2025 Phenotype Phebruary Week 2

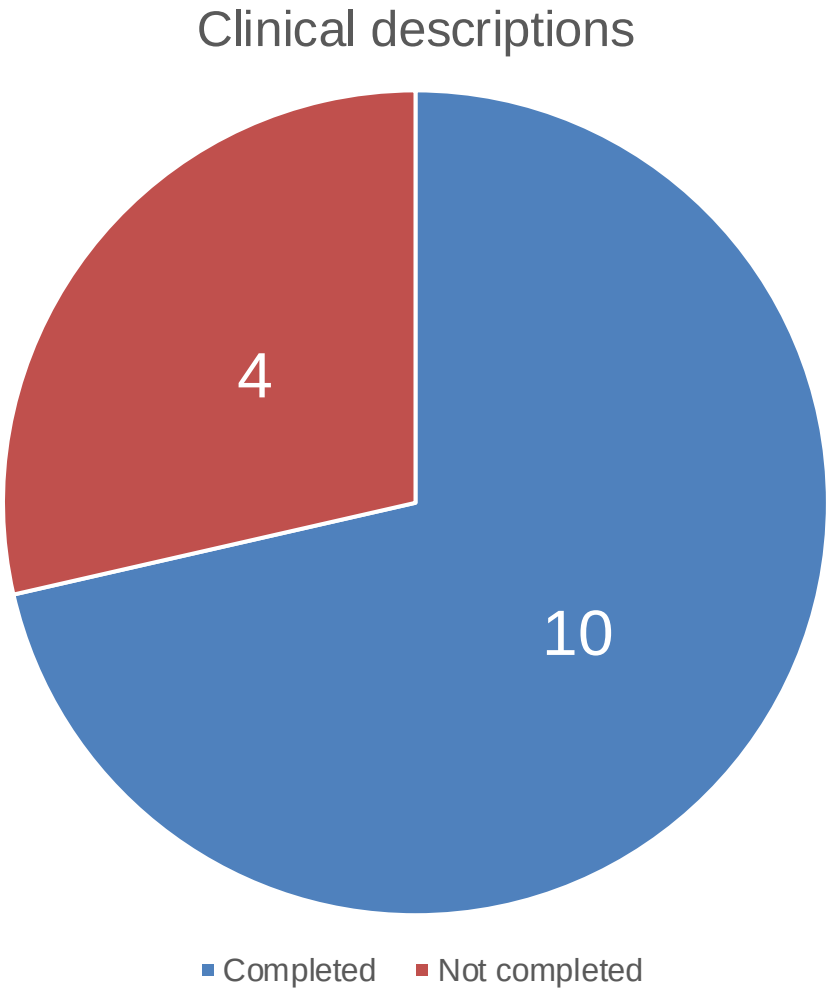
Phenotype Phebruary 2025 Calendar

Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
Week 1: Clinical Descriptions & Prior Work						
				Study team's complete clinical description	*Office hours*	
6	7	8				
ets and Logic						
				Study team's complete 1st drafts of cohorts in Atlas	*Office hours*	
13	14	15				
s & Iterations						
				Study team's complete cohort review and iterations	*Office hours*	
20	21	22				
ools for Evaluation						
		Community call update & evaluation demos	*Office hours*	*Study team's wrap up cohorts and document evaluation*	*Office hours*	*All cohorts are done!*
23	24	25	26	27	28	

- **Week 1 – Clinical Description:**
  A session is planned to discuss clinical descriptions. Participants are expected to use a Gen AI prompt (developed by @Gowtham_Rao) that extracts necessary information to form a phenotype. This step also includes a literature search and an exploration of existing phenotype definitions (for example, checking for pre-existing definitions of AKI or obesity management).
- **Week 2 – Concept Set and Logic Building:**
  The second week focuses on creating the concept sets and building the logical framework of the phenotype. Here, many participants already familiar with tutorial work on concept sets building are expected to contribute. The study leads are expected to be fully engaged.
- **Weeks 3–4 – Evaluation and Iteration:**
  The final two weeks are dedicated to evaluating the developed phenotypes using tools such as cohort diagnostics. Iterations and refinements will be made based on these diagnostics. There is also mention of showcasing additional tools and validation approaches during these weeks.

https://forums.ohdsi.org/t/ohdsi-phenotype-phebruary-and-workgroup-updates/20940/69

Week 1 results



2 page – 25 page documents

3 useful conversations stemming just from clinical descriptions

30+ people attended the office hours

Crohn's disease clinical description

Overview: *Crohn's disease (CD) is an idiopathic inflammatory bowel disease that can affect any portion of the intestinal tract with focal, asymmetric, transmural, and granulomatous inflammation. Intestinal involvement is discontinuous; "skip areas" of apparently normal tissue separate severely involved segments. Reported incidence varies geographically, ranging from 0-20.02 cases per 100,000 py in North America and 0.3-12.7 cases per 100,000 in Europe. Reported prevalence is highest in Europe (322 cases per 100,000 persons in Germany) and North America (319 cases per 100,000 persons in Canada) [4]. Risk factors include smoking, diet, and genetic factors.*

Presentation: *Symptoms can be non-specific; patients are oft syndrome [5]. Diarrhea and abdominal pain are most commo fatigue, weight loss, fever, anemia, and recurrent fistulas or c display prominent extraintestinal manifestations such as arth oral ulcers, anemia, clubbing, kidney stones, uveitis, and abn principal pathologic features of CD include discontinuous foc granulomas, and perianal involvement [1-3].*

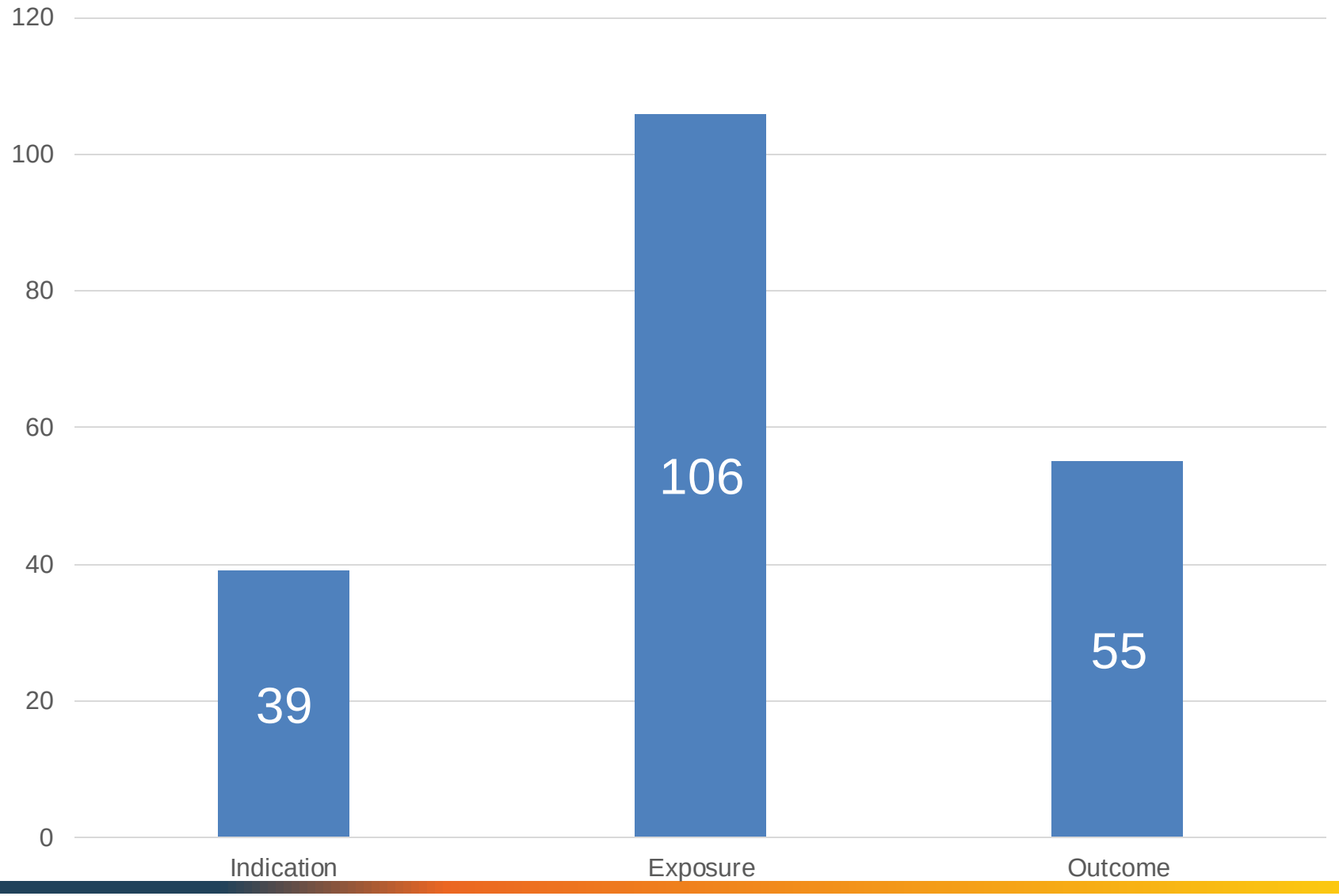
Assessment: *Diagnosis includes serological and fecal biomark utrasonography, CTE and MRI) and endoscopic imaging (e.g. diagnosis includes Infections (amebiasis, campylobacter, C. di ischemic colitis, sarcoidosis, small vessel vasculitis*

Plan: *One of the primary goals of treatment is mucosal healing [4]. Corticosteroids (e.g. budesonide and prednisone) have been the cornerstone of CD management for decades and are recommended for treatment of mild-to-moderate ileal and moderate-to-severe ileocolonic CD. Steroids are recommended to induce remission but not for maintenance. Antibiotics are indicated in the case of perianal complications. For maintenance therapy, immunusuppressants (e.g. azathioprine and methotrexate) and biologic agents (e.g. anti-TNFs, ustekinumab, vedolizumab) are recommended. Biologics may also be used for induction of remission. In both the adult and pediatric CD patient populations, the overall goal of treatment is to achieve remission of acute, active disease and to maintain disease remission [2, 4, 6]. However, many patients do not attain, or subsequently lose, clinical benefit even with combinations of these therapies.*

Prognosis: *CD is a chronic, systemic, progressive disease. The natural history of CD comprises a progression from intestinal inflammation to the development of strictures and/or penetrating disease (ie, with fistulas and/or abscesses). Intestinal obstruction ultimately occurs in 20% to 30% of patients and fistula formation affects approximately a quarter of patients [4-6]. Patients often experience an impact of disease on quality of life.*

Week 1 results

201 phenotype
63 available in phenotype library or in other sources



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Concept Set creation

First-episode psychosis

Tatiana Skugarevskaya

How did we get here?

