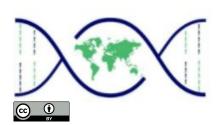




# IHCC Cohort Atlas Workshop

Thomas Keane, EMBL-EBI







# **Agenda for today**

Time (BST)	Subject	Presenter		
16:00 - 16:10	IHCC project and data team overview (10')	Thomas Keane (EMBL-EBI)		
16:10 - 16:20	Atlas overview (10') Brandon Chan (OICR			
16:20 - 16:35	Atlas Data harmonization (15')	Melanie Courtot (EMBL-EBI)		
16:50 - 18:00	Live demo (including processing steps)	Carles Garcia, Isuru Liyanage (EMBL-EBI)		



#### **Data and Infrastructure Team**

"Deliver <u>interoperable cross cohort infrastructure</u> to enable population scale biomolecular <u>data to be accessible across international borders</u> accelerating research and improving the health of individuals resident across continents."



#### **Challenges**

Challenge 1: Federated cohort data discovery

Challenge 2: Harmonized cohort level metadata

Challenge 3: Cohort access and authorization

Challenge 4: Federated analysis interoperability for research



#### **Challenges**

Challenge 1: Federated cohort data discovery

Challenge 2: Harmonized cohort level metadata

Challenge 3: Cohort access and authorization

Challenge 4: Federated analysis interoperability for research



#### **Global Standards**



Cohort interoperability standards are emerging

Global Alliance for Genomics and Health (GA4GH)

- 8 workstreams (e.g. Discovery, Data Use, Clinical and phenotypes, Ethics, Security)
- Clinical metadata standards (e.g. HL7/FIHR, OMOP etc)

# 2000+ Subscribers 8 8 500+ Organisational Members 70+ Countries

#### IHCC Data team and GA4GH

- Foundation for all IHCC products to interop with other cohorts
- e.g. IHCC cohort atlas cross queries with other aggregate resources



#### **Global Standards**



Cohort interoperability standards are emerging

Global Alliance for Genomics and Health (GA4GH)

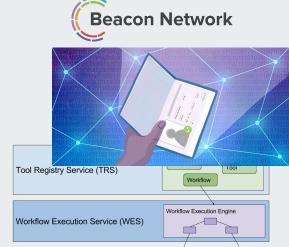
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#### IHCC Data team and GA4GH

- Foundation for all IHCC products to interop with other cohorts
- e.g. IHCC cohort atlas cross queries with other aggregate resources









#### **Progress to date**

#### **IHCC Cohort Atlas**

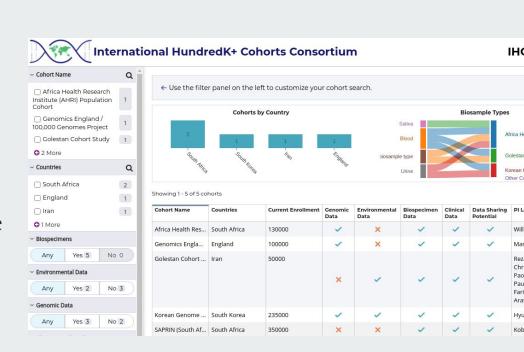
- Enables researchers to search across IHCC cohort metadata
- 13 cohorts, >100 variables harmonised

