



**11AM**

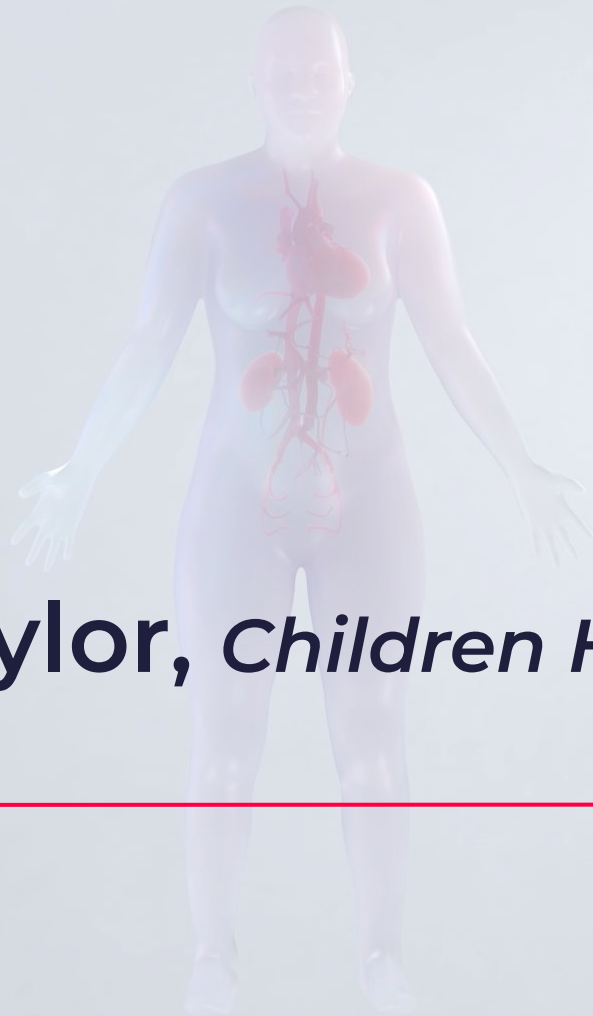
**4PM in London (GMT), 1AM in Tokyo (GMT+9)**

**FAIR & Scalable Infrastructures**

**Moderator:** Katy Börner, *Indiana University*

**Presenters:**

- Deanne Taylor, *Children's Hospital of Philadelphia*
- Ino de Bruijn, *Memorial Sloan Kettering Cancer Center*
- Bruce W. Herr II, *Indiana University*
- Jonathan Silverstein, *University of Pittsburgh*



# **Deanne Taylor, *Children Hospital of Philadelphia***

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# From Genes to Systems: Modeling Human Functional Biology with Knowledge Graphs

Deanne Taylor  
15 Dec 2024



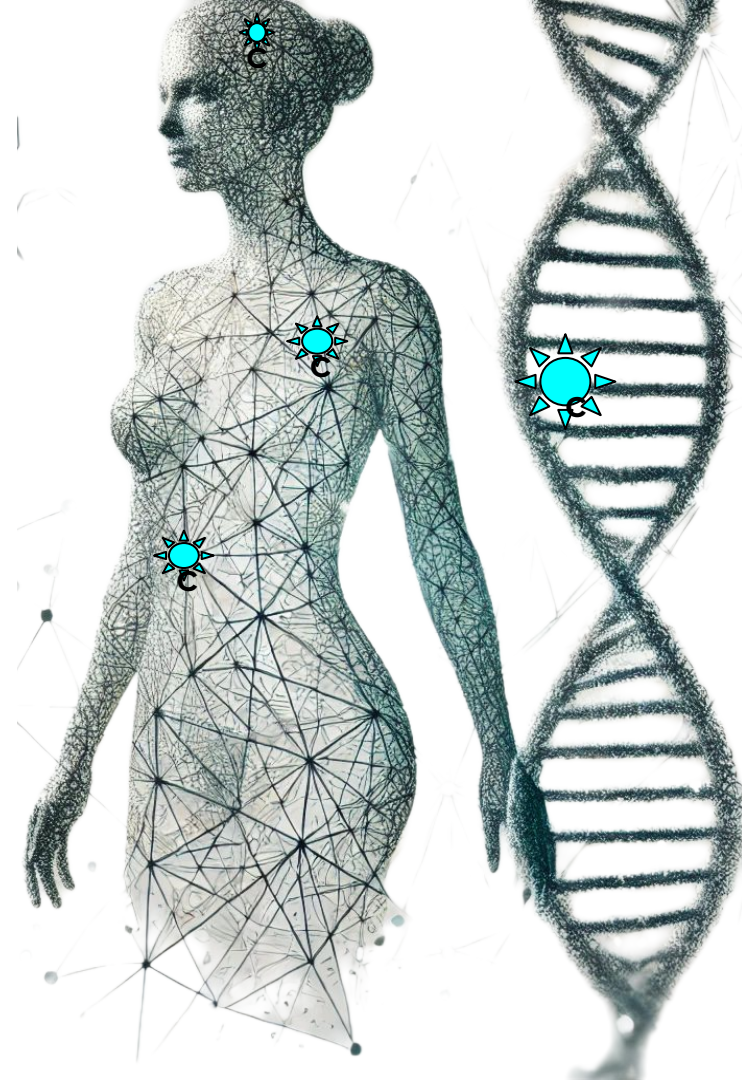
# Big Questions

How are gene variations and disorders/diseases related?

How do we predict a therapy given a gene variation?

**Reachable question:**

Can we interconnect large biomedical datasets and use those structures to answer complex biological questions linking gene variations to diseases, and potential therapeutics?



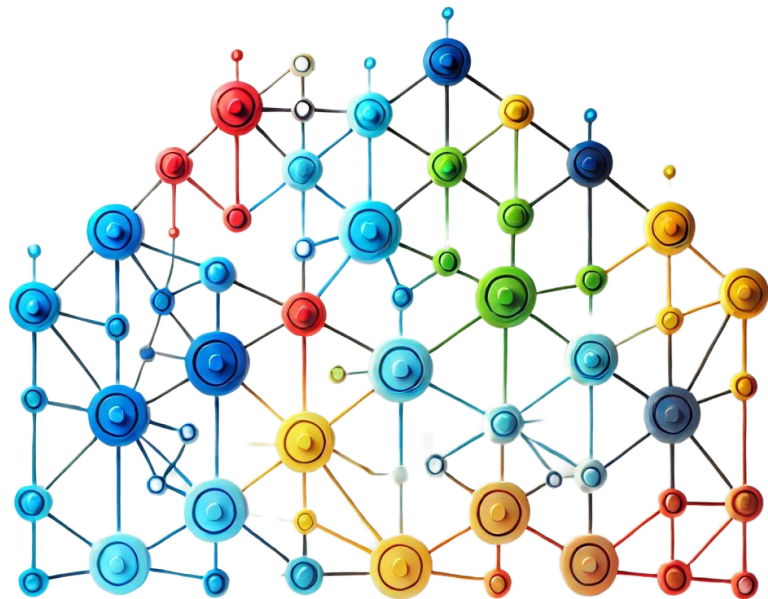
# Introduction to Knowledge Graphs (KGs)

KGs represent information as networks of nodes (entities) and edges (relationships).

For data models of the human at all scales:

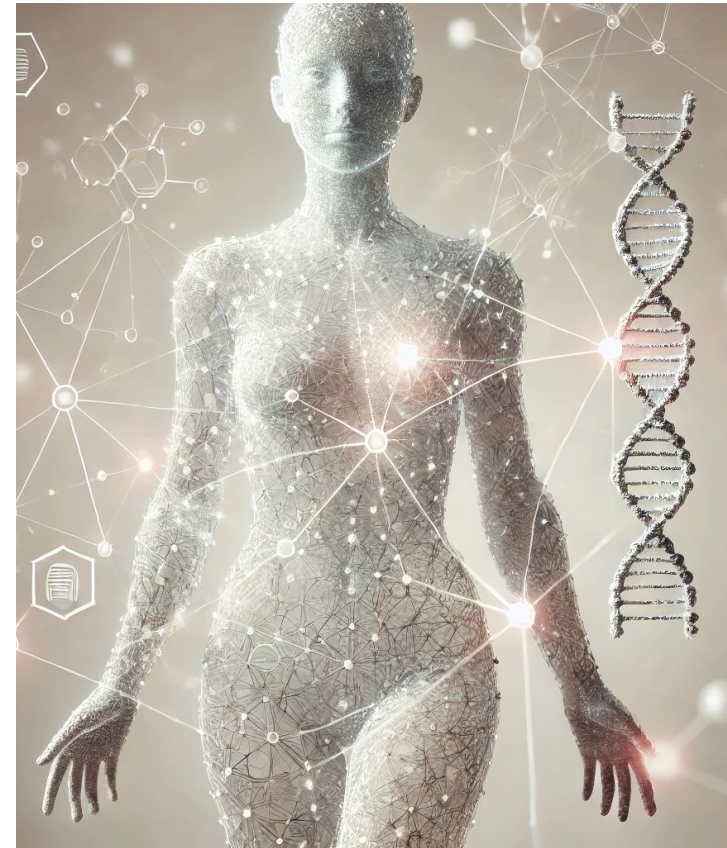
- **Node Types:** Molecules, Genes, Proteins, Diseases, Tissues, Organs, Treatments...
- **Edge Types** "Encodes," "Regulates," "Is associated with," "Treats" ...