Study idea → Clinical description → List of codes from literature

Clinical descriptions of the phenotypes

Introduction

Psychotic features can accompany a big number of mental disorders and can be caused by a variety of different causes. Since the idea of this study originated from an evidence gap in treatment guidelines for first-episode schizophrenia, we will be focusing on first-episode psychosis. The terms 'first-episode schizophrenia' and 'first-episode psychosis' are often used interchangeably, however, some differences need to be highlighted.

When a first-in-a-lifetime psychotic episode manifests, it is often extremely difficult or impossible to diagnose a patient with a specific disorder (e.g. schizophrenia, schizoaffective disorder, newly diagnosed depression, etc.). This is reflected in the variety of diagnostic systems and their new emerging versions. Diagnosis of a chronic disorder is often made later, if subsequent episodes happen, enriching the clinical picture. We therefore do not want to capture patients with pre-existing conditions (such as drug/alcohol abuse or bipolar disorder).

Here, a decision was made to define psychosis phenotype through Condition domain only, since psychiatric prescriptions can intersect between diagnoses a lot, and thus are not condition specific.

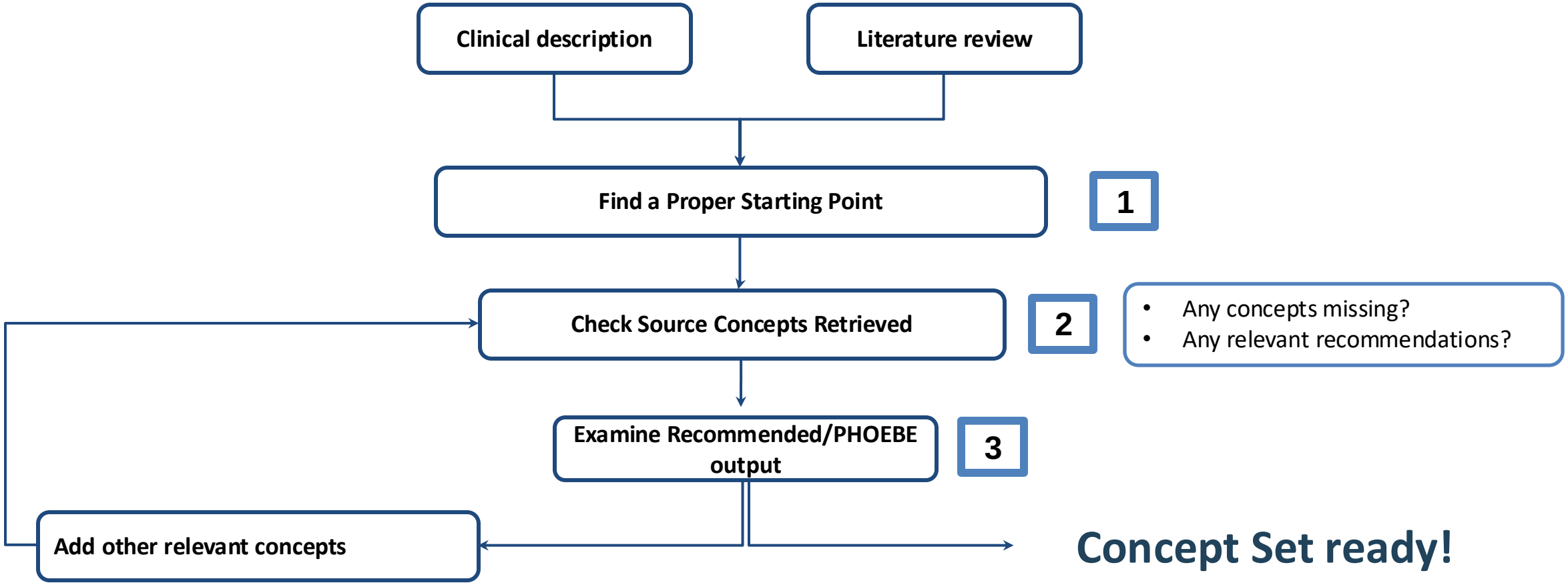
Commonly used codes in the literature (ICD10):

- Schizophrenia-spectrum psychotic disorders (F20.x-F29)
- Manic episode with psychotic symptoms (F30.2)
- Depressive episode with psychotic symptoms (F32.3)

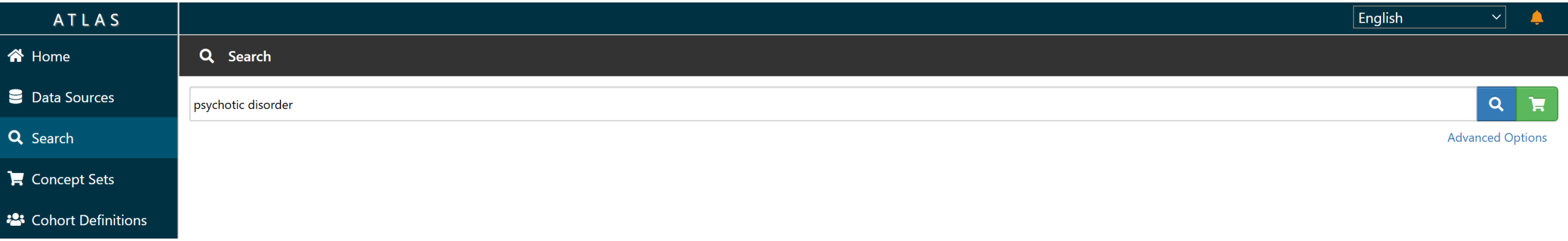
Exclude:

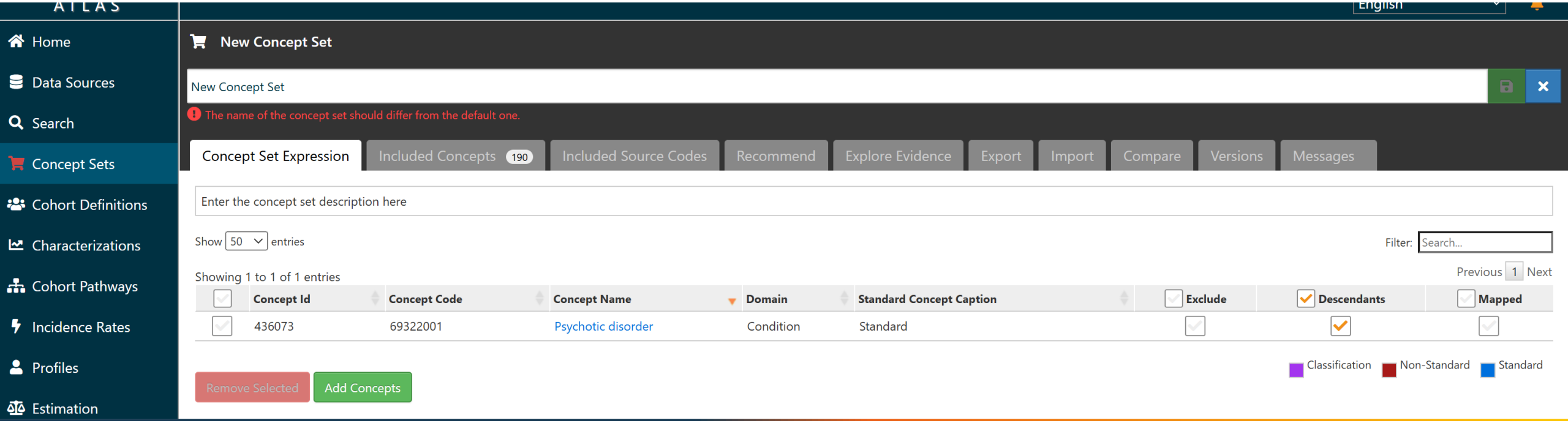
- Drug-induced psychoses (e.g. F10.5 Mental and behavioural disorders due to use of alcohol, Psychotic disorder)
- Organic psychotic conditions (e.g. F06.2 Organic delusional [schizophrenia-like] disorder)

