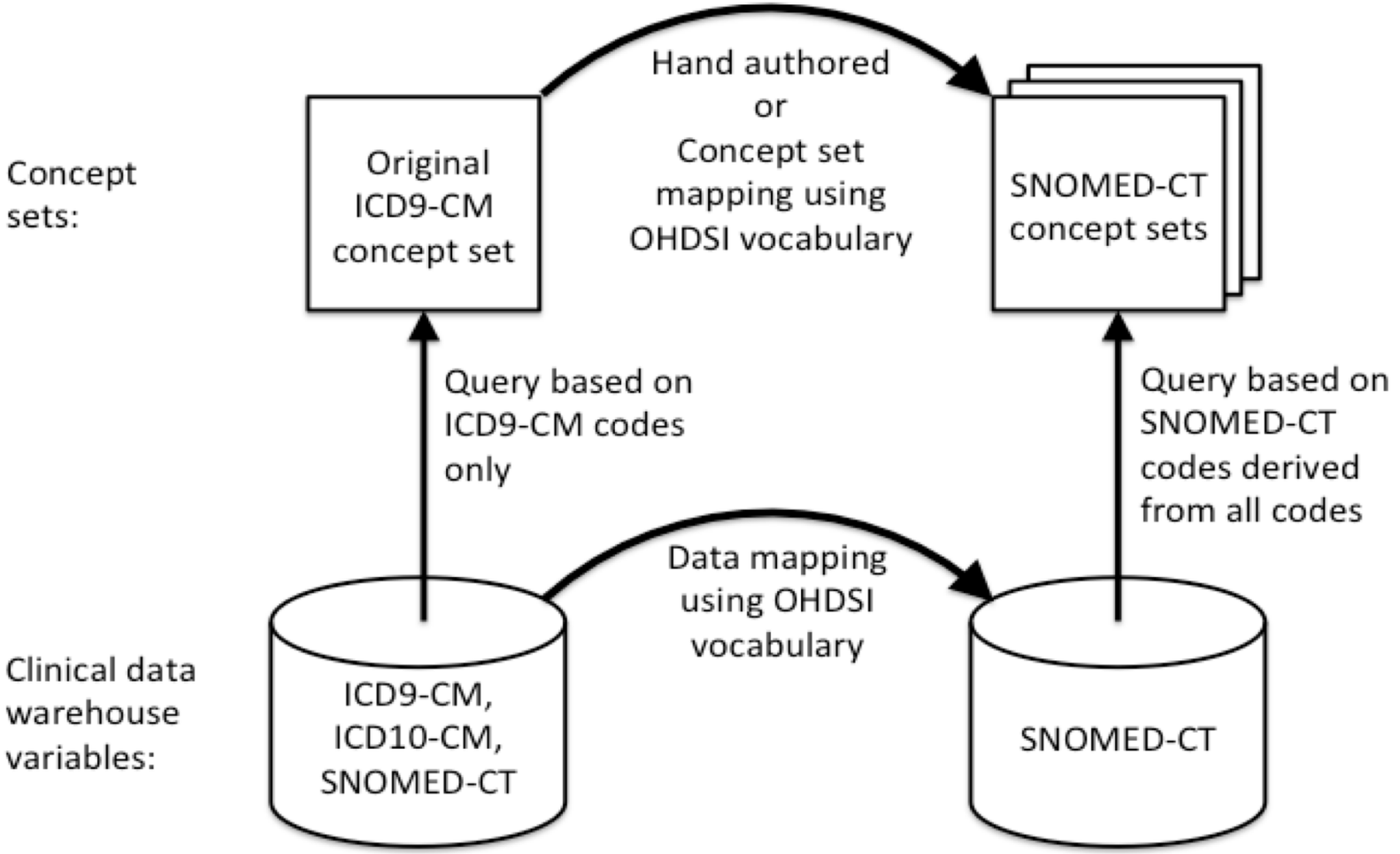


Experimental Design: Compare patient cohorts defined by a) original concept sets on unmapped data, b) mapped concept sets on mapped data



Original ICD9-CM concept set: no mappings

Algorithm (from eMERGE)	Original ICD9-CM concept set‡
Heart failure (HF) [1]	428.*
Heart failure as exclusion diagnosis (HF2) [2]	428.*
Type-1 diabetes mellitus (T1DM) [3]	250.x1, 250.x3
Type-2 diabetes mellitus (T2DM) [4]	250.x0, 250.x2
Appendicitis (Appy) [6]	540.*
Attention deficit hyperactivity disorder (ADHD) [5]	314, 314.0, 314.01, 314.1, 314.2, 314.8, 314.9
Cataract (Catar) [7]	366.10, 366.12, 366.13, 366.14, 366.15, 366.16, 366.17, 366.18, 366.19, 366.21, 366.30, 366.41, 366.45, 366.8, 366.9
Crohn's disease (Crohn) [8]	555, 555.0, 555.1, 555.2, 555.9
Rheumatoid arthritis (RA) [9]	714, 714.0, 714.1, 714.2 (M05*, M06*)

What: Original ICD9-CM concept set generated by the phenotype author.