**KGs are ideal** for integrating diverse biomedical datasets because they preserve connections and context.



# Functional Biology represented in Knowledge Graphs

- Functional biology focuses on how genes and molecular processes drive development and functioning of cells, tissues, and organs.
- KGs can provide an integrated "functional biology" layer to use in concert with anatomical maps/KGs like the Human Reference Atlas (see Bruce Herr's talk coming up!)
- This integration can also give us tools to explore how genetic variations influence human development, disorders, and disease.



# Knowledge Graphs as connected data warehouses

## UBKG

The **Unified Biomedical Knowledge Graph (UBKG)** is a knowledge graph infrastructure that represents a set of interrelated concepts from biomedical ontologies and vocabularies (Jonathan Silverstein, HuBMAP: <https://ubkg.docs.xconsortia.org/>)

## Petagraph

- Built with UBKG to integrate over 20 large multi-omics human and mouse genomic datasets
- Incorporates 180+ ontologies and annotation resources like STRING, Reactome, and GENCODE.
- Focused on harmonizing omics data for rapid feature selection and hypothesis generation.
- 32 million nodes and 118 million relationships

## Data Distillery Knowledge Graph (DDKG)

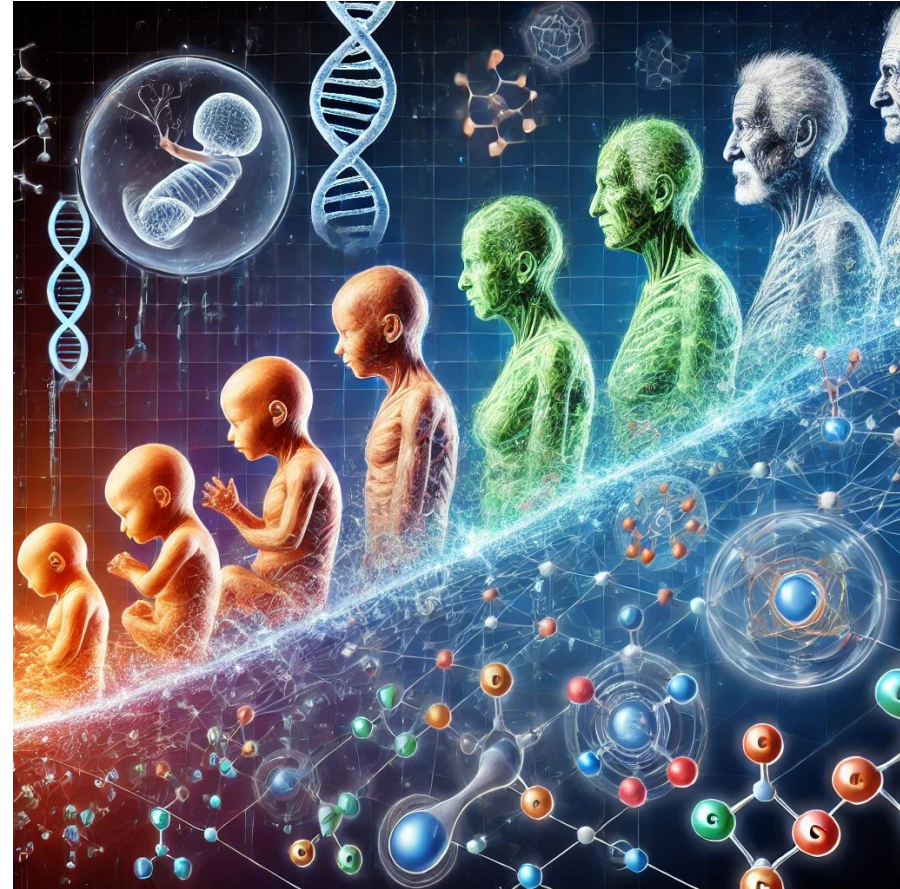
- Extends Petagraph's schema to integrate over 30 large genomics datasets in collaboration with NIH Common Fund DCCs (e.g., HuBMAP, GTEx, 4D Nucleome, LINCS...)
- Like Petagraph, is scalable for new genomics data types and cross-dataset analyses.
- Includes development of user-friendly interfaces (API, UI) for secure and efficient querying
- 40 million nodes and 300 million relationships

# The future of modeling: KGs can enable a multi-dimensional human model

Precision medicine would benefit from knowledge graphs to create whole-human models from:

- Maps of human organs and tissues across human development (4D)
- Functional biology genomics data from single cells to whole body physiology
- Human genetics data
- Observational medical data
- Dynamic biomedical process models

... towards prediction and treatments of diseases and disorders.



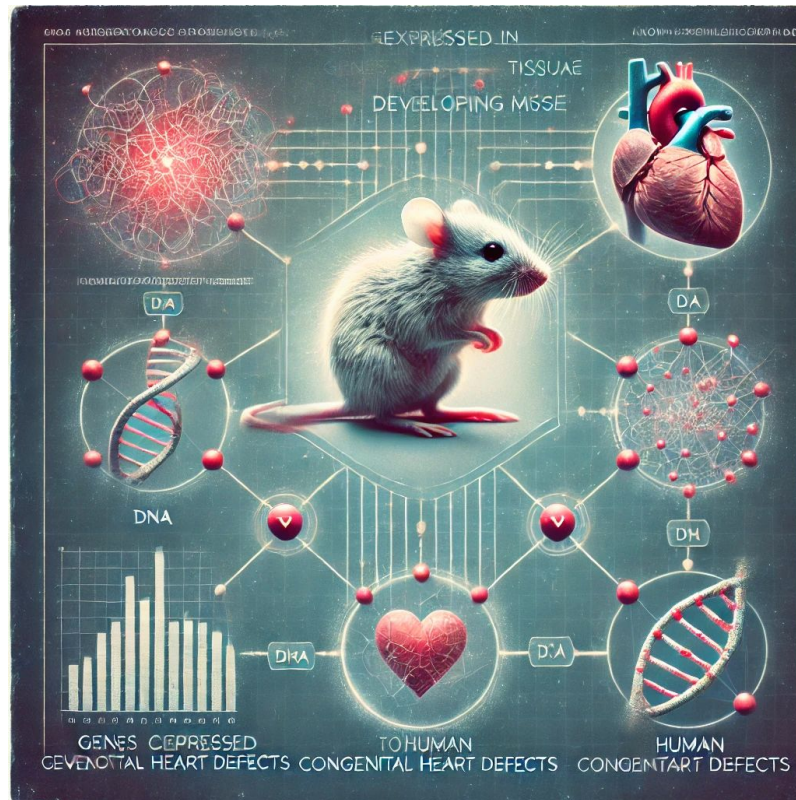


# Data Distillery Use Case: Exploring Fetal Heart Development

We can use the Data Distillery KG's corpus of integrated data to ask complex questions that would ordinarily take weeks of data integration effort.

**Example:** What genes linked to heart defects in mice are also actively expressed in developing human heart cells?

This information would help in predicting heart defect risks in affected families by pinpointing which genes to examine for genetic variation.





# Petagraph Use Case: Drug vs Tissue Effect

**Question:** How to predict what human tissues are most likely going to be affected by a drug?

**Example:** Rofecoxib (Vioxx) was recalled due to high cardiovascular risks.

Petagraph already has useful data integrated:

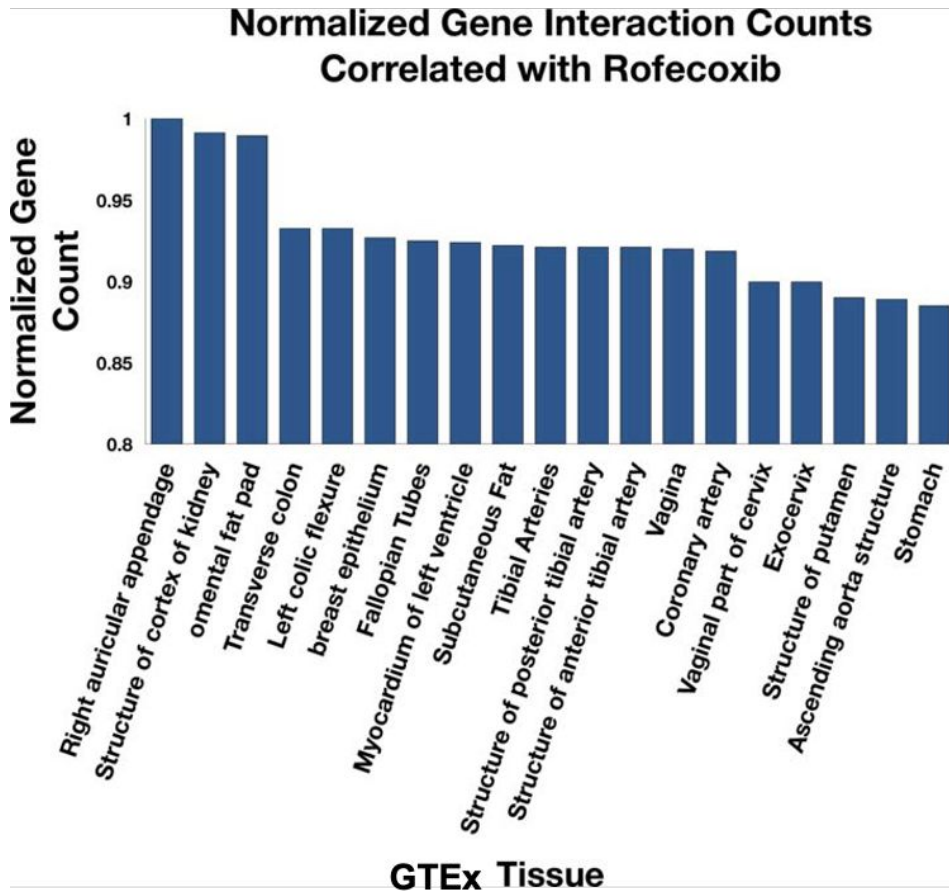
- Rofecoxib's gene expression signatures of dosing human cell cultures (LINCS/L1000 projects)
- Integrated with gene expression signatures across many human tissues (GTEx project).