Condition concept set construction algorithm



Starting to make a concept set





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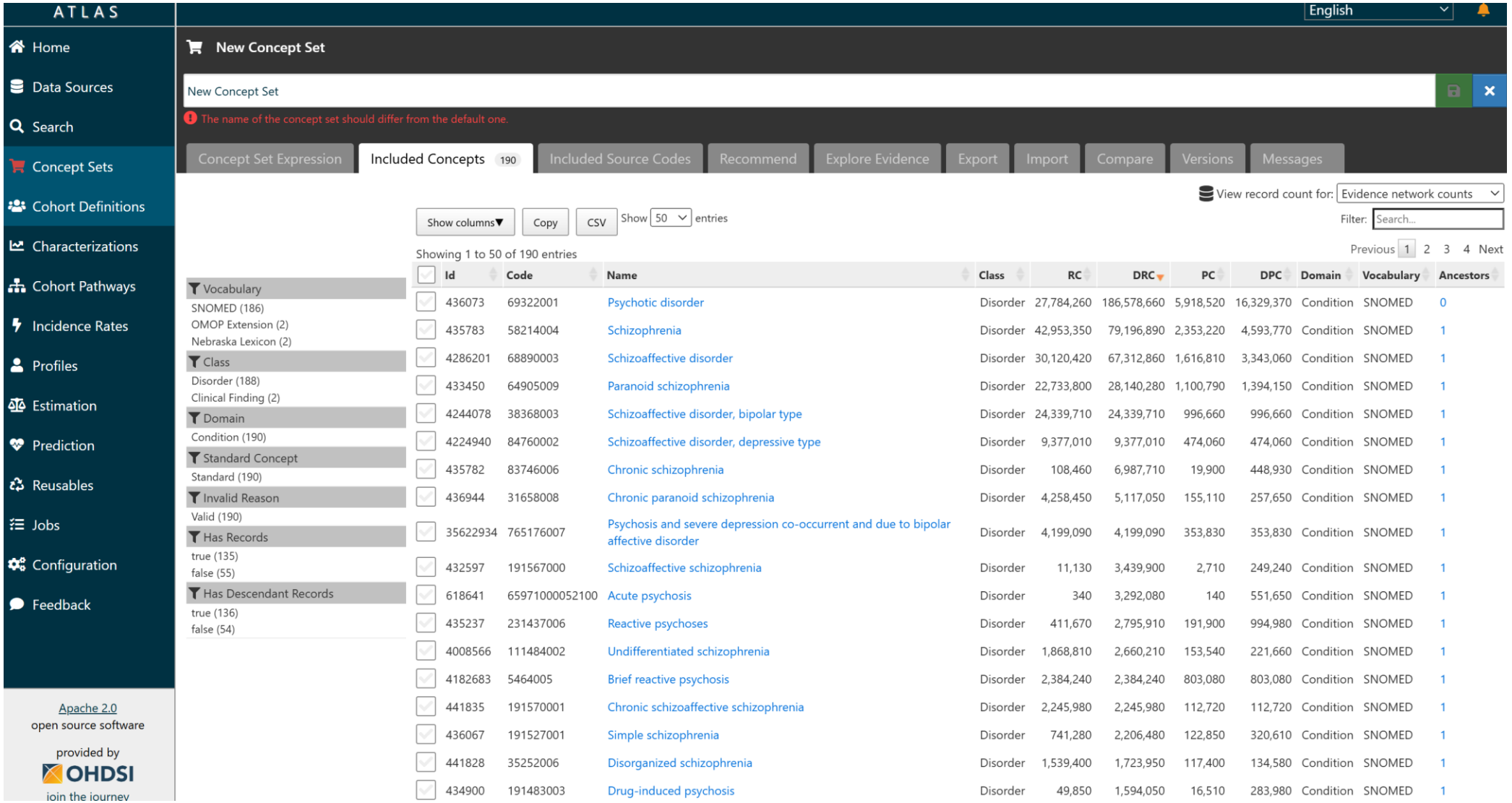
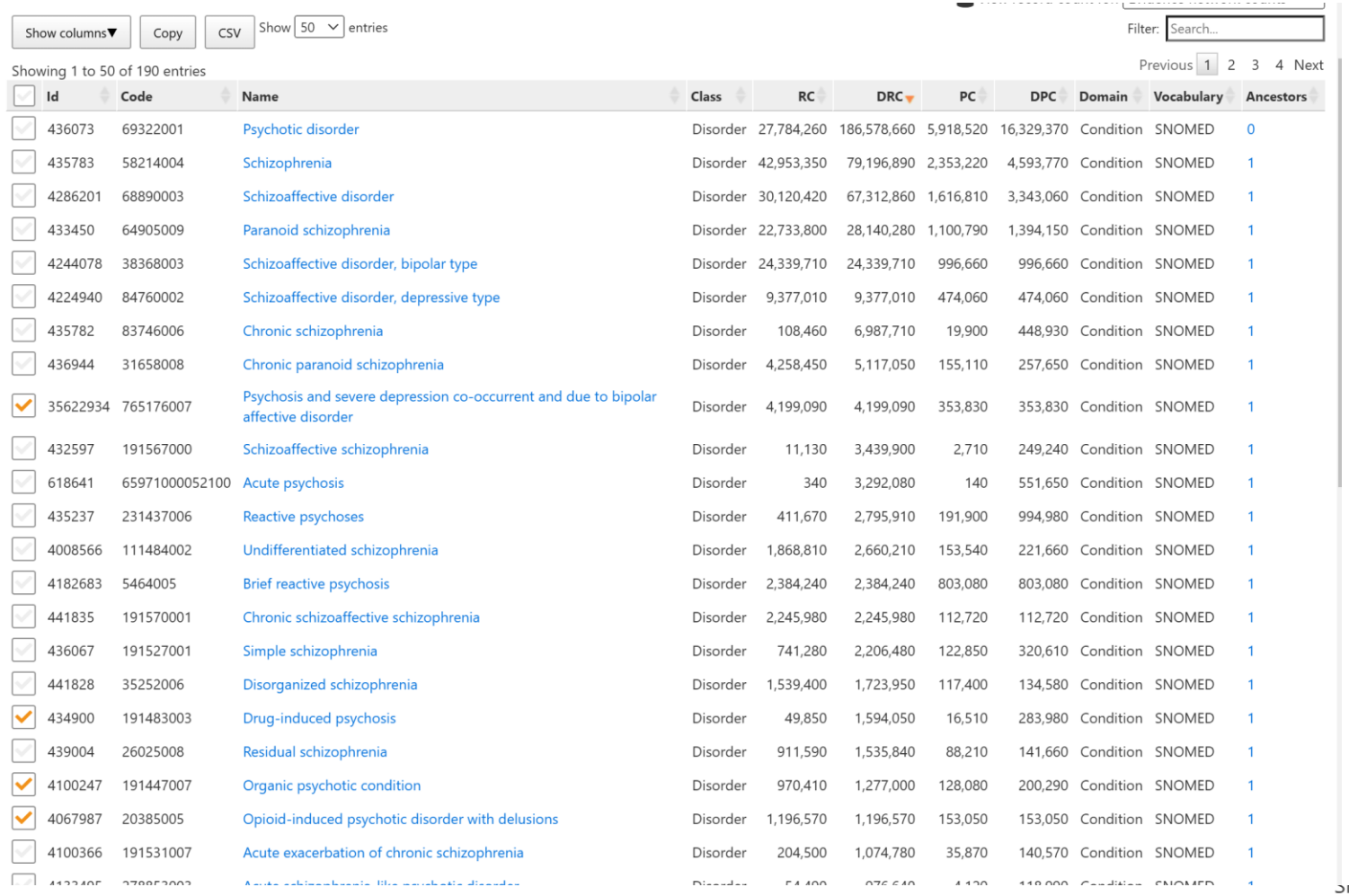


Excluding codes based on the clinical description



Show columns▼ Copy CSV Show 50 entries Filter: Search...

Showing 1 to 50 of 190 entries Previous 1 2 3 4 Next

	Id	Code	Name	Class	RC	DRC	PC	DPC	Domain	Vocabulary	Ancestors
☐	436073	69322001	Psychotic disorder	Disorder	27,784,260	186,578,660	5,918,520	16,329,370	Condition	SNOMED	0
☐	435783	58214004	Schizophrenia	Disorder	42,953,350	79,196,890	2,353,220	4,593,770	Condition	SNOMED	1
☐	4286201	68890003	Schizoaffective disorder	Disorder	30,120,420	67,312,860	1,616,810	3,343,060	Condition	SNOMED	1
☐	433450	64905009	Paranoid schizophrenia	Disorder	22,733,800	28,140,280	1,100,790	1,394,150	Condition	SNOMED	1
☐	4244078	38368003	Schizoaffective disorder, bipolar type	Disorder	24,339,710	24,339,710	996,660	996,660	Condition	SNOMED	1
☐	4224940	84760002	Schizoaffective disorder, depressive type	Disorder	9,377,010	9,377,010	474,060	474,060	Condition	SNOMED	1
☐	435782	83746006	Chronic schizophrenia	Disorder	108,460	6,987,710	19,900	448,930	Condition	SNOMED	1
☐	436944	31658008	Chronic paranoid schizophrenia	Disorder	4,258,450	5,117,050	155,110	257,650	Condition	SNOMED	1
☑	35622934	765176007	Psychosis and severe depression co-occurrent and due to bipolar affective disorder	Disorder	4,199,090	4,199,090	353,830	353,830	Condition	SNOMED	1
☐	432597	191567000	Schizoaffective schizophrenia	Disorder	11,130	3,439,900	2,710	249,240	Condition	SNOMED	1
☐	618641	65971000052100	Acute psychosis	Disorder	340	3,292,080	140	551,650	Condition	SNOMED	1
☐	435237	231437006	Reactive psychoses	Disorder	411,670	2,795,910	191,900	994,980	Condition	SNOMED	1
☐	4008566	111484002	Undifferentiated schizophrenia	Disorder	1,868,810	2,660,210	153,540	221,660	Condition	SNOMED	1
☐	4182683	5464005	Brief reactive psychosis	Disorder	2,384,240	2,384,240	803,080	803,080	Condition	SNOMED	1
☐	441835	191570001	Chronic schizoaffective schizophrenia	Disorder	2,245,980	2,245,980	112,720	112,720	Condition	SNOMED	1
☐	436067	191527001	Simple schizophrenia	Disorder	741,280	2,206,480	122,850	320,610	Condition	SNOMED	1
☐	441828	35252006	Disorganized schizophrenia	Disorder	1,539,400	1,723,950	117,400	134,580	Condition	SNOMED	1
☑	434900	191483003	Drug-induced psychosis	Disorder	49,850	1,594,050	16,510	283,980	Condition	SNOMED	1
☐	439004	26025008	Residual schizophrenia	Disorder	911,590	1,535,840	88,210	141,660	Condition	SNOMED	1
☑	4100247	191447007	Organic psychotic condition	Disorder	970,410	1,277,000	128,080	200,290	Condition	SNOMED	1
☑	4067987	20385005	Opioid-induced psychotic disorder with delusions	Disorder	1,196,570	1,196,570	153,050	153,050	Condition	SNOMED	1
☐	4100366	191531007	Acute exacerbation of chronic schizophrenia	Disorder	204,500	1,074,780	35,870	140,570	Condition	SNOMED	1

Showing 1 to 50 of 190 entries

☑ Exclude ☑ Descendants ☐ Mapped Preview... Add To Concept Set

Expression with exclusions

Concept Set Expression | Included Concepts 120 | Included Source Codes | Recommend | Explore Evidence | Export | Import | Compare | Versions | Messages

Enter the concept set description here

Show 50 entries

Filter: Search...