How: Run against patients' original ICD9-CM terms.

Why: Show what would have happened before either data or concept sets were mapped.

Author's INTENT source concept set: no mappings

- We extended original ICD9-CM concepts to include similar ICD10 and SNOMED CT codes
- This allows us to acquire a cohort, using unmapped data/queries, that would reflect the author's intent under a broader availability of terminologies
- Also corrected obvious errors in original concept set

Knowledge engineered concept set (map data only)

- What: By-hand SNOMED CT concept sets
- How: Run against OHDSI-mapped data in the form of SNOMED CT terms
- 2 intentions of concept set mapping:
  1. SNOMED ***"mimic"***
     - Designed to mimic the original ICD9-CM concept set as much as possible, ignoring data from other vocabularies
  2. SNOMED ***"optimize"***:
     - Designed to carry out phenotype author's intent to ICD9-CM, ICD10-CM, and SNOMED-CT

Knowledge engineered concept set: SNOMED mimic

- Mimic original ICD9-CM concept set as much as possible
- Create a SNOMED concept set expected to ONLY find people who had the original ICD9 codes
- E.g. Does not try to find patients who might have a related ICD10-CM code

Knowledge engineered concept set: SNOMED optimize

- Interpret and extend author's original intent (e.g. Find people with appendicitis)
- Find patients with relevant ICD9-CM, ICD10-CM, and SNOMED-CT codes (e.g. that would reflect author's intent, as demonstrated by their ICD9 concept set)

Automatically generated concept sets (map data AND concept sets)

- What: generated automatically from the original ICD9-CM set using OHDSI vocabulary mappings
- How: Run against OHDSI-mapped data in the form of SNOMED CT terms.
- 4 granularities for concept set mapping:
  1. SNOMED "no descendants"
     - OHDSI mappings from ICD9-CM to SNOMED-CT, ***without*** SNOMED hierarchy.
  2. SNOMED "all descendants"
     - includes descendants of mapped terms
  3. SNOMED "descendants x child"
     - includes descendants of mapped terms **only if none of the term's CHILDREN are also in the concept set.** (limited descendants)
  4. SNOMED "descendants x descendants"
     - includes descendants of mapped terms **only if none of the term's DESCENDANTS are also in the concept set.** (more limited descendants)

Results – code level

- Some knowledge engineered mappings just worked
- Multiple source codes (ICD) to one standard code (SNOMED CT)
- One source code (ICD) to multiple standard codes (SNOMED CT)
- Missing OMOP codes
- Information gain

Results – code level

- Some knowledge engineered mappings just worked
  - Acute appendicitis – 1 code and descendants
  - Crohn’s disease – 2 codes and descendants
  - Heart failure as an exclusion diagnosis – 3 codes and descendants
  - Heart failure as an inclusion diagnosis – 29 code and some descendants

Results – code level

- Multiple source codes (ICD) to one standard code (SNOMED CT)
  - Biggest challenge
  - Causes ambiguity so that either need to gain or lose patients
  - Attention deficit hyperactivity disorder
    - Includes ICD9-CM 314.0 “Attention deficit disorder of childhood” but excludes its child, 314.00 “Attention deficit disorder without mention of hyperactivity”
    - Both of these terms map to SNOMED CT 192127007 “Child attention deficit disorder”
  - Type-2 diabetes mellitus
  - Type-1 diabetes mellitus
  - Rheumatoid arthritis
  - Cataract

Results – code level

- One source code (ICD) to multiple standard codes (SNOMED CT)
  - Mostly happens with compound ICD9-CM terms
  - Can solve with conjunction in SNOMED
  - Type-1 diabetes mellitus
    - More of an oversight; did not need conjunction
  - Type-2 diabetes mellitus
    - More of an oversight; did not need conjunction

Results – code level

- Missing OMOP codes
  - Generally new codes that have not been added to OHDSI yet
  - Type-1 diabetes mellitus
    - Not used for patient data yet so no consequence
  - Type-2 diabetes mellitus
    - Not used for patient data yet so no consequence