**Petagraph KG Query: What human tissues are most likely to be perturbed by rofecoxib?**

**Result:** Heart and blood vessels, such as the right auricular appendage and myocardium of the left ventricle, were predicted as the most affected tissues based on the number of shared genes with rofecoxib's dosing profile.

Predictions align with the known cardiovascular risks of rofecoxib.

Query took seconds to complete.



# KGs as FAIR and useful tools

Data Distillery and Petagraph can be freely downloaded with a UMLS license (<https://www.nlm.nih.gov/databases/umls.html>)

Users with a UMLS license can follow site instructions to obtain and utilize a UMLS license key at <https://ubkgdownloads.xconsortia.org/>

Installable with a 16G memory laptop!

# Acknowledgements

**Ben Stear**

**Kids First**

Mano R. Maurya

MW, DRC

**Taha Mohseni Ahooyi**

**Kids First**

Srinivasan Ramachandran

MW, DRC

**Alan Simmons**

**HuBMAP**

Eoin Fahy

MW

**Jonathan C. Silverstein\***

**HuBMAP**

Shankar Subramaniam

MW, DRC

Christopher Nemarich

KF

Bernard de Bono

SPARC

Christophe G. Lambert

IDG

Jeffrey S. Grethe

SPARC

Vincent T. Metzger

IDG

Thomas H. Gillespie

SPARC

John Erol Evangelista

LINCS, DRC

Fahim T. Imam

SPARC

Daniel J. B. Clarke

LINCS, DRC

Natallia Kokash

SPARC

Zhuorui Xie

LINCS, DRC

Matthew, E. Roth

ERCC

Heesu Kim

LINCS, DRC

Robert Fullem

ERCC

Sherry L. Jenkins

LINCS, DRC

Dubravka Jevtic

ERCC

Avi Ma'ayan

LINCS, DRC

Aleks Milosavljevic

ERCC

George Papanicolaou, Christy Kano NIH

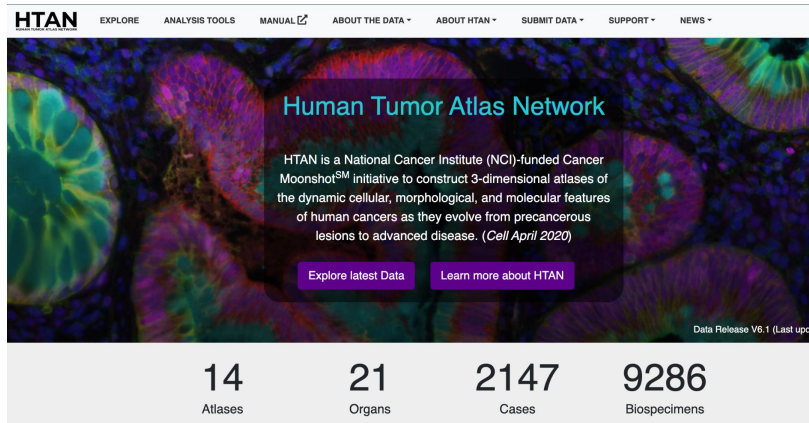
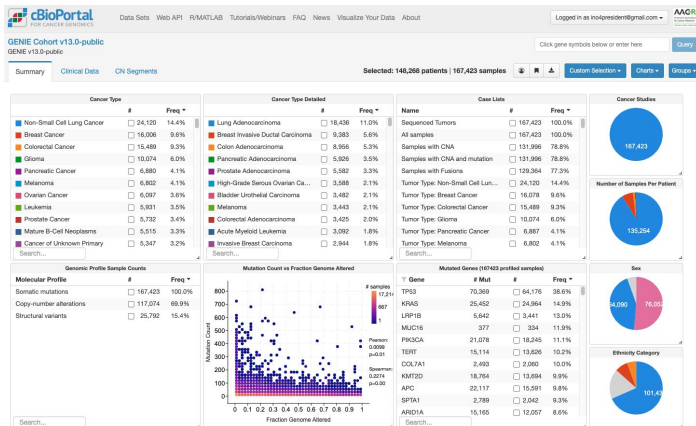


**Ino de Bruijn, *Memorial Sloan  
Kettering Cancer Center***

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<https://cbiportal.org> & <https://humantumoratlas.org>

# FAIR Data Portals for multimodal cancer data



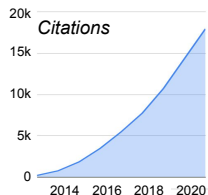
**Bulk Genomics & Clinical Data Focus**

**Wide variety of assays incl single cell, multiplex imaging**

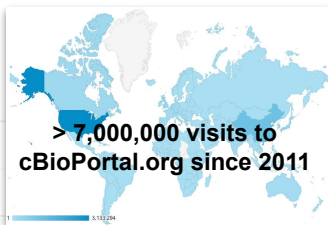


# cBioPortal over time

<https://cbioportal.org>



- Curated Studies >350
- Monthly Users ~34k
- Total Citations ~18k



Weekly user visits



Start of cBioPortal Query/Results view

Patient View Study View

Open source transition (MSK, DFCI & Hyve)

PMCC & CHOP joined

Architecture upgraded Comparison View

Start of TCGA

First SU2C cBioPortal

CANCER DISCOVERY Cerami et al. Cancer Discov.

Gap et al. Sci Signal. cBioPortal @MSK

cBioPortal @PMCC cBioPortal @DFCI Project GENE

cBioPortal @CHOP

NCI ITCR funding

HTAN Start of HTAN Count Me In

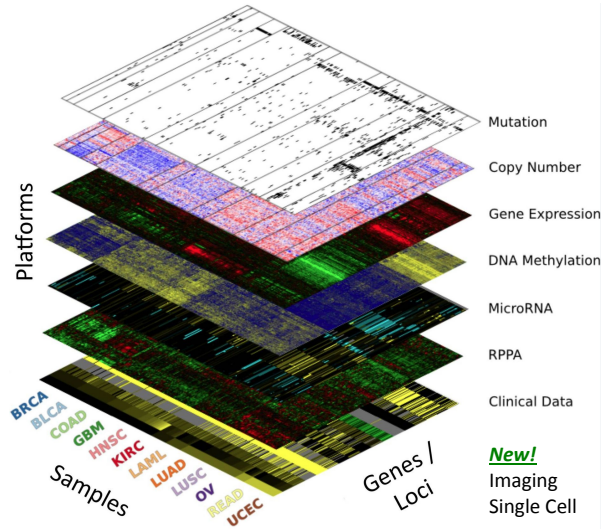
AAGR GENIE BPC

Webinars

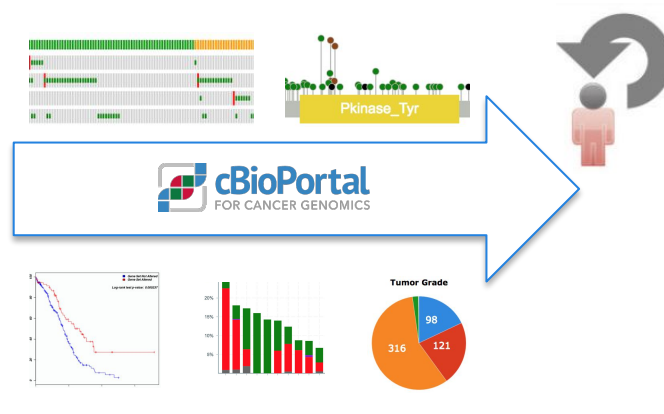


# Goal of cBioPortal

Make complex cancer genomic data accessible and interpretable for cancer biologists and clinicians



Adapted from TCGA Nat Genet 2014



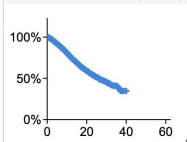
Biological  
discovery

Clinical  
interpretation

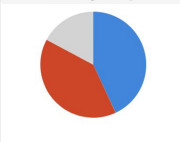
# Cohort exploration

Cancer Type	#	Freq
Non-Small Cell Lung Cancer	1,668	15.2%
Breast Cancer	1,324	12.1%
Colorectal Cancer	1,007	9.2%
Prostate Cancer	717	6.6%
Glioma	553	5.1%
Pancreatic Cancer	502	4.6%
Soft Tissue Sarcoma	443	4.0%
Bladder Cancer	423	3.9%
Melanoma	365	3.3%
Renal Cell Carcinoma	361	3.3%
Hepatobiliary Cancer	355	3.2%