Showing 1 to 5 of 5 entries

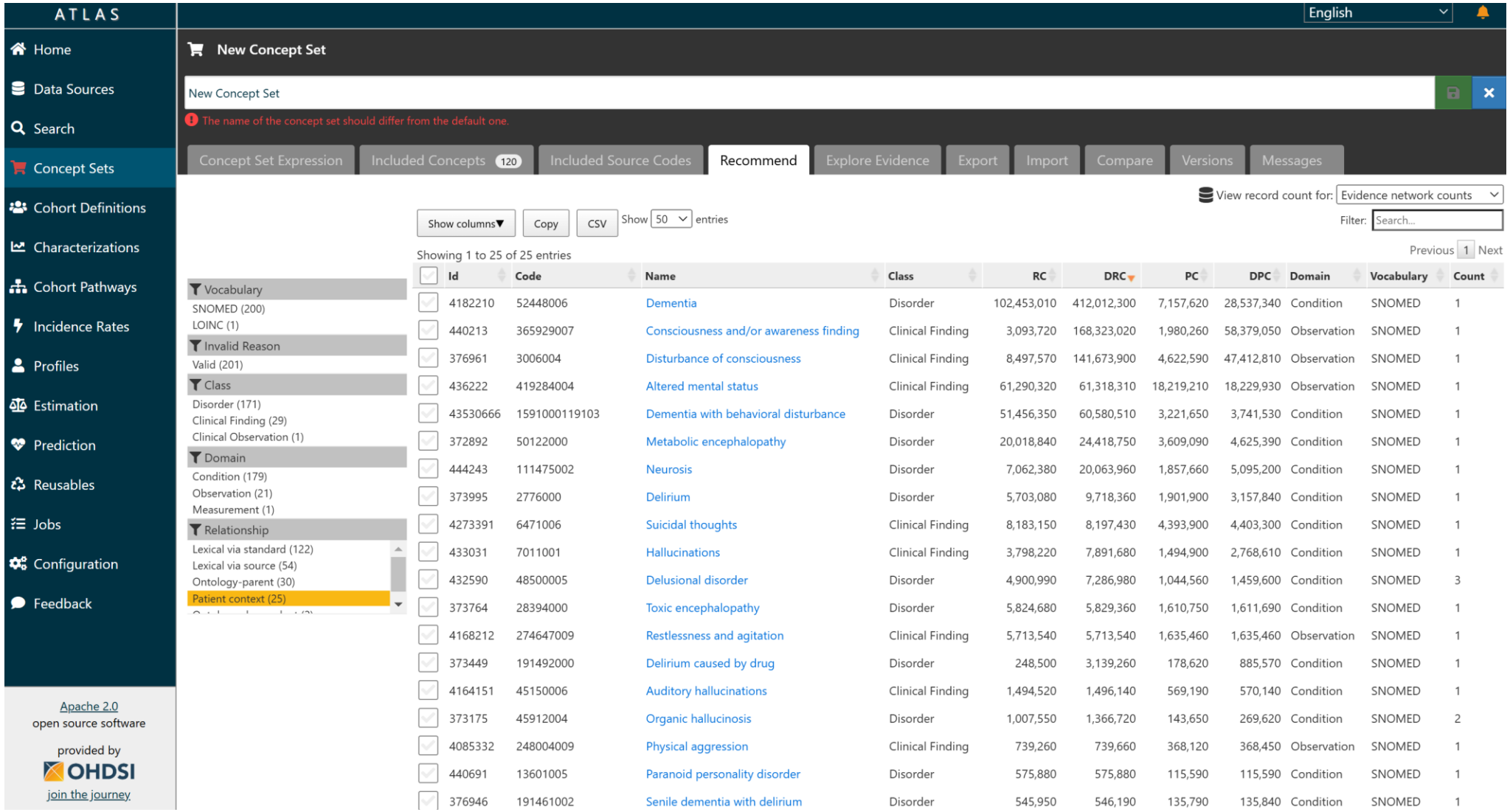
Previous 1 Next

Concept Id	Concept Code	Concept Name	Domain	Standard Concept Caption	Exclude	Descendants	Mapped
436073	69322001	Psychotic disorder	Condition	Standard	☐	☑	☐
35622934	765176007	Psychosis and severe depression co-occurrent and due to bipolar affective disorder	Condition	Standard	☑	☑	☐
4100247	191447007	Organic psychotic condition	Condition	Standard	☑	☑	☐
4067987	20385005	Opioid-induced psychotic disorder with delusions	Condition	Standard	☑	☑	☐
434900	191483003	Drug-induced psychosis	Condition	Standard	☑	☑	☐

Classification Non-Standard Standard

Remove Selected | Add Concepts

Explore recommendations



Id	Code	Name	Class	RC	DRC	PC	DPC	Domain	Vocabulary	Count
4182210	52448006	Dementia	Disorder	102,453,010	412,012,300	7,157,620	28,537,340	Condition	SNOMED	1
440213	365929007	Consciousness and/or awareness finding	Clinical Finding	3,093,720	168,323,020	1,980,260	58,379,050	Observation	SNOMED	1
376961	3006004	Disturbance of consciousness	Clinical Finding	8,497,570	141,673,900	4,622,590	47,412,810	Observation	SNOMED	1
436222	419284004	Altered mental status	Clinical Finding	61,290,320	61,318,310	18,219,210	18,229,930	Observation	SNOMED	1
43530666	1591000119103	Dementia with behavioral disturbance	Disorder	51,456,350	60,580,510	3,221,650	3,741,530	Condition	SNOMED	1
372892	50122000	Metabolic encephalopathy	Disorder	20,018,840	24,418,750	3,609,090	4,625,390	Condition	SNOMED	1
444243	111475002	Neurosis	Disorder	7,062,380	20,063,960	1,857,660	5,095,200	Condition	SNOMED	1
373995	2776000	Delirium	Disorder	5,703,080	9,718,360	1,901,900	3,157,840	Condition	SNOMED	1
4273391	6471006	Suicidal thoughts	Clinical Finding	8,183,150	8,197,430	4,393,900	4,403,300	Condition	SNOMED	1
433031	7011001	Hallucinations	Clinical Finding	3,798,220	7,891,680	1,494,900	2,768,610	Condition	SNOMED	1
432590	48500005	Delusional disorder	Disorder	4,900,990	7,286,980	1,044,560	1,459,600	Condition	SNOMED	3
373764	28394000	Toxic encephalopathy	Disorder	5,824,680	5,829,360	1,610,750	1,611,690	Condition	SNOMED	1
4168212	274647009	Restlessness and agitation	Clinical Finding	5,713,540	5,713,540	1,635,460	1,635,460	Observation	SNOMED	1
373449	191492000	Delirium caused by drug	Disorder	248,500	3,139,260	178,620	885,570	Condition	SNOMED	1
4164151	45150006	Auditory hallucinations	Clinical Finding	1,494,520	1,496,140	569,190	570,140	Condition	SNOMED	1
373175	45912004	Organic hallucinosis	Disorder	1,007,550	1,366,720	143,650	269,620	Condition	SNOMED	2
4085332	248004009	Physical aggression	Clinical Finding	739,260	739,660	368,120	368,450	Observation	SNOMED	1
440691	13601005	Paranoid personality disorder	Disorder	575,880	575,880	115,590	115,590	Condition	SNOMED	1
376946	191461002	Senile dementia with delirium	Disorder	545,950	546,190	135,790	135,840	Condition	SNOMED	1

Expression with the recommendations

Concept Set Expression | Included Concepts 154 | Included Source Codes | Recommend | Explore Evidence | Export | Import | Compare | Versions | Messages

Enter the concept set description here

Show 50 entries

Filter: Search...