#### Davos Alzheimer's Collaborative

Example of disease specific expansion of the atlas

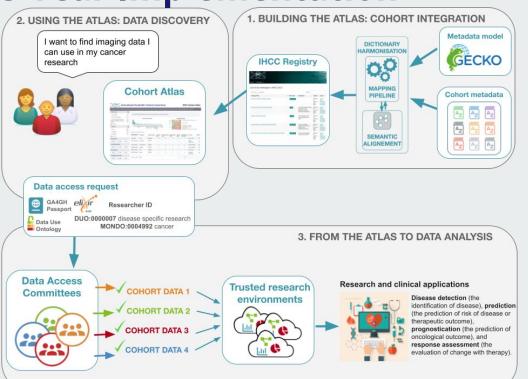
#### Future vision for atlas

• Cornerstone of future IHCC data science platform





#### **5 Year Implementation**

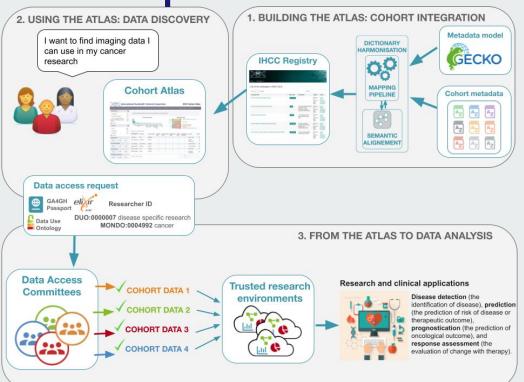


#### 1. IHCC Atlas Content

- Expansion of cohort content
- Expansion of the harmonised metadata model
- Standardisation of core cohort descriptors
- Data model alignment with other cross cohort resources
- New modes of discovery, e.g. data use



#### **5 Year Implementation**

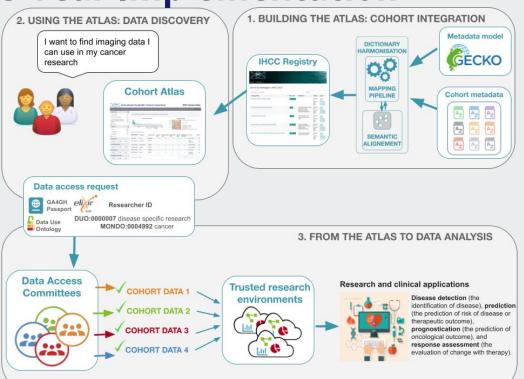


#### 2. Using the Atlas

- Complex interactive queries
- Integration with discovery networks, e.g. Beacon network
- FAIR cohort data access
- Integration of cohort data access processes, e.g. GA4GH researcher Passports



#### **5 Year Implementation**



- 3. Federated cohort analysis
  - Federated cloud analysis model for cohorts, e.g. AoU, UKB, AD workbench
  - Discover, access, analysis
  - Atlas integration with cohort platforms
  - Exemplar for deployment of GA4GH standards



# **Agenda for today**

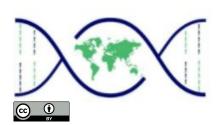
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# IHCC Cohort Atlas Overview

Melanie Courtot / Brandon Chan







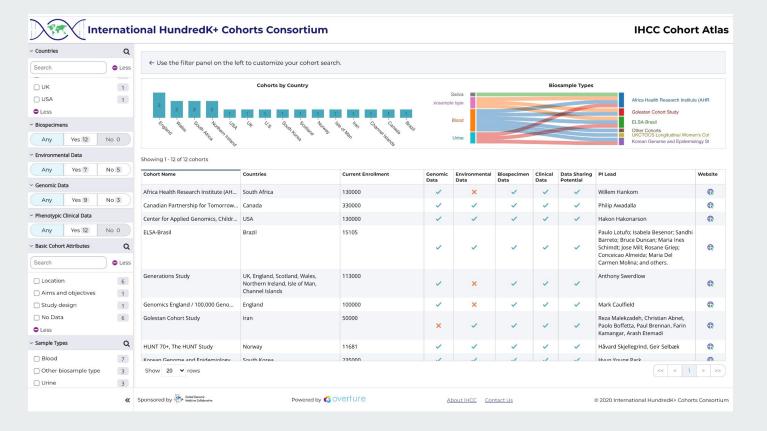
#### **IHCC Cohort Atlas Overview**

- The IHCC Cohort Atlas fits directly into the overall IHCC vision and goal to bring large cohorts together to encourage data sharing, improve efficiencies and maximize benefits in addressing scientific questions no party can answer alone.
- The Atlas Browser provides a simple-to-use, easy-to-access UI for researchers to quickly search for a cohort of interest.
- Available cohorts and their metadata are first consolidated and harmonized before being published to the Browser.
- Users can then further explore those cohorts of their own accord.
- Acts as a gateway or portal for users to access their cohorts of interest and pursue further detailed, specific exploration on each cohort.