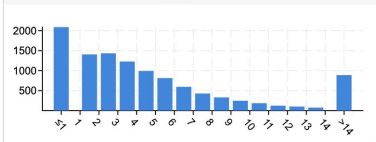
KM Plot: Overall Survival (months)



Smoking History

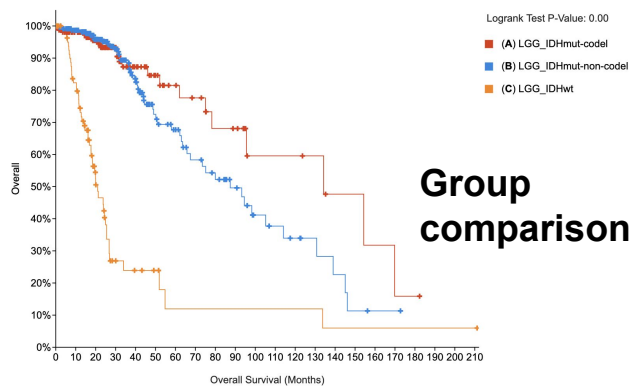


Mutation Count

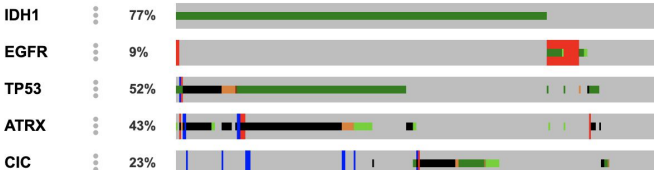


Mutated Genes (10945 profiled samples)

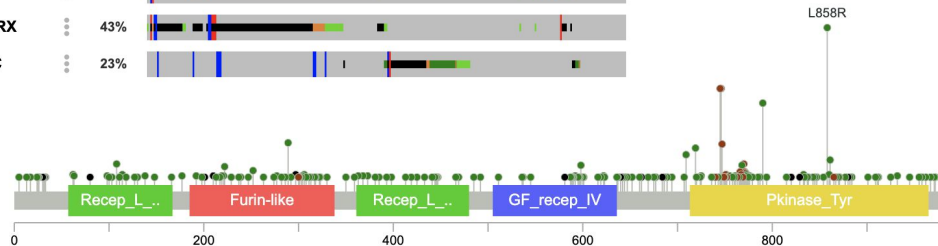
Gene	# Mut	#	Freq
TP53	4,954	4,538	41.5%
KRAS	1,670	1,643	15.0%
TERT	1,549	1,460	13.3%
PIK3CA	1,517	1,355	12.4%
APC	1,674	1,121	10.2%
ARID1A	1,072	875	8.0%
KMT2D	1,210	851	7.8%
PTEN	764	665	6.1%
KMT2C	818	642	5.9%
EGFR	799	636	5.8%
FAT1	773	595	5.4%



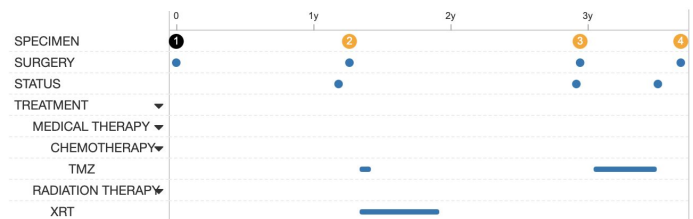
Group comparison



## Gene-centric queries



## Patient visualization



Samples	Gene	Protein Change	Annotation
1 2 3 4	IDH1	R132C	🔍 📄 📉 🔥
1 2 3 4	TP53	R248Q	🔍 📄 📉 🔥
3	ERC1	L283Fs*20	🔍
3	HSP90AB1	K72E	🔍
3	SPRNT	F404Lfs*3	🔍
1	OR10V1	R273W	🔍
1 2 3 4	TEAD3	P51L	🔍

# The HTAN Portal <https://humantumoratlas.org/>

**HTAN**  
HUMAN TUMOR ATLAS NETWORK

EXPLORE ANALYSIS TOOLS MANUAL ABOUT THE DATA ABOUT HTAN SUBMIT DATA SUPPORT NEWS

## Human Tumor Atlas Network

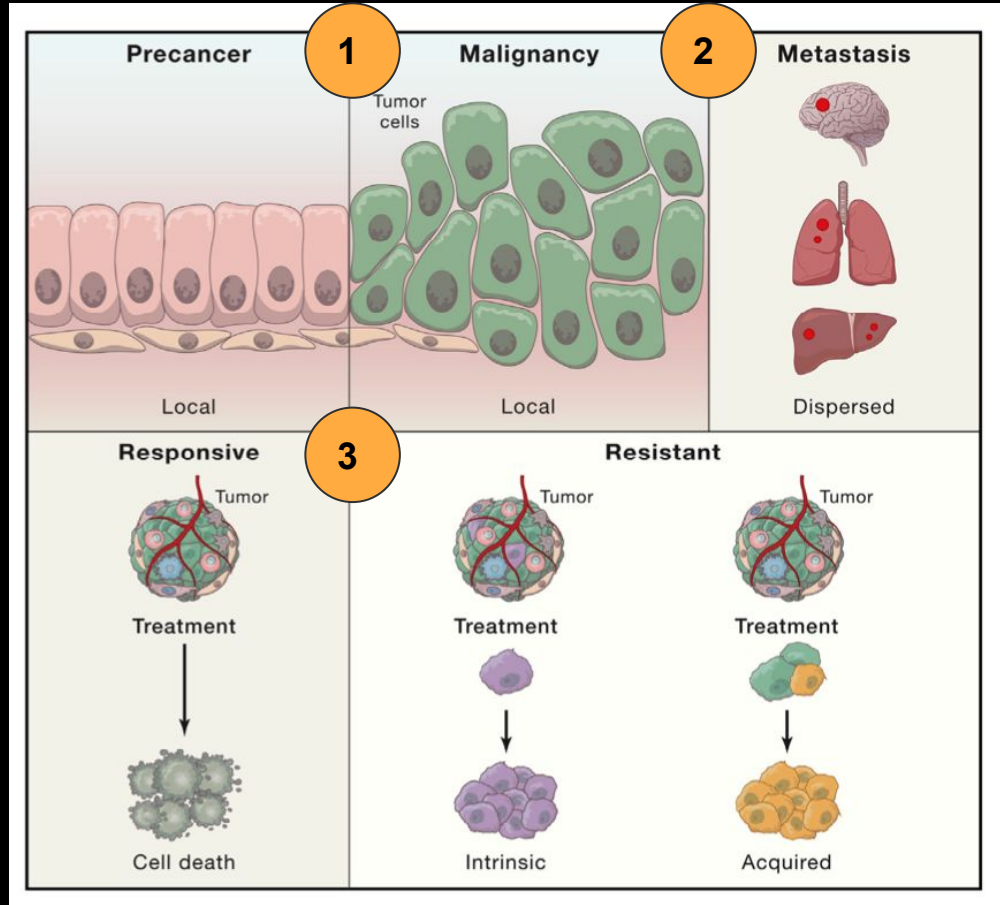
HTAN is a National Cancer Institute (NCI)-funded Cancer Moonshot<sup>SM</sup> initiative to construct 3-dimensional atlases of the dynamic cellular, morphological, and molecular features of human cancers as they evolve from precancerous lesions to advanced disease. (*Cell April 2020*)

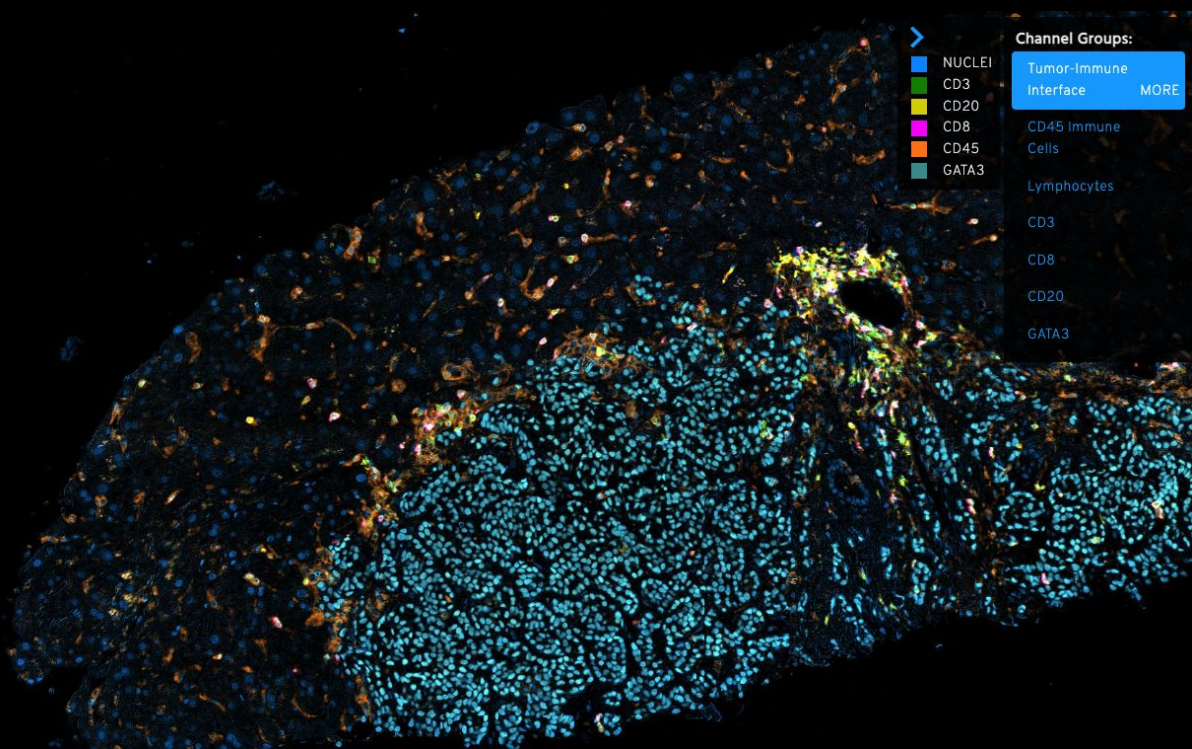
Explore latest Data Learn more about HTAN