Showing 1 to 6 of 6 entries

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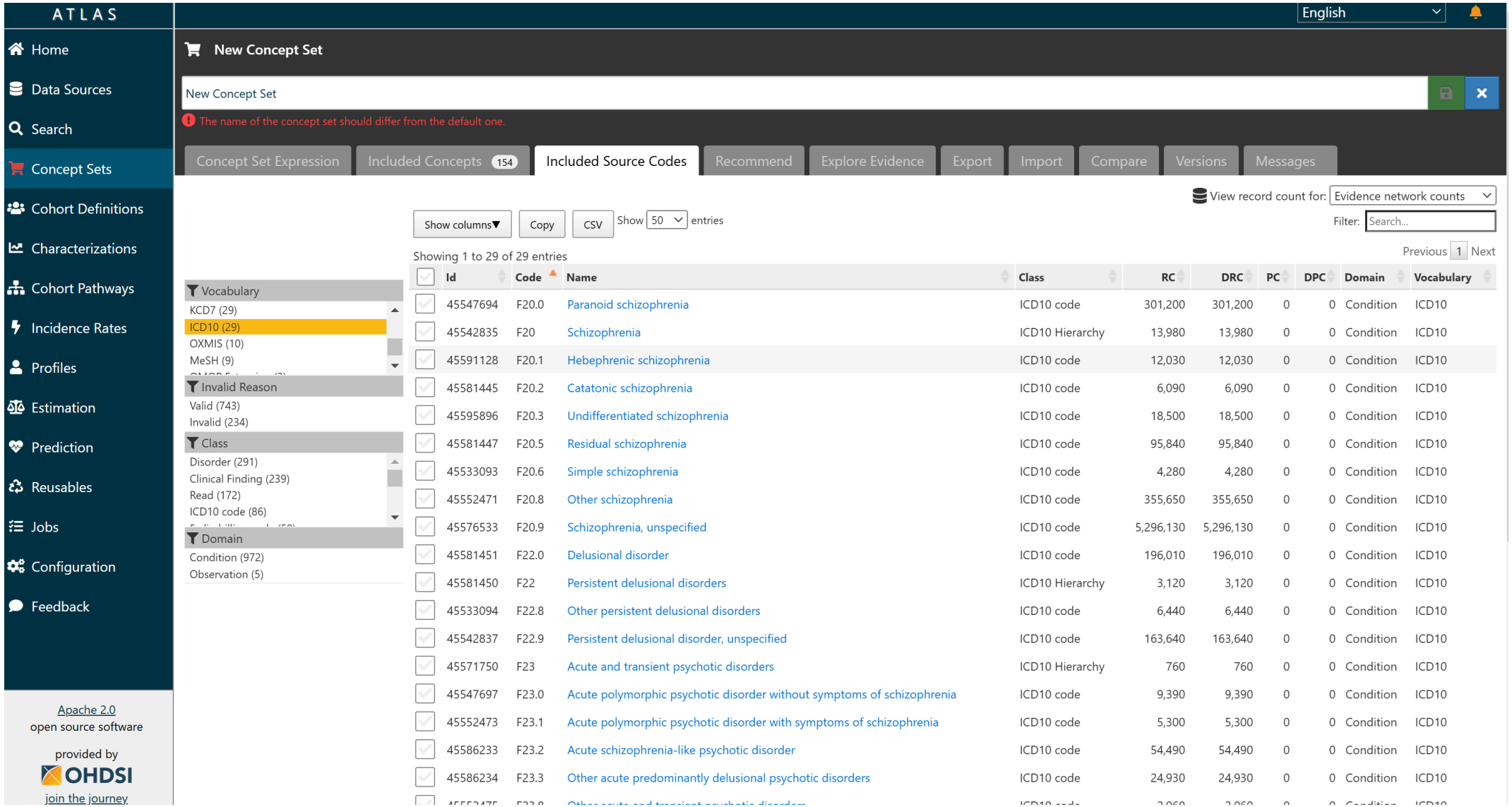
434900	191483003	Drug-induced psychosis	Condition	Standard	☑	☑	☐
432590	48500005	Delusional disorder	Condition	Standard	☐	☑	☐