#### **IHCC Cohort Atlas Browser**

#### https://ihccglobal.org/







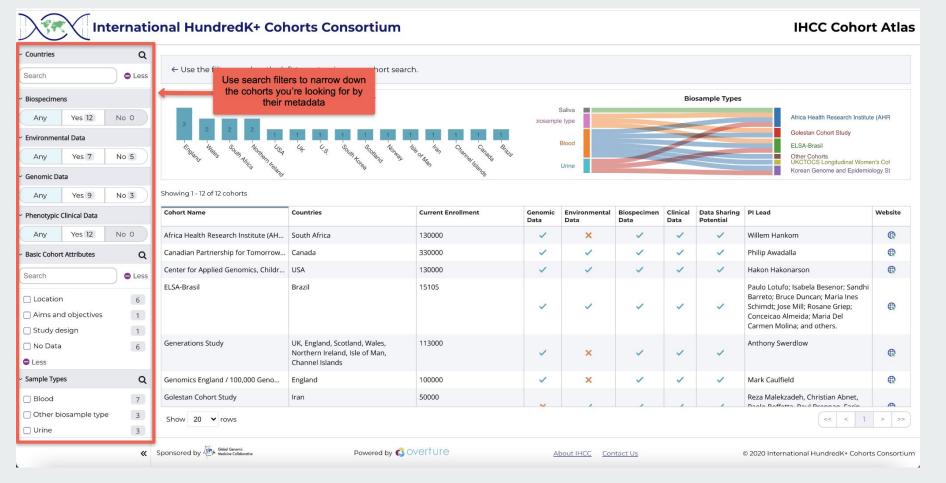






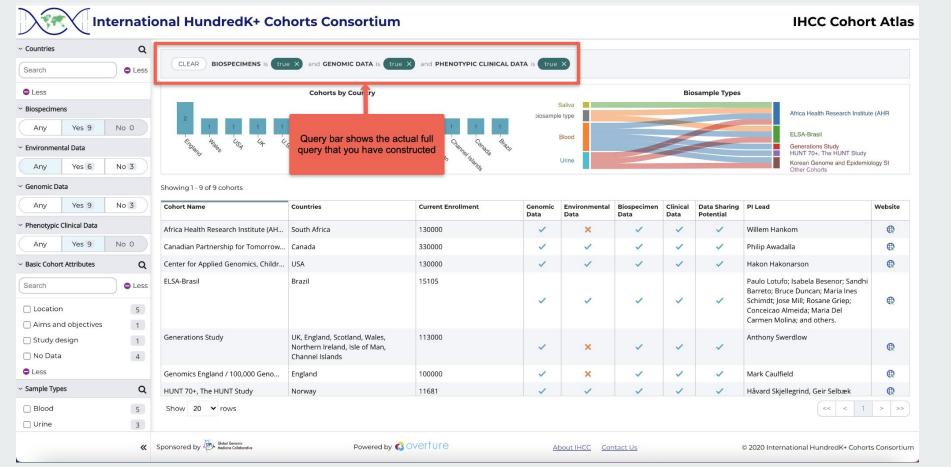


#### **Search & Filter Controls**



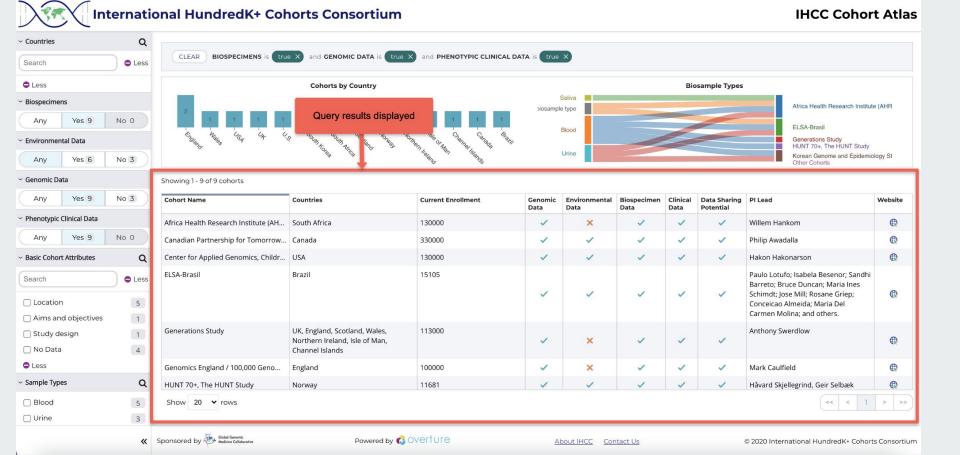


#### **Query Bar**



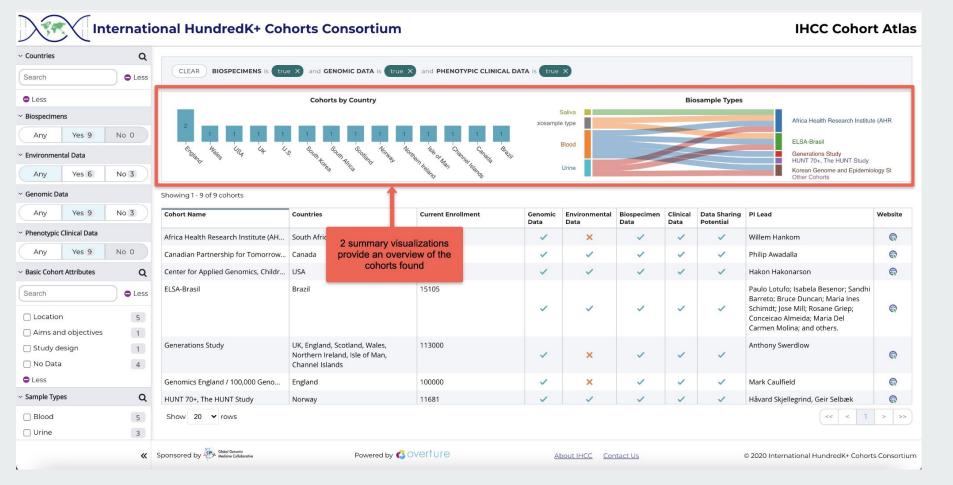


#### **Query Results**



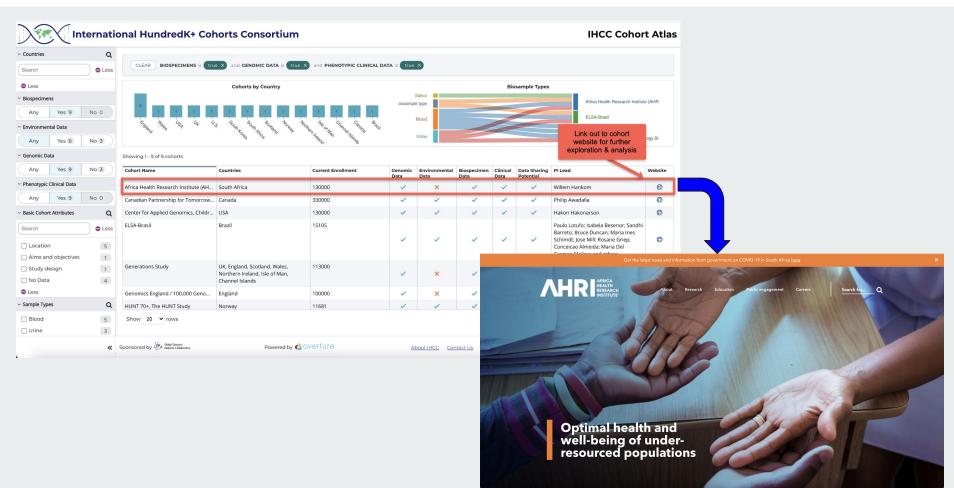


#### **Cohort Summary Visualizations**





#### **Link Out to Cohorts for Further Exploration**





#### **Atlas Browser Underlying Technology**

#### https://www.overture.bio/

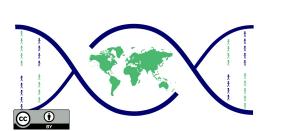






# Data harmonisation

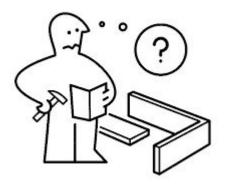
Mélanie Courtot EMBL-EBI





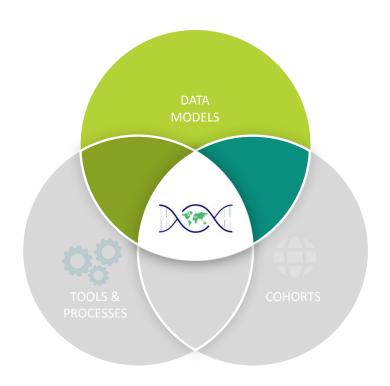
#### **Building a common framework**





- 1. **Data models** to represent both access conditions and cohort data
- 2. **Tools** and processes for implementations
- 3. Deployment over clinical cohorts