Results – code level

- Information gain
  - Superior hierarchy of SNOMED CT versus strict hierarchy of ICD9-CM can improve the concept set
  - E.g., find codes in different areas of the ICD9-CM hierarchy
  - Heart failure as an exclusion diagnosis
    - Added terms

Results – patient cohort level

- Mapped Cohorts vs Original ICD9 cohort
- Mapped Cohorts vs Author’s Intent
- Automated batch analysis
  - 122 eMERGE concept sets
  - original ICD9 cohort vs AUTO mappings

Results: Appendicitis

-0 patient loss with any query

Pheno	#Cases	ICD9 set‡		SNOMED mimic		SNOMED optimize		SNOMED no desc		SNOMED all desc	
		Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss
VS original	9,887	0	0	0	0	0	0	0	0	0	0

‡ This column is used as the gold standard and therefore must have perfect performance

Results: Appendicitis

-0 patient loss with any query (vs original)
-same queries are extendable to capture author's intent

Pheno	#Cases	ICD9 set‡		SNOMED mimic		SNOMED optimize		SNOMED no desc		SNOMED all desc	
		Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss
VS original	9,887	0	0	0	0	0	0	0	0	0	0
VS intent	9,920	0	33	0	0	0	0	0	8	0	0

‡ This column is used as the gold standard and therefore must have perfect performance

Results: ADHD

-no perfect mapped query
-tradeoff between gain and loss
-automated don't perform well
-0.1% loss from knowledge engineering is probably better than 9.4% gain with auto-mapping
-KE queries are extendable to capture author's intent

Pheno	#Cases	ICD9 set‡		SNOMED mimic		SNOMED optimize		SNOMED no desc		SNOMED all desc	
		Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss
VS original	14,399	0	0	0	19	0	19	1362	0	1362	0
VS intent	14,547	0	148	0	39	0	19	1359	19	1359	0

Results: Mapped Cohorts vs Original ICD9 cohort

-0 patient loss from automatic mappings
-MIMIC does a good job a mimicing the patient cohort

Pheno	#Cases	ICD9 set‡		SNOMED mimic		SNOMED optimize		SNOMED no desc		SNOMED all desc	
		Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss
HF	75,312	0	0	0	0	0	0	0	0	1262	0
HF2	75,312	0	0	0	0	0	0	0	0	1262	0
T1DM	27,861	0	0	0	23	0	23	108	0	943	0
T2DM	125,342	0	0	3	30	3	30	34	0	1318	0
Appy	9,887	0	0	0	0	0	0	0	0	0	0
ADHD	14,399	0	0	0	19	0	19	1362	0	1362	0
Catar	50,879	0	0	50	0	74	0	50	0	2491	0
Crohn	4,679	0	0	0	0	0	0	0	0	0	0
RA	9,655	0	0	0	16	0	16	0	0	25103	0

‡ This column is used as the gold standard and therefore must have perfect performance

Results: Mapped Cohorts vs Author's Intent

- Author's Intent brings in new patients, compared to only original ICD9 codes.

Pheno	#Cases	ICD9 set	
		Gain	Loss
HF	75,626	0	314
HF2	76,958	0	1646
T1DM	27,935	0	74
T2DM	126,828	0	1486
Appy	9,920	0	33
ADHD	14,547	0	148
Catar	50,953	0	194
Crohn	4,679	0	0
RA	9,655	0	0