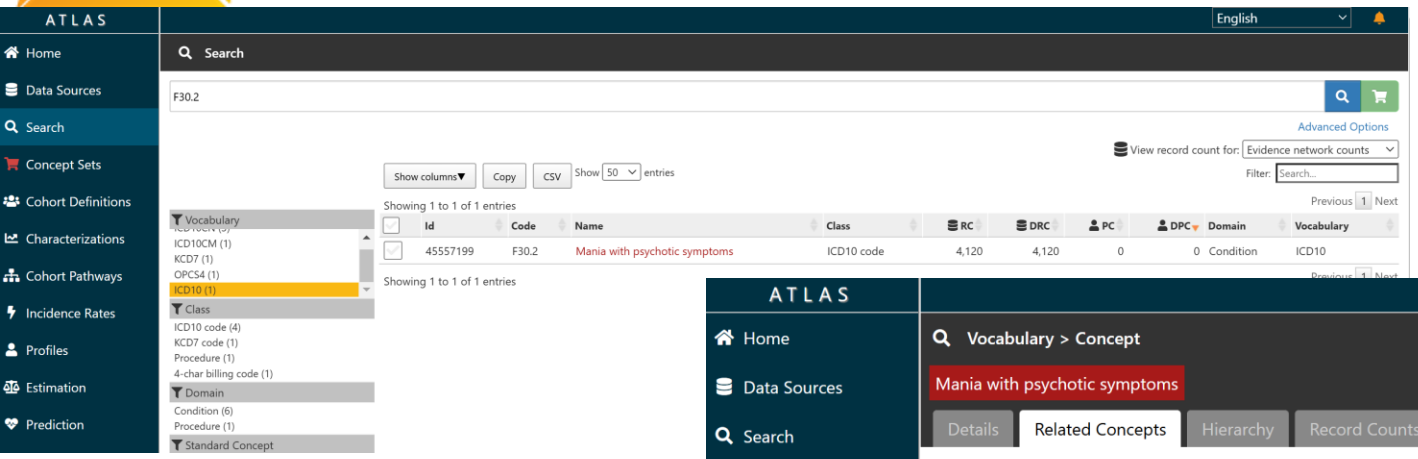
Classification Non-Standard Standard

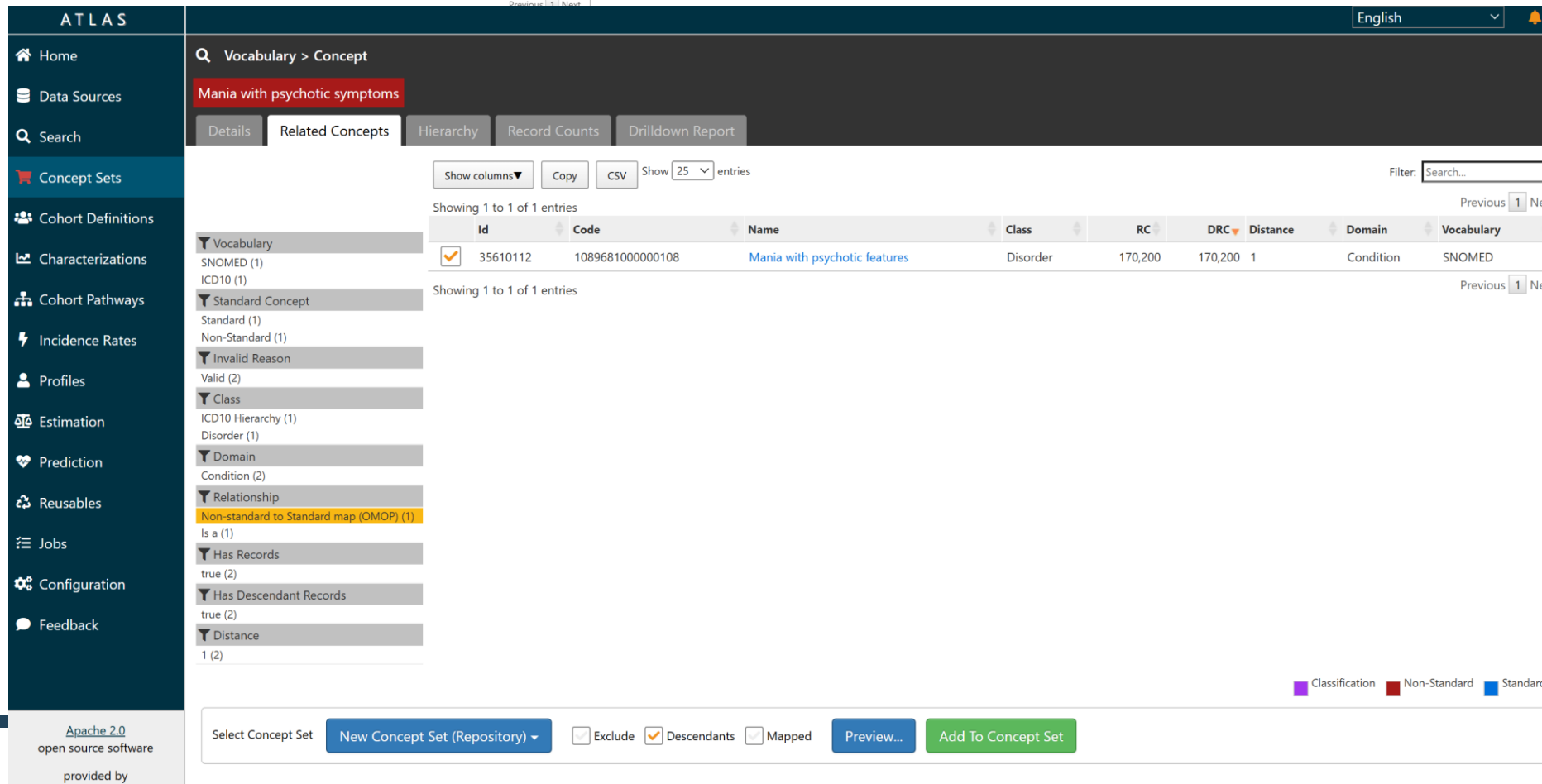
Remove Selected | Add Concepts

Checking source codes included

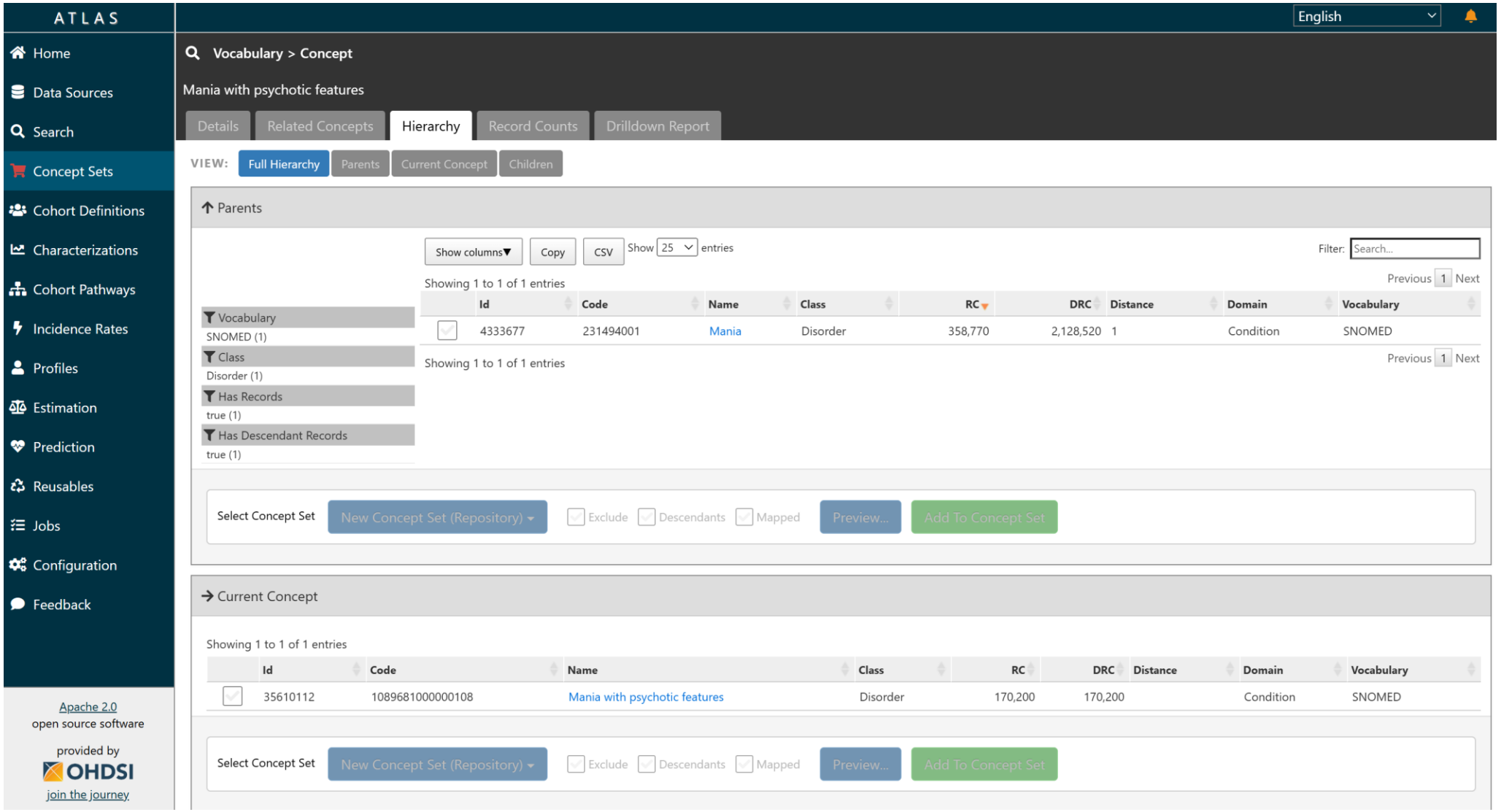


Adding other codes from the clinical description





Codes may come from different hierarchy branches



Cohort Definition creation

Inflammatory bowel disease

Kevin Haynes

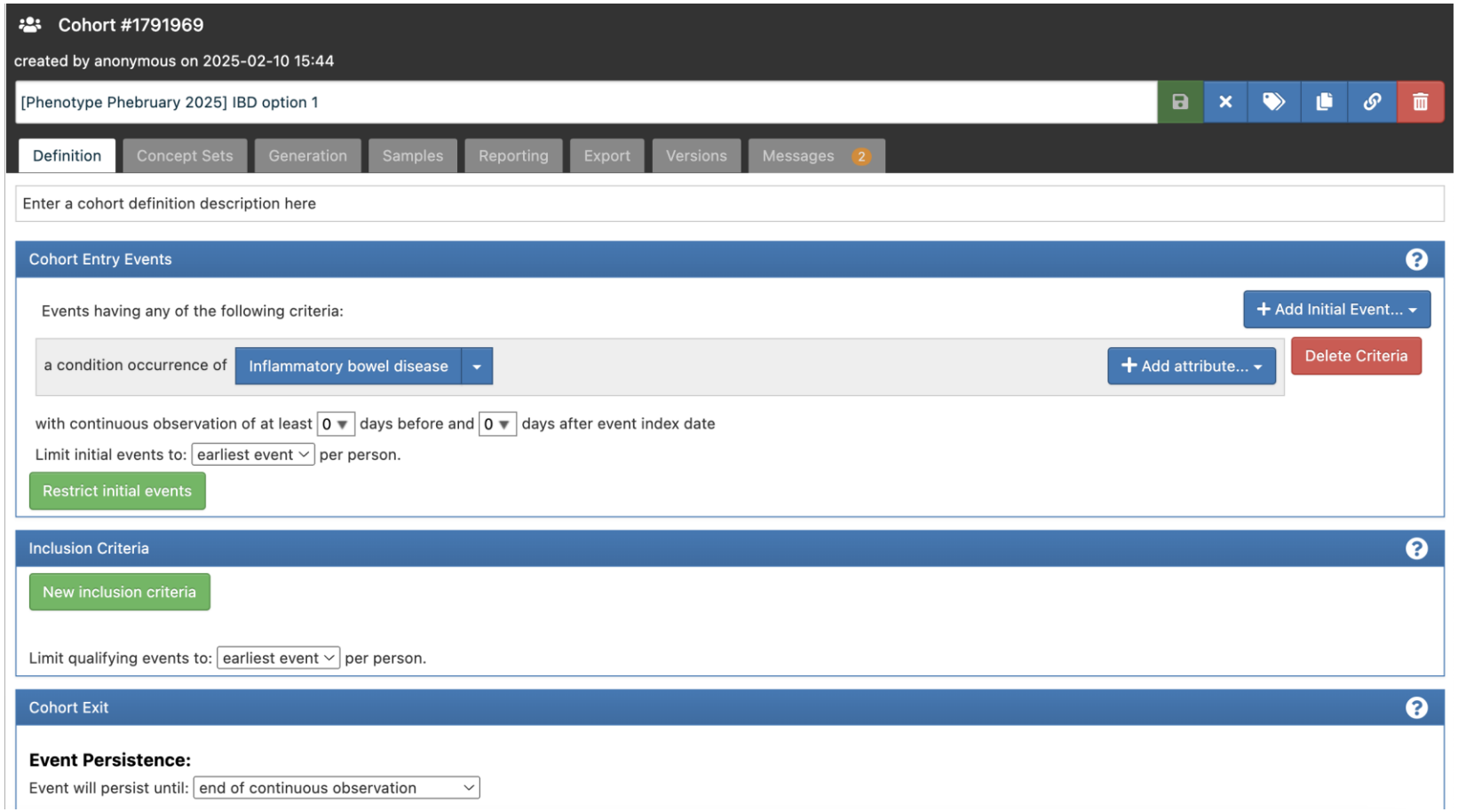
Inflammatory Bowel Disease: option 1

>=1 occurrence of an IBD code

Entry event: Condition occurrence of IBD for the first time in the person's history

Inclusion rules: -

Exit criteria: End of observation period



https://pubmed.ncbi.nlm.nih.gov/25874650/

Inflammatory Bowel Disease: option 2

Development and Validation of Claims-Based Definitions to Identify Incident and Prevalent Inflammatory Bowel Disease in Administrative Healthcare Databases

Table 1. PPV of the incident claim–based algorithms to identify the date of incidence diagnosis of IBD within 90 days of that recorded in the medical records using HIRD data.

Algorithm	Number of charts reviewed	Correctly identified incidence date	PPV (%)	95% CI (%)
High probability	57	52	91	81-97
Low probability	26	22	85	65-96
High probability with longer lag time between lower endoscopy or surgery and first IBD diagnosis	26	19	73	52-88

Abbreviations: CI, confidence interval; HIRD, HealthCore Integrated Research Database; IBD, inflammatory bowel disease; PPV, positive predictive value.

https://pmc.ncbi.nlm.nih.gov/articles/PMC10697409/

Inflammatory Bowel Disease: option 2

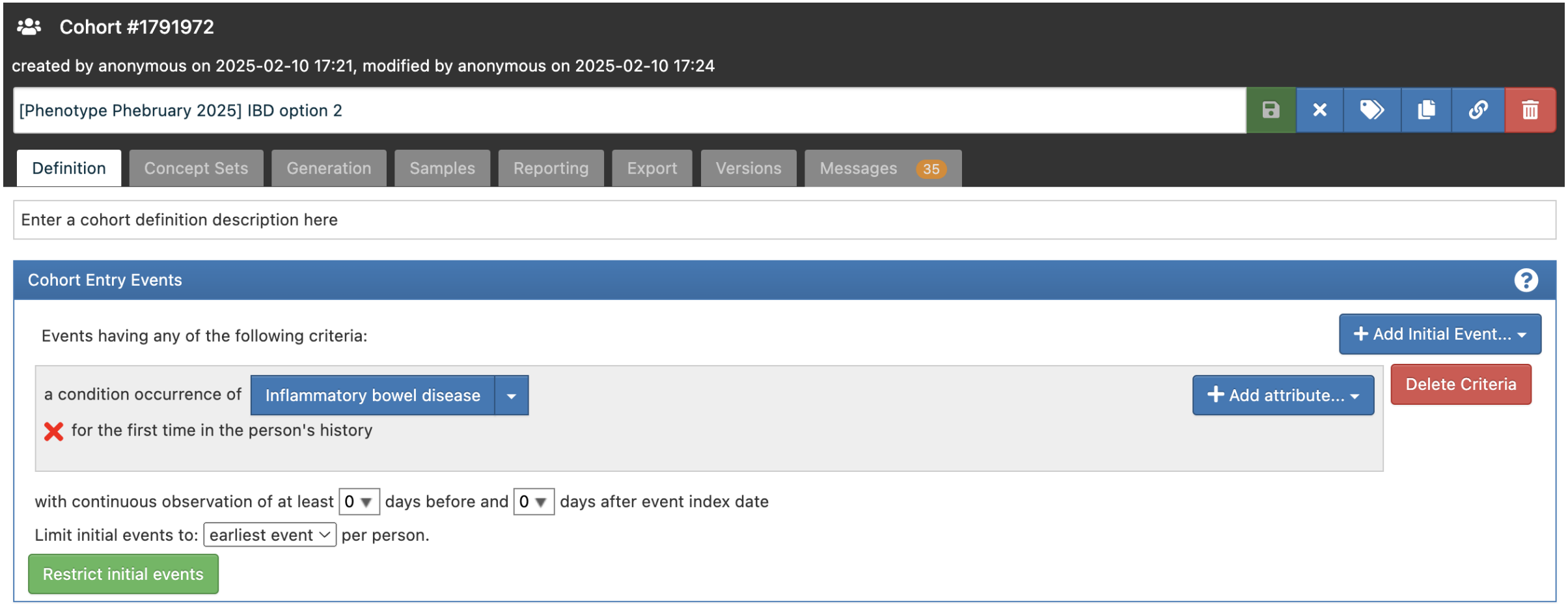
Entry event: Condition occurrence of IBD for the first time in the person's history