#### **Genomics Cohort Knowledge Ontology (GECKO)**

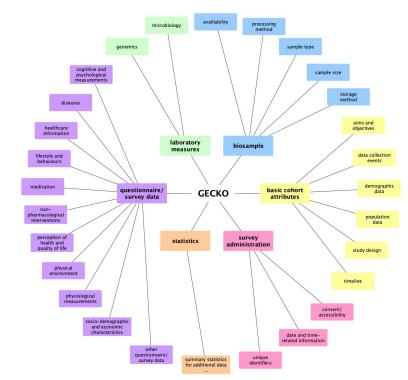
- Commonly used attributes to describe cohort metadata
- "Medication", "sample type", "genomics datatypes"...



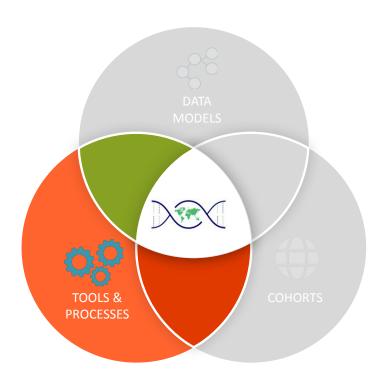








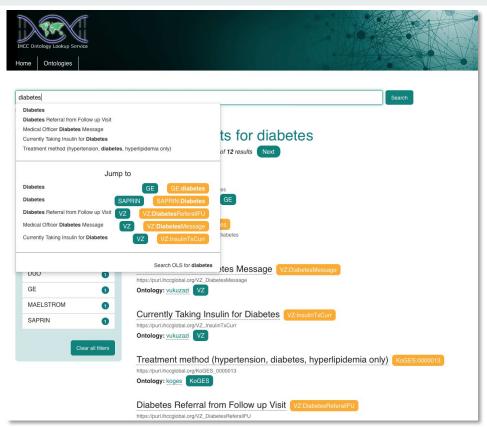




## Registry and mapping

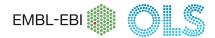
#### **IHCC** cohort registry

- Human readability of cohort dictionaries
- Version and change detection for update
- Built on the EMBL-EBI Ontology Lookup Service platform



https://registry.ihccglobal.app/index

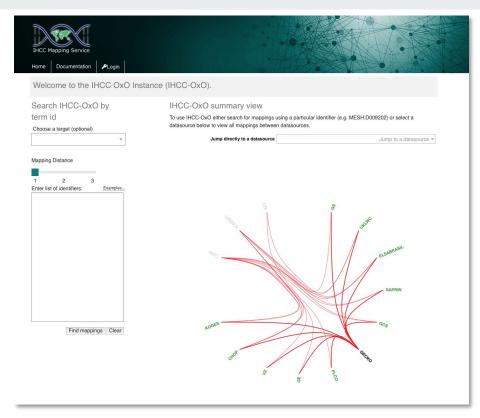






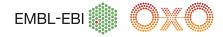
#### **IHCC** cohort mappings

- Stores mapping between GECKO and cohort terms
- Accessible through APIs
- Parameter to bridge between mappings If A ⇔ B and B ⇔ C then can infer A⇔ C