Results: Mapped Cohorts vs Author's Intent

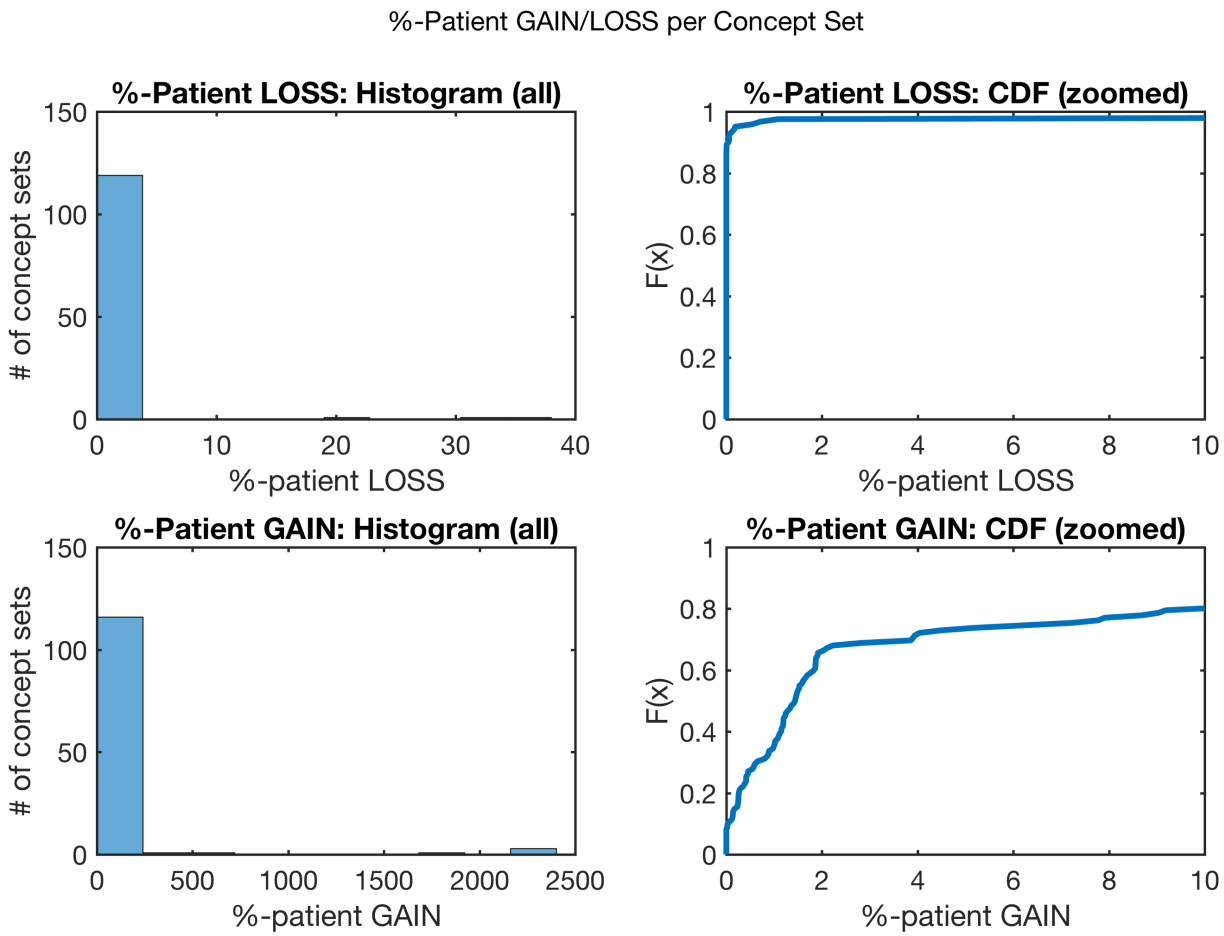
- Author's Intent brings in new patients, compared to only original ICD9 codes.
- OPTIMIZE does a good job of finding patients that reflect the author's intent
  - Generally <1:1000 (worst is RA at 0.013)

Pheno	Cases#	ICD9 set		SNOMED mimic		SNOMED optimize		SNOMED no desc		SNOMED all desc	
		Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss	Gain	Loss
HF	75,626	0	314	0	0	0	0	0	0	1332	0
HF2	76,958	0	1646	0	1332	0	0	0	1332	0	0
T1DM	27,935	0	74	0	23	0	23	108	67	943	0
T2DM	126,828	0	1486	3	1412	3	30	34	1486	1317	0
Appy	9,920	0	33	0	0	0	0	0	8	0	0
ADHD	14,547	0	148	0	39	0	19	1359	19	1359	0
Catar	50,953	0	194	39	26	39	2	39	26	2451	0
Crohn	4,679	0	0	0	0	0	0	0	0	0	0
RA	9,655	0	0	113	0	113	16	113	0	25289	0

Results: Patient Gain/Loss using only automatically mapped queries

Results: Patient Gain/Loss using only automatically mapped queries

- Patient loss was almost always exactly 0.
  - Exceptions occurred when invalid ICD9 codes were specified
- Patient gain was usually <4% (70% of concept sets).
- Patient gain was sometimes very large (e.g. > 100%)
- Concpet sets with largest %-gain had obvious flaws:
  - Missing icd9 codes that probably should be included
  - Typos



Discussion 1

- 5/9 concept sets had code mapping issues
- But effect on patient cohort is minimal
  - 8/9 concept sets produced error < 1/700, 9th was 1.3%
  - Small compared to coding error 2% to 50%
- Mapping process can improve queries
  - 2/9 mapping revealed codes that were clearly intended but missed from the original list

Discussion 2

- Automated mappings did not perform well consistently (versus
- OHDSI retains the source data so can always go back to th needed