Inclusion rules:

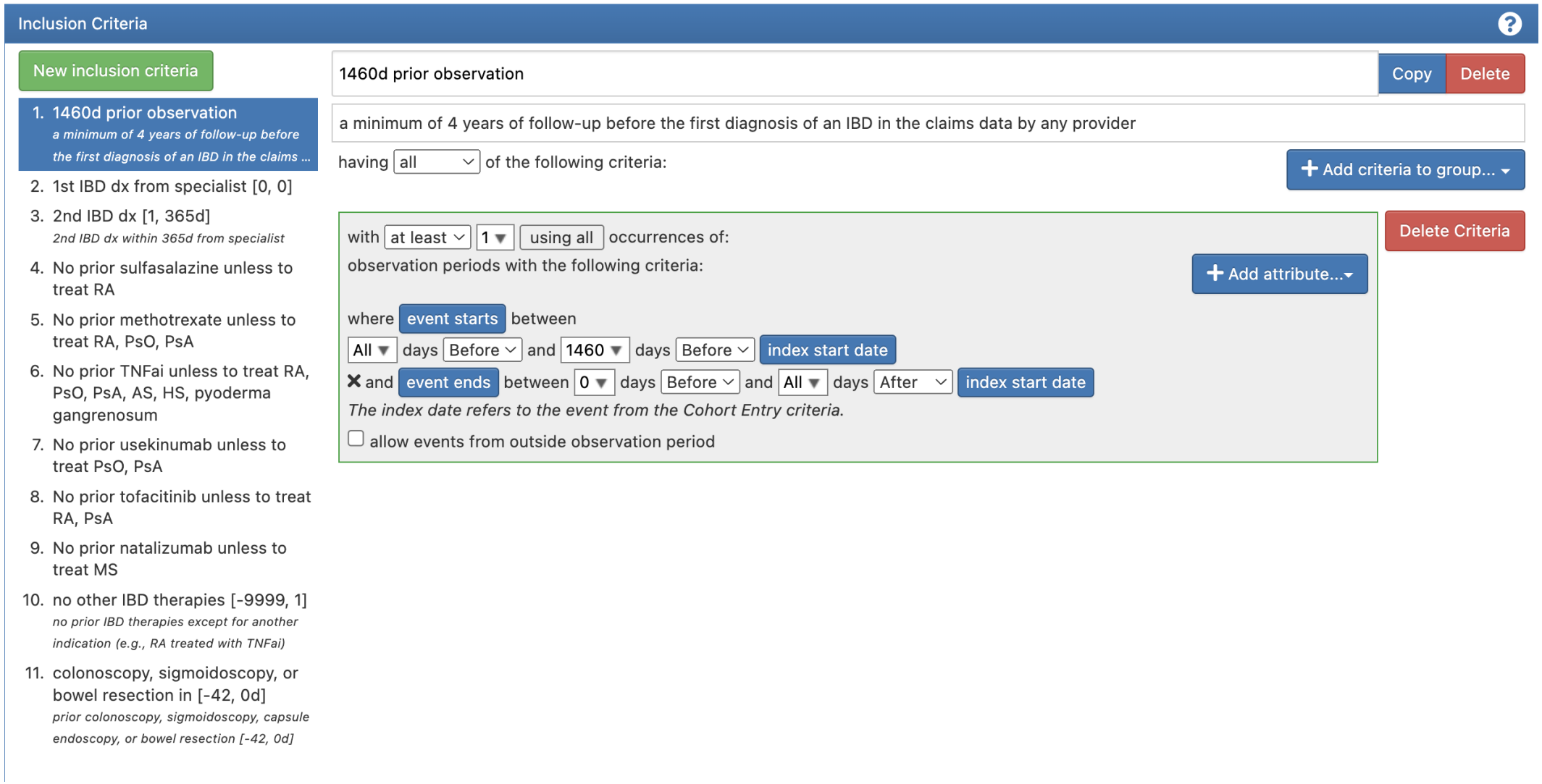
1. 1460d prior observation
2. 1st IBD dx from specialist [0, 0d]
3. 2nd IBD dx from specialist [1, 365d]
4. No prior sulfasalazine unless to treat RA
5. No prior methotrexate unless to treat RA, PsO, PsA
6. No prior TNFai unless to treat RA, PsO, PsA, AS, HS, pyoderma gangrenosum
7. No prior usekinumab unless to treat PsO, PsA
8. No prior tofacitinib unless to treat RA, PsA
9. No prior natalizumab unless to treat MS
10. No prior other IBD therapies [-9999, 1d]
11. Prior colonoscopy, sigmoidoscopy, capsule endoscopy, or bowel resection procedures [-42, 0d]

Exit criteria: End of observation period

Cohort entry



Inclusion/exclusion criteria



Inclusion/exclusion criteria

1st IBD dx from specialist [0, 0] | Copy | Delete

enter an inclusion rule description

having all of the following criteria: + Add criteria to group...

with at least 1 using all occurrences of: | Delete Criteria

a condition occurrence of Inflammatory bowel disease + Add attribute...

with a Provider Specialty of: Colorectal Surgery | Gastroenterology | Pediatric Gastroenterology | Add | Import

where event starts between

0 days Before and 0 days After index start date *add additional constraint*

The index date refers to the event from the Cohort Entry criteria.

☐ restrict to the same visit occurrence

☐ allow events from outside observation period

Inclusion/exclusion criteria

New inclusion criteria

1. 1460d prior observation
   a minimum of 4 years of follow-up before the first diagnosis of an IBD in the claims ...
2. 1st IBD dx from specialist [0, 0]
3. 2nd IBD dx [1, 365d]
   2nd IBD dx within 365d from specialist
4. No prior sulfasalazine unless to treat RA
5. No prior methotrexate unless to treat RA, PsO, PsA
6. No prior TNFai unless to treat RA, PsO, PsA, AS, HS, pyoderma gangrenosum
7. No prior usekinumab unless to treat PsO, PsA
8. No prior tofacitinib unless to treat

2nd IBD dx [1, 365d] | Copy | Delete

2nd IBD dx within 365d from specialist

having all of the following criteria: + Add criteria to group...

Delete Criteria

with at least 1 using all occurrences of:

a condition occurrence of Inflammatory bowel disease + Add attribute...

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1 days After and 365 days After index start date *add additional constraint*

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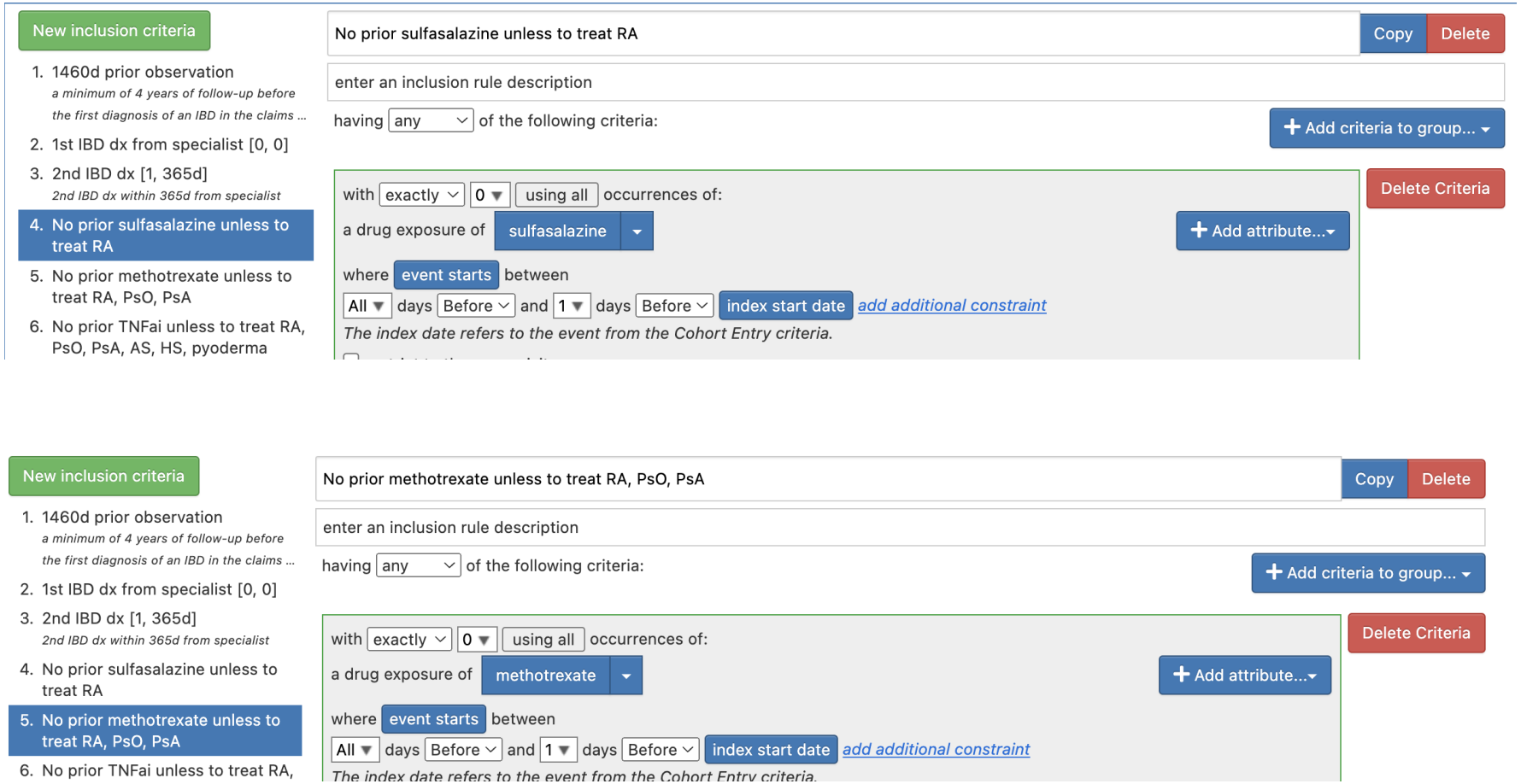
Inclusion/exclusion criteria