Data Release V6.1 (Last updated 2024-11-05)

# Sharing data from the HTAN Consortium

HTAN is focused on **transitions** in cancer

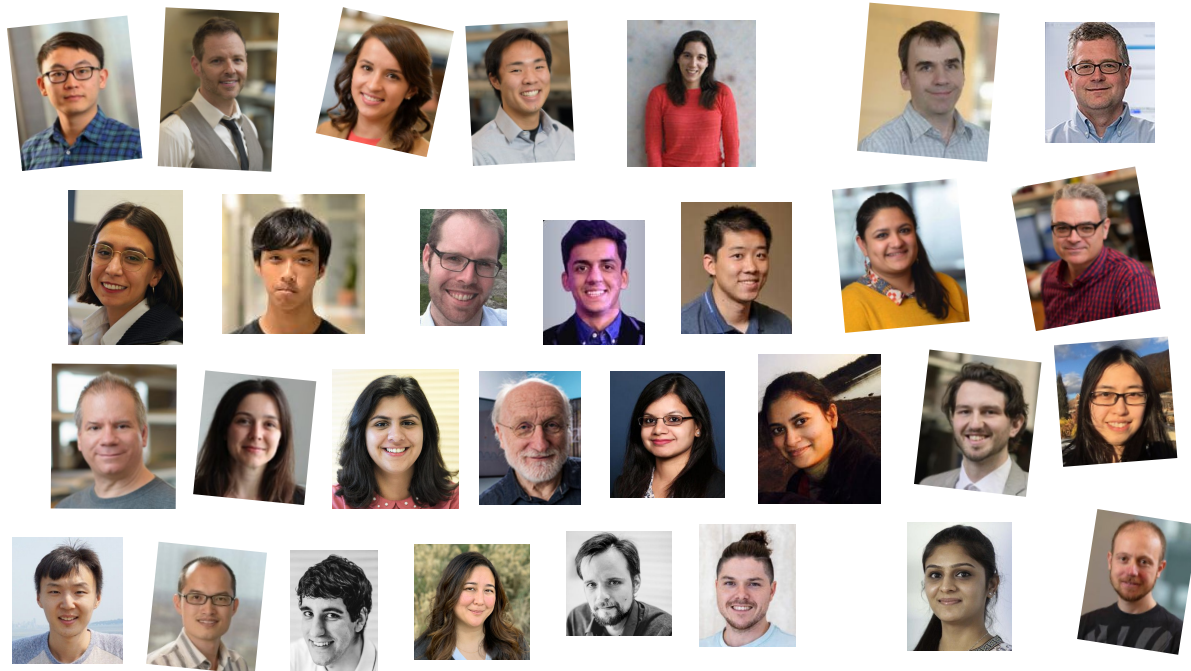





Lymphocytes in breast cancer liver metastasis biopsy (OHSU HTAN Atlas).

Most HTAN groups have a focus on **single cell** and **multi-plex imaging** modalities

Many thanks to these amazing teams!





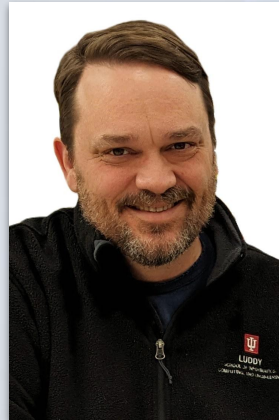
**Bruce W. Herr II, *Indiana University***  
***(HuBMAP)***

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# Human Reference Atlas 5-star Linked Open Data: Construction, Publication, and Usage

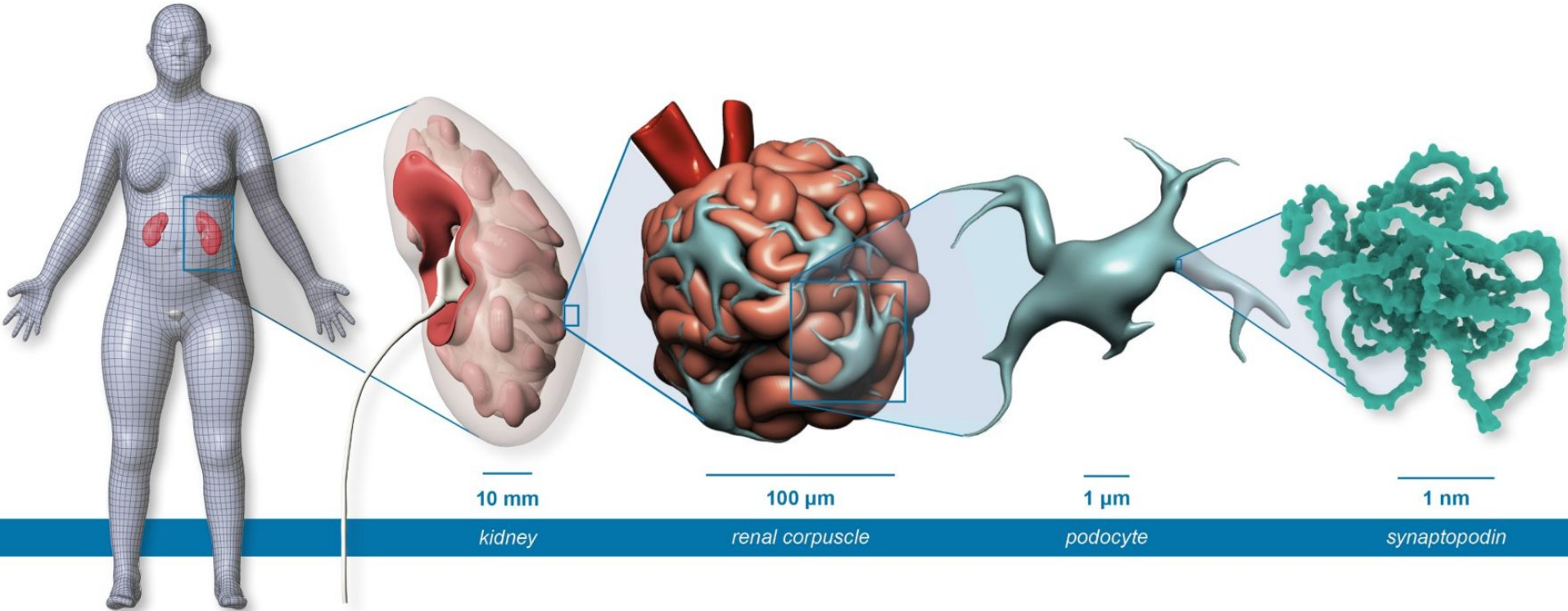


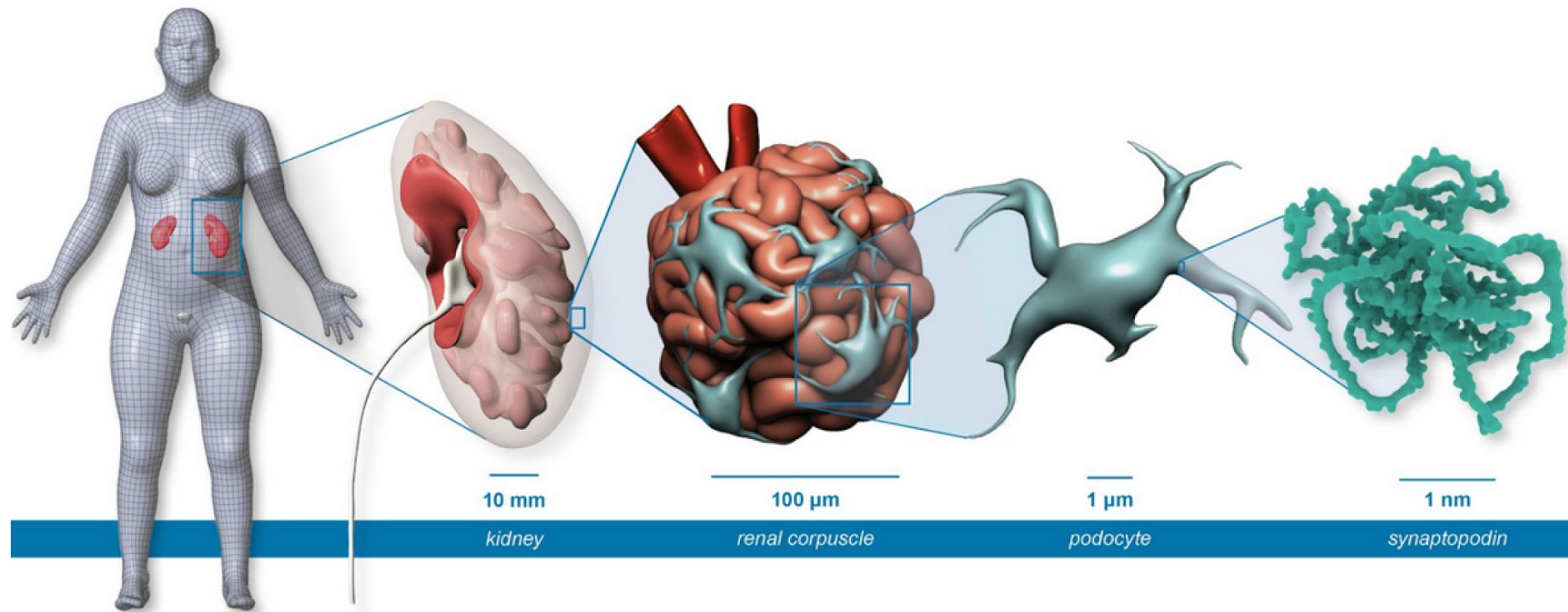
Bruce W. Herr II  
Technical Director

*Cyberinfrastructure for Network Science Center  
Department of Intelligent Systems Engineering  
Luddy School of Informatics, Computing, and Engineering  
Indiana University, Bloomington, IN, USA*

# Human Reference Atlas (HRA)

A comprehensive, ontologically aligned, high-resolution, three-dimensional, multiscale atlas of anatomical structures and cells in the healthy human body



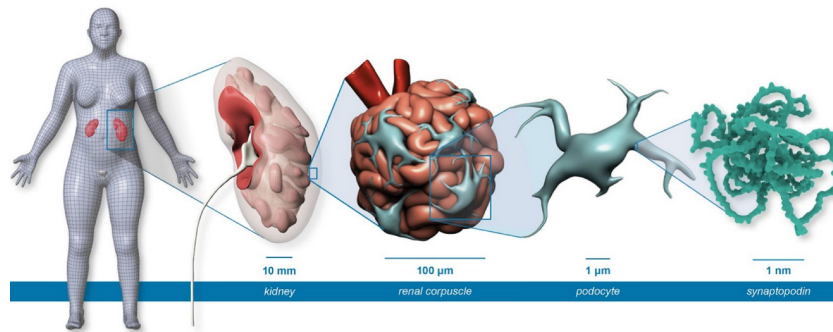


Anatomical Structures

Functional  
Tissue Units

Cell Types

Biomarkers  
Genes, Proteins, ..



Anatomical Structures

Functional  
Tissue Units

Cell Types

Biomarkers  
Genes, Proteins, ..

Biology

Anatomical Structures, Cell Types, and Biomarkers Tables

Atlas

3D Reference Organs

2D FTU  
Illustrations

Organ Mapping Antibody Panels

Vascular Geometry

Cell Type  
Annotations



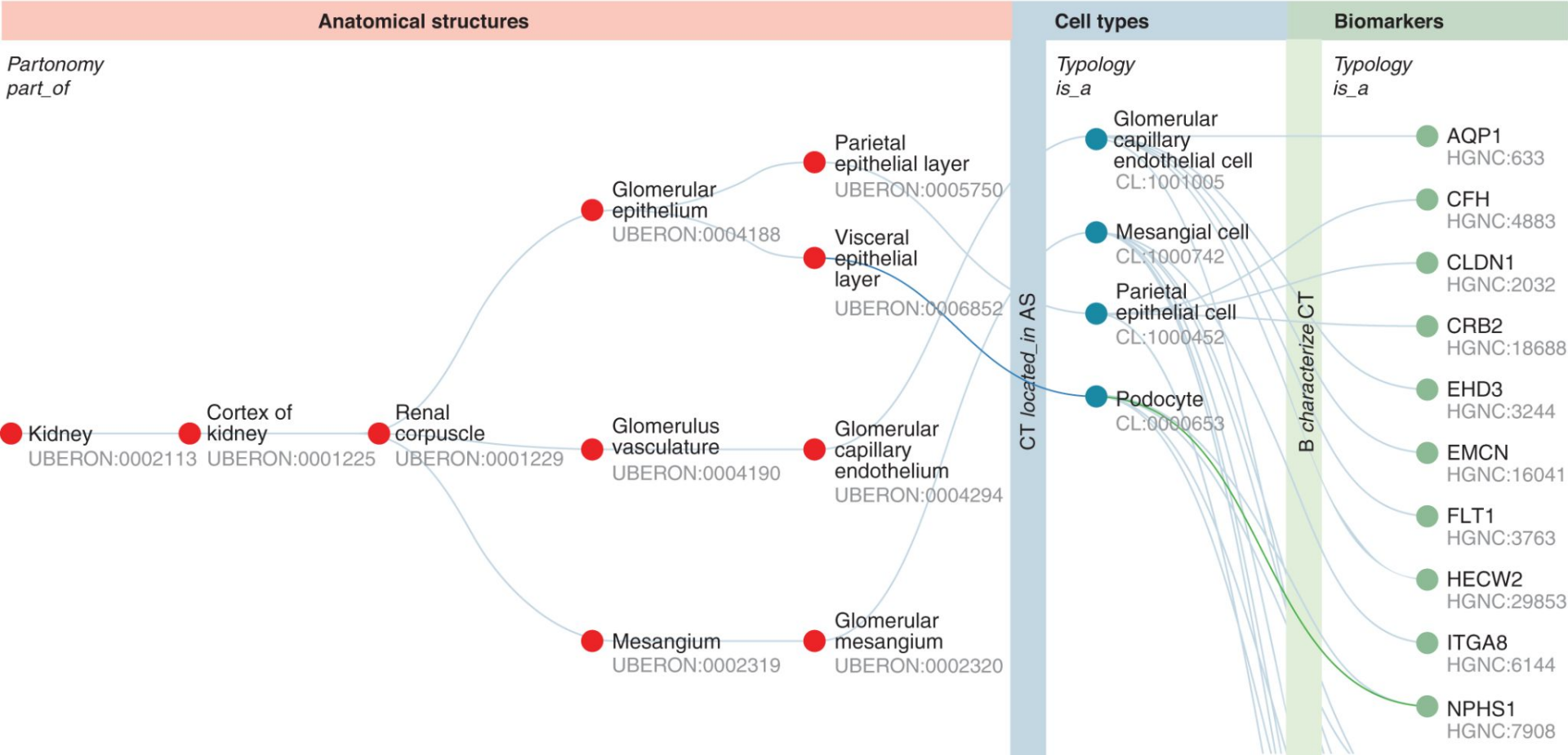
Experimental Data (Donors, Tissues,  
Extraction Sites, Datasets)

Atlas++

HRApop (Experimental Data + Cell Summaries)

HRAlit (HRA-relevant Literature)

# HRA ASCT+B Tables



# HRA Functional Tissue Unit (FTU) Illustrations

Ascending thin limb of loop of Henle (20  $\mu$ m)

Descending thin limb of loop of Henle (20  $\mu$ m)

Thick ascending limb of loop of Henle (26  $\mu$ m)

Collecting duct (outer medulla) (45  $\mu$ m)

Cortical collecting duct (45  $\mu$ m)

Collecting duct (inner medulla) (50  $\mu$ m)

Dermal papilla (50  $\mu$ m width)

Epidermal ridge (150  $\mu$ m depth)

Intercalated duct of pancreas (5  $\mu$ m)

Acinus of pancreas (30  $\mu$ m)

0.1 mm

Islets of Langerhans (0.1 mm)

Pulmonary alveolar parenchyma (0.2 mm)

Renal corpuscle (0.2 mm)

Bronchial submucosal gland (0.3 mm)

Villus of small intestine (1-1.5 mm)

Crypt of Lieberkuhn (0.5-0.7 mm)

Prostatic glandular acinus (0.2 mm)

0.2 mm

Liver lobule (1-2.5 mm)

Thymus lobule (0.5-2 mm)

Red pulp of spleen (variable)

White pulp of spleen (0.5-1.0 mm)

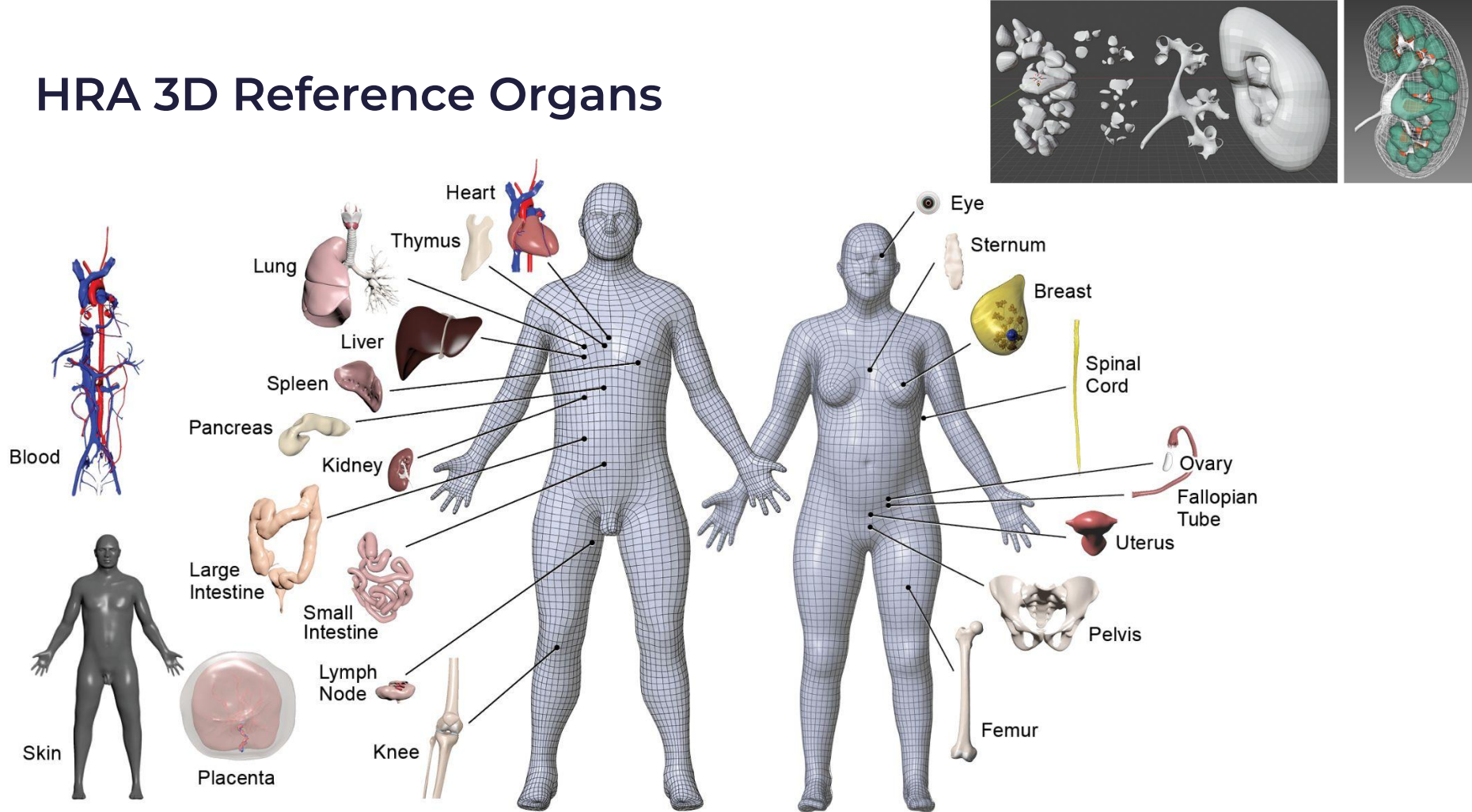
0.5 mm

Nephron tubule (3-5 mm)

1 mm

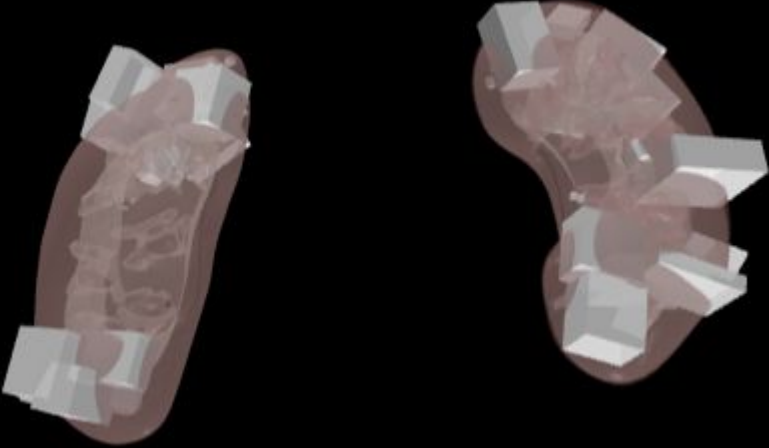


# HRA 3D Reference Organs

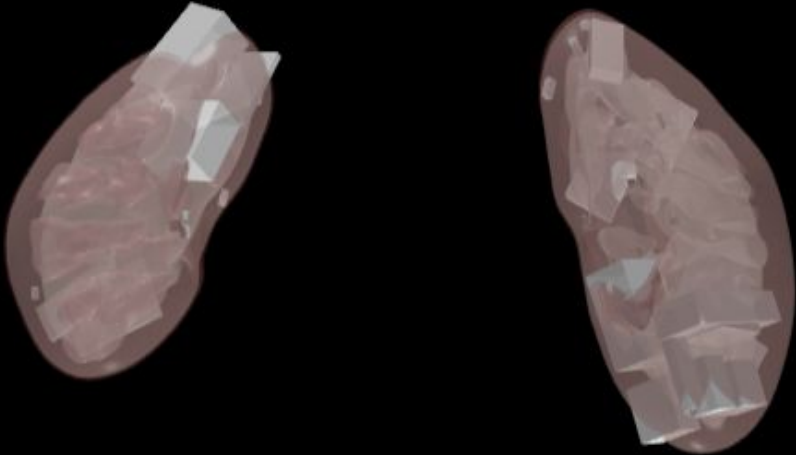


# HRA 3D Reference Organs: kidneys

Male



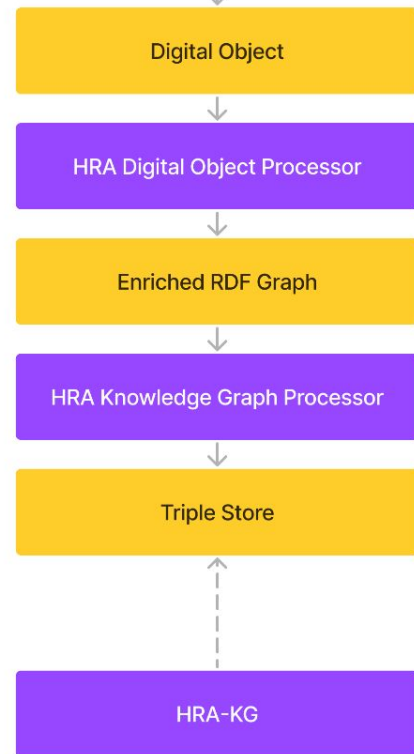
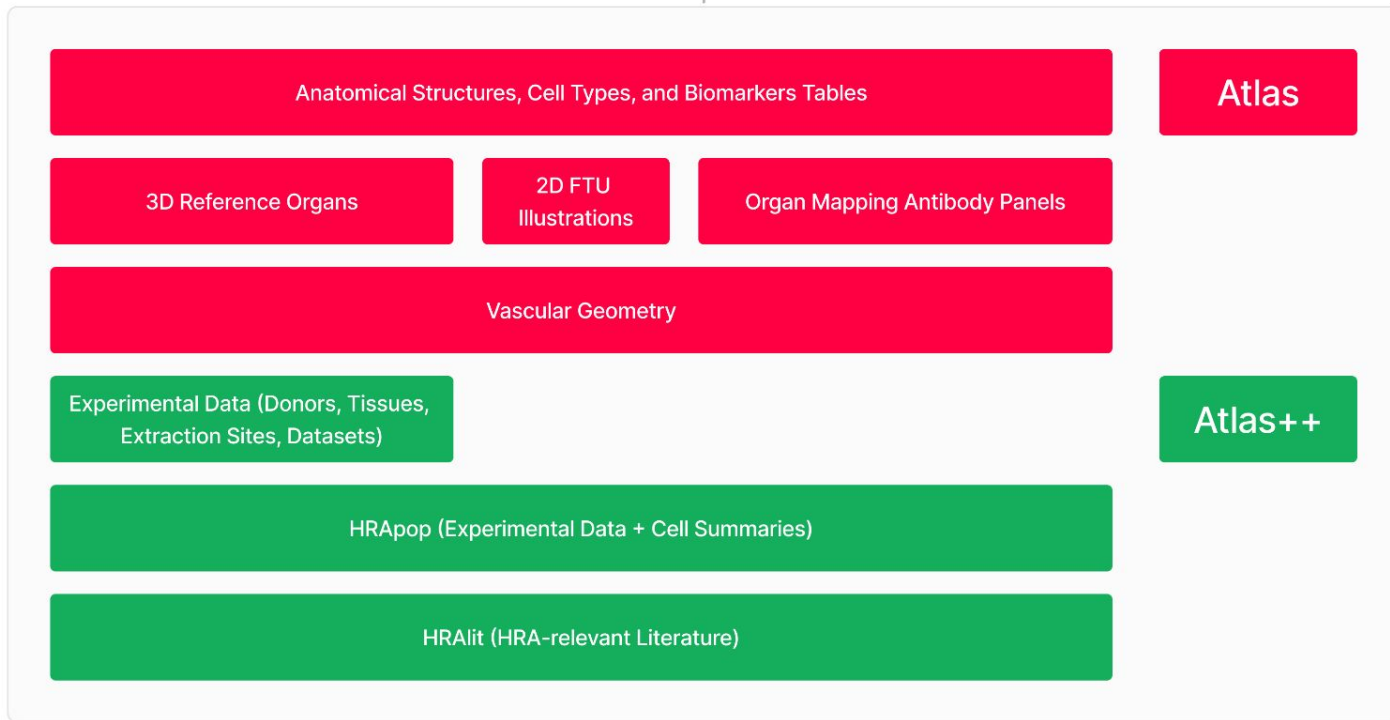
Female



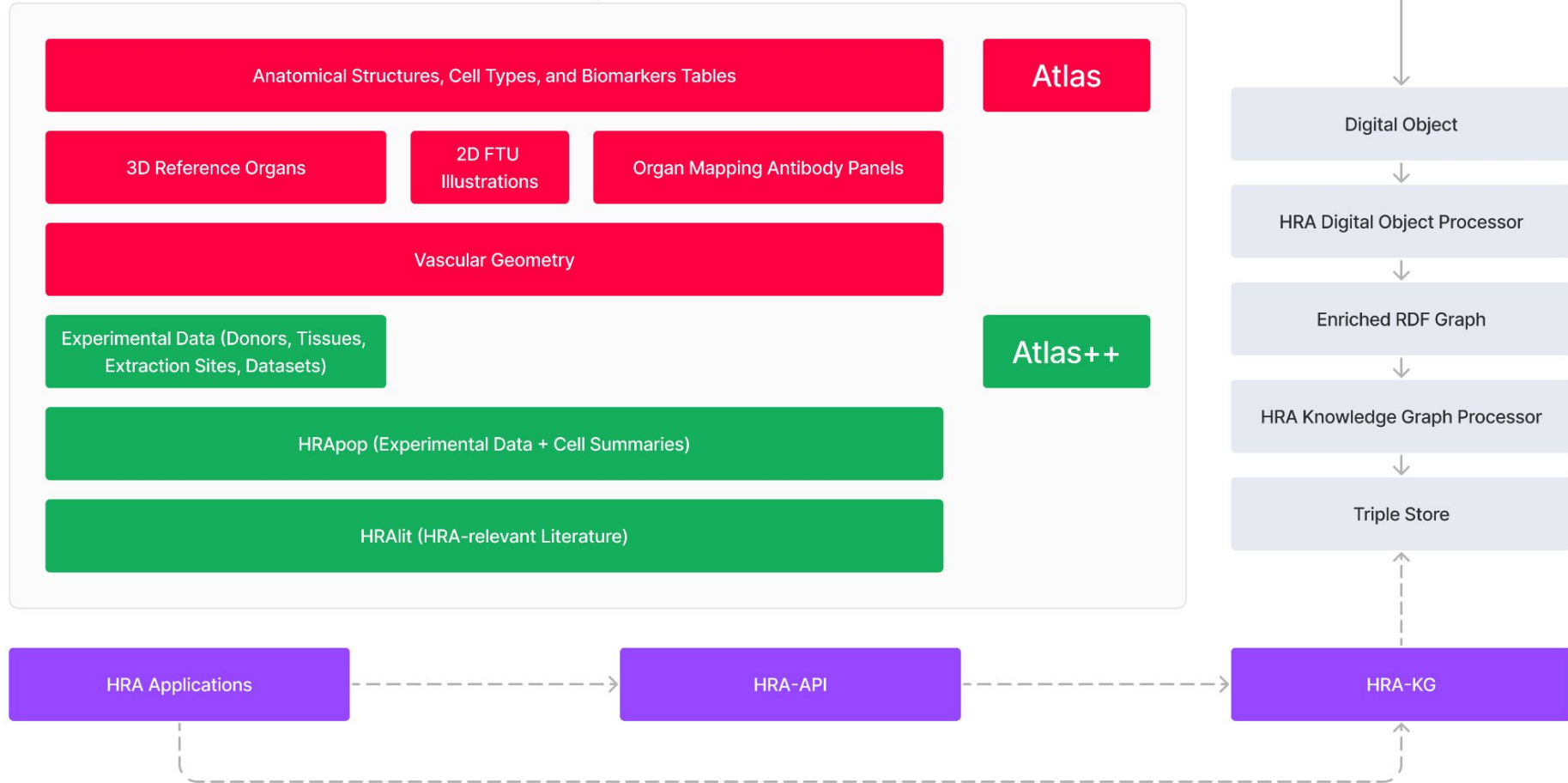
126 tissue blocks with 1,198 datasets from 4 consortia



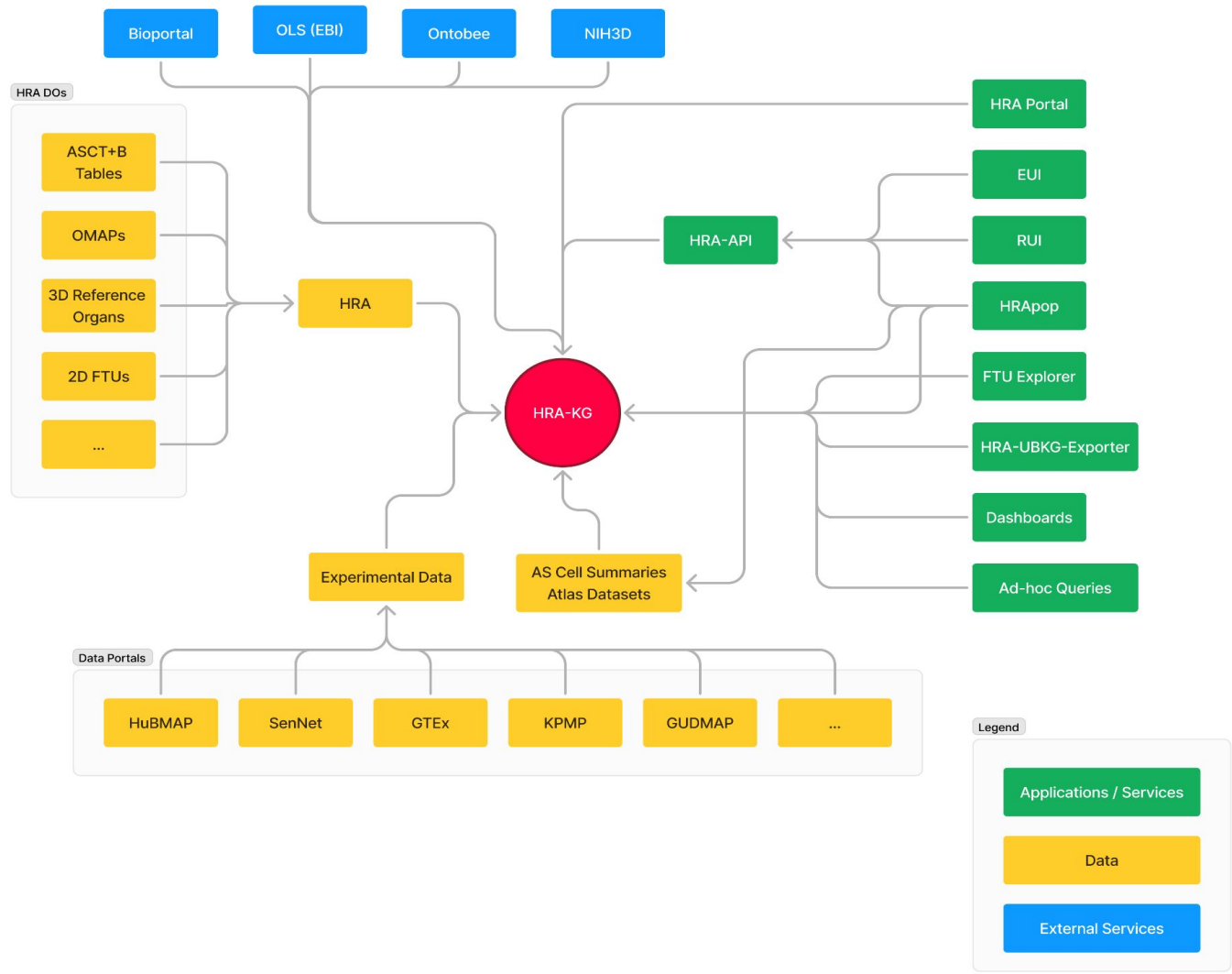
# HRA Knowledge Graph Framework



# HRA API and Applications



# HRA Ecosystem



# HRA KG Data

## Digital Objects ⓘ

<b>36</b> ASCT+B Tables	<b>23</b> OMAP Tables	<b>22</b> FTU Illustrations	<b>71</b> 3D Organ Models	<b>1</b> Blood Vessel Segments	<b>28</b> SOPs
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## Size of the HRA Knowledge Graph ⓘ