https://mapping.ihccglobal.app

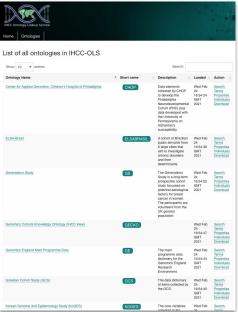




## **Automated mapping pipeline for cohort**

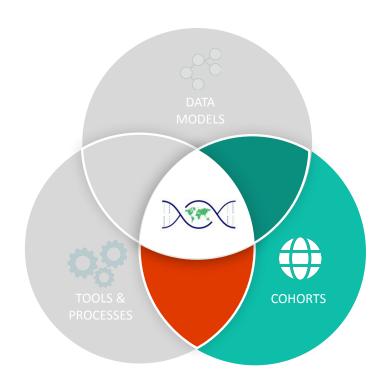
**owners** 



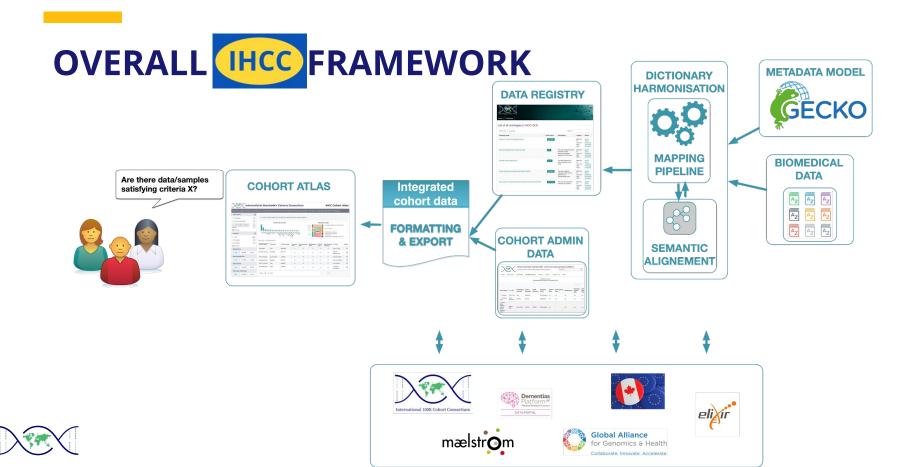


**IHCC** cohort registry



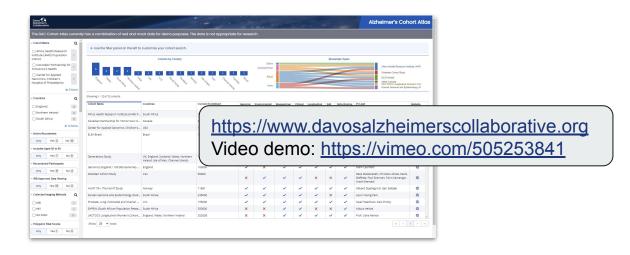


Applying these techniques to clinical cohorts...



#### **Next steps**

Pipeline can be reused



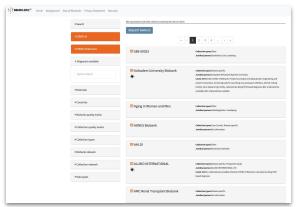


#### **Next steps**

# Pipeline can be reused

Model can be extended







Morris Swertz

https://directory.bbmri-eric.eu/



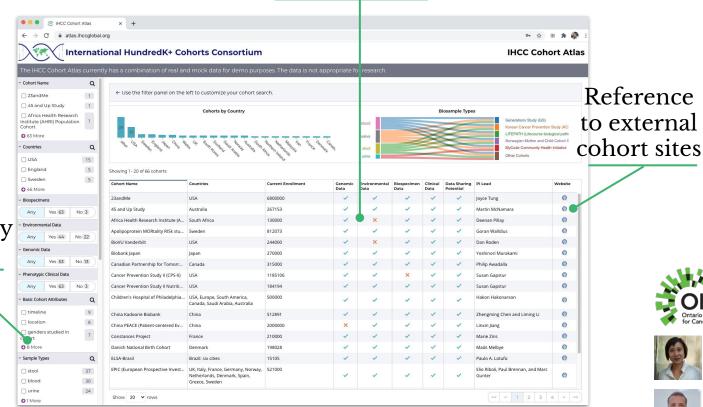


#### IHCC cohort atlas

#### Cohort presentation and display



https://ihccglobal.org











Philip Awadalla October 27th, 2021



# Cohort Data Processing as a User Perspective

Carles Garcia Linares EMBL-EBI





I would like to register & map my data dictionary



# The IHCC data harmonization project: Documentation and Instructions

Documentation and Instructions for the International HundredK+ Cohorts Consortium (IHCC) data harmonization project

How to register your Data Dictionary with the International HundredK+ Cohorts Consortium (IHCC)

Email of the IHCC data harmonization team: ihcc-browser@googlegroups.com

#### Initial submission

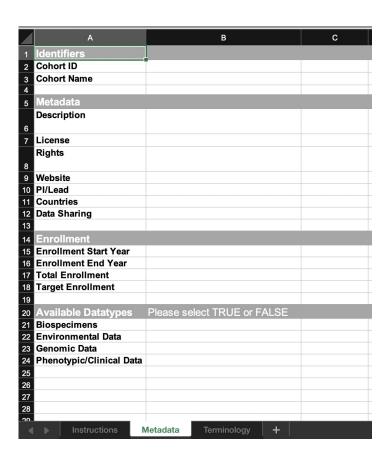
- Download the data dictionary registration template. It is an Excel Workbook in XLSX format, containing the following three sheets:
  - 1. Instructions. Please read this table carefully before you continue.
  - Metadata. Contains basic metadata about your data dictionary, such as name, description and license.
  - 3. Terminology. Contains the terms of your data dictionary, alongside a few optional columns.
- 2. Follow the instructions in the Instructions sheet.

https://ihcc-cohorts.github.io/DataDictionaryMapping.html



A	В	С	D	Е	F	G		
IHCC Data Harmonization								
Please fill the Metadata and Terminology sheets to the best of your ability.								
Email to	ihcc-browser@	<u>googlegroups.com</u>						
Metadata								
Cohort ID	Required	The short identifier for the cohort. Must be one wo	rd, uppercase, and u	inique among the II	HCC cohorts			
Cohort Name	Required	The title of the cohort.						
Description	Required	The description of the cohort, in one or two senter	nces.					
License	Required	The link to the license of the cohort data dictionar	y.					
Rights	Optional	An optional text description of the usage rights for	the cohort's data did	ctionary.				
Website	Optional	A link to the cohort website.						
2 PI/Lead	Required	The cohort PI/Lead (comma-separated for more th	nan one lead).					
Countries	Required	Country(ies) in which the cohort participants are le	ocated (comma-sepa	arated).				
Data Sharing	Required	TRUE/FALSE value for if the cohort has IRB appr	roval for data sharing	<b>j</b> .				
Enrollment Data	Required*	Enrollment years and numbers for the cohort. Enr	ollment start year an	d total enrollment va	alues are required,	other values are o	ption	
Available Datatypes	Required	TRUE/FALSE values for the types of data collected	ed by the cohort.					
7								
Terminology		One row for each data dictionary term.						
Label	Required	The label of your term.						
Parent Term	Optional	The parent term or category: this is optional, but if	filled, the value mus	at be the label of and	other term in this she	eet.		
Definition	Optional	The definition of your term.						
Internal ID	Optional	If your term has an internal id, like a database ide	your term has an internal id, like a database identifier, you can add it here.					
Comment	Optional	Other comments for the IHCC data harmonization	team.					
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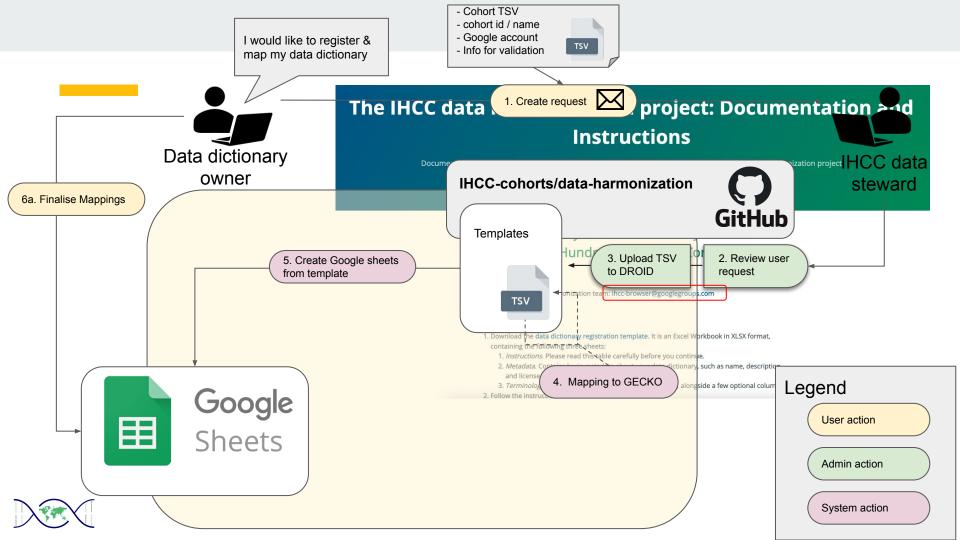


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2 Nationality		The country of origin of the inc	dividual (the country of which	the person is/was a citizen by birth)
3 Surname		Family or last name		
4 First Name		The first given name of the inc	dividual	
5 Sex		Male (1), female(2) or unknow	vn(0)	
6 Birth Date		Date of Birth		
7 Employment Type		Employed, Unemployed, Part	time, Retired	
8 Birth Weight		Birth weight in grams		
9 Blood cell count		Complete blood cell count		
10 Glucose		Fasting glucose level		
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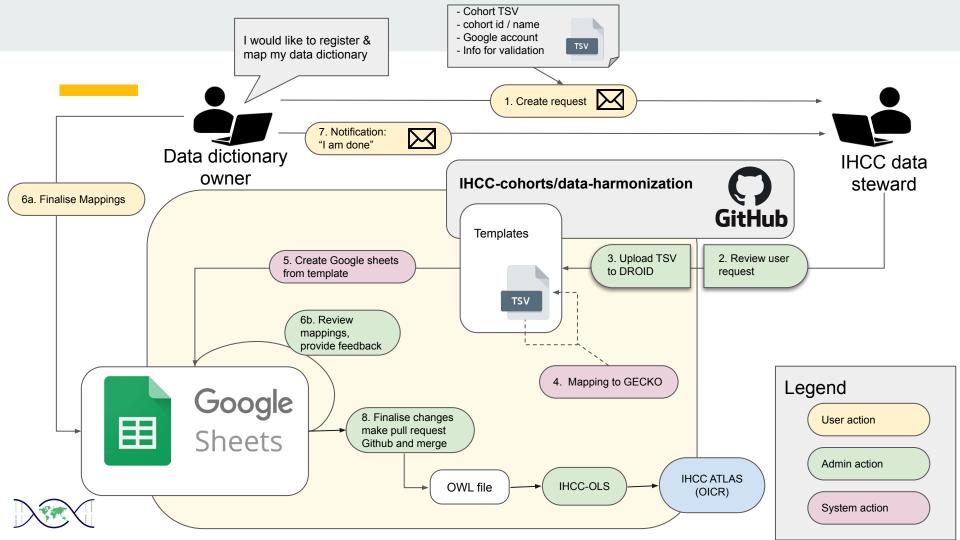


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3	Surname		Family or last name		
4	First Name		The first given name of the ir	ndividual	
5	Sex		Male (1), female(2) or unkno	wn(0)	
6	Birth Date		Date of Birth		
7	Employment Type		Employed, Unemployed, Par	t time, Retired	
8	Birth Weight		Birth weight in grams		
9	Blood cell count		Complete blood cell count		
10	Glucose		Fasting glucose level		
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3	COA:0000003	Surname		Family or last na	survey admir 🔻		1.00 GECKO:00	00052 survey	administration   0.1	6 MONDO:000508	
4	COA:0000004	First Name		The first given n	survey admir		1.00 GECKO:00	00052 survey	administration   0.1	4 GECKO:0000093	
5	COA:0000005	Sex		Male (1), female	biological sex		1.00 PATO:0000	0047 biological	sex   0.16 MONDO	0:0005084 mental a	
6	COA:0000006	Birth Date		Date of Birth	age/birthdate		1.00 PATO:0000	011 age/birthd	ate   0.90 STATO:0	000093 date and t	
7	COA:0000007	Employment Typ	pe	Employed, Uner	socio-demog		1.00 PATO:0001	1995 socio-dem	ographic and ecor	omic characteristic	
8	COA:0000008	Birth Weight		Birth weight in g	reproduction 🔻		1.00 GECKO:00	00114 reprodu	ction   0.37 CMO:0	000012 weight	
9	COA:0000009	Blood cell count		Complete blood	blood		1.00 UBERON:0	0000178 blood	0.14 OGMS:0000	020 signs and sym	
10	COA:0000010	Glucose		Fasting glucose	urine		1.00 UBERON:0	0001088 urine	0.40 UBERON:00	00178 blood   0.28	
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#### Contact us! ihcc-browser@googlegroups.com

### **Acknowledgements**



**Thomas** Keane



Philip Awadalla



Christina Yung



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Melanie Courtot



Eric Plummer



Minh Ha



Brandon Chan



Carles Garcia



Isuru Liyanage



Dan Brake



Chris Lunt



Knocean

James Overton Rebecca Jackson Nicolas Matentzoglu