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7. No prior usekinumab unless to treat PsO, PsA
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2. 1st IBD dx from specialist [0, 0]
3. 2nd IBD dx [1, 365d]
   2nd IBD dx within 365d from specialist
4. No prior sulfasalazine unless to treat RA
5. No prior methotrexate unless to treat RA, PsO, PsA
6. No prior TNFai unless to treat RA, PsO, PsA, AS, HS, pyoderma gangrenosum
7. No prior usekinumab unless to treat PsO, PsA
8. No prior tofacitinib unless to treat RA, PsA
9. No prior natalizumab unless to treat MS
10. no other IBD therapies [-9999, 1]
    no prior IBD therapies except for another indication (e.g., RA treated with TNFai)
11. **colonoscopy, sigmoidoscopy, or bowel resection in [-42, 0d]**
    prior colonoscopy, sigmoidoscopy, capsule endoscopy, or bowel resection [-42, 0d]

colonoscopy, sigmoidoscopy, or bowel resection in [-42, 0d] — Copy — Delete

prior colonoscopy, sigmoidoscopy, capsule endoscopy, or bowel resection [-42, 0d]

having any of the following criteria: + Add criteria to group...

with at least 1 using all occurrences of: — Delete Criteria

a procedure occurrence of Colonoscopy, sigmoidoscopy,... + Add attribute...

where event starts between

42 days Before and 0 days After index start date *add additional constraint*

The index date refers to the event from the Cohort Entry criteria.

☐ restrict to the same visit occurrence

☐ allow events from outside observation period

or with at least 1 using all occurrences of: — Delete Criteria

an observation of Colonoscopy, sigmoidoscopy,... + Add attribute...

where event starts between

42 days Before and 0 days After index start date *add additional constraint*

The index date refers to the event from the Cohort Entry criteria.

☐ restrict to the same visit occurrence

☐ allow events from outside observation period

or with at least 1 using all occurrences of: — Delete Criteria

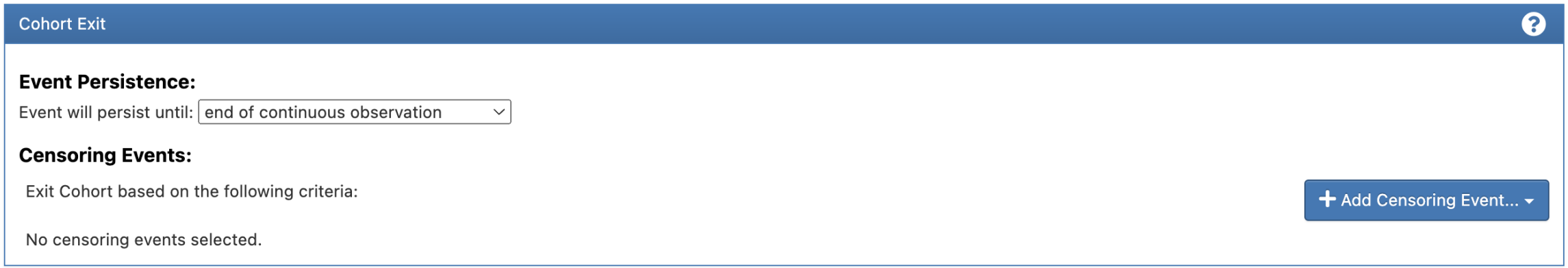
a procedure occurrence of Bowel resection procedures + Add attribute...

where event starts between

42 days Before and 0 days After index start date *add additional constraint*

The index date refers to the event from the Cohort Entry criteria.

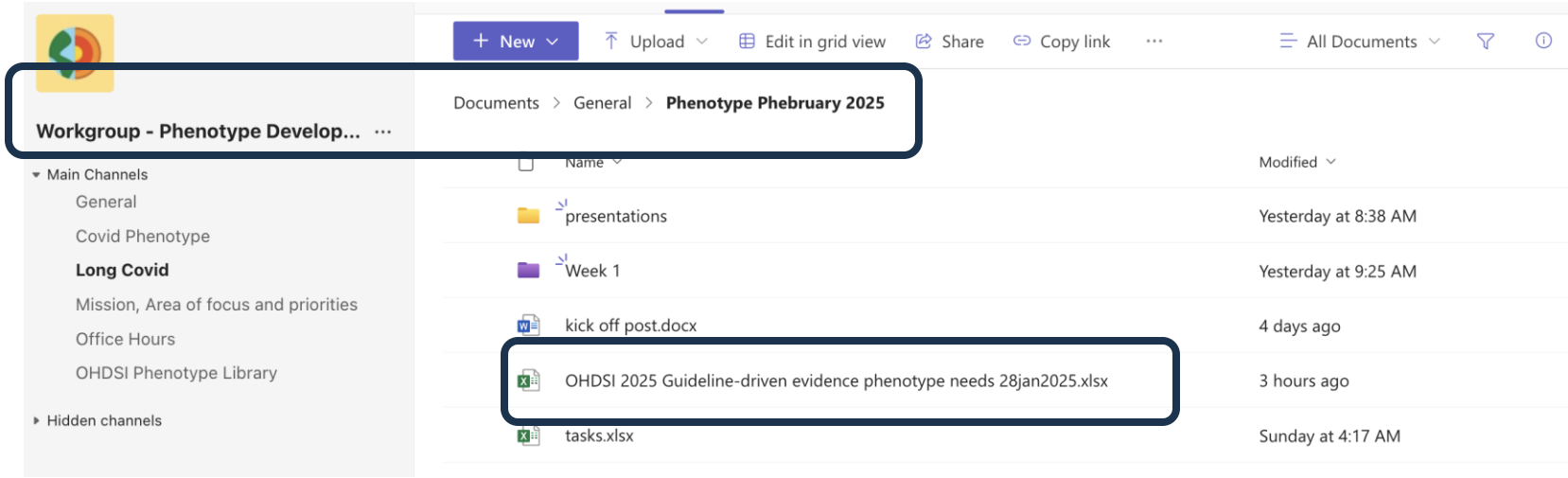
Cohort exit


Cohort Exit
Event Persistence:
Event will persist until: end of continuous observation
Censoring Events:
Exit Cohort based on the following criteria:
+ Add Censoring Event...
No censoring events selected.

Next steps

Study leads

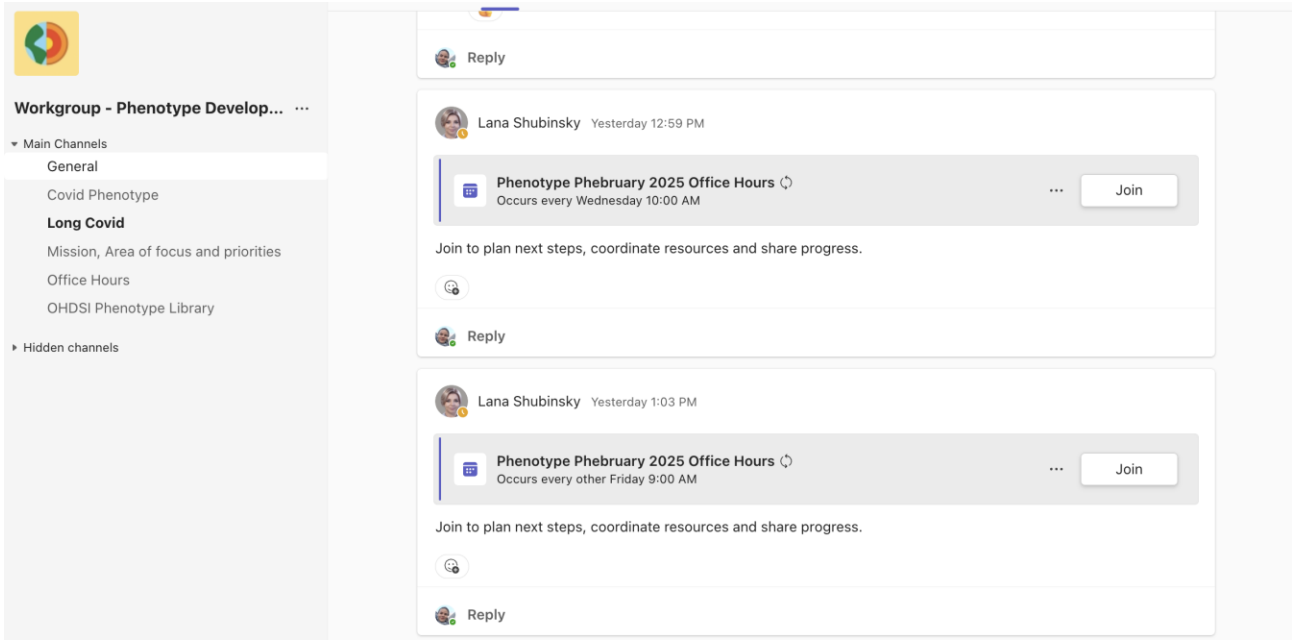
1. Together with the interested participant (tagged in channels and in emails) create the first draft of the cohorts. By Friday Feb 14th EOB use the spreadsheet to indicate which cohorts are ready for us to run Cohort Diagnostics on.
2. Come to the office hours on Wednesday 10am EST
   Friday 9am EST



	A	B	F	G	H	I	J	
1	Study	Lead	Clinical	Cohort definition	CohortDiag	Iteration		Cohort id in Atlas

Next steps

Anybody who wants to participate

1. If you have not already, reach out to the study leads or us to participate in cohort building.

2. Come to the office hours on Wednesday 10am EST
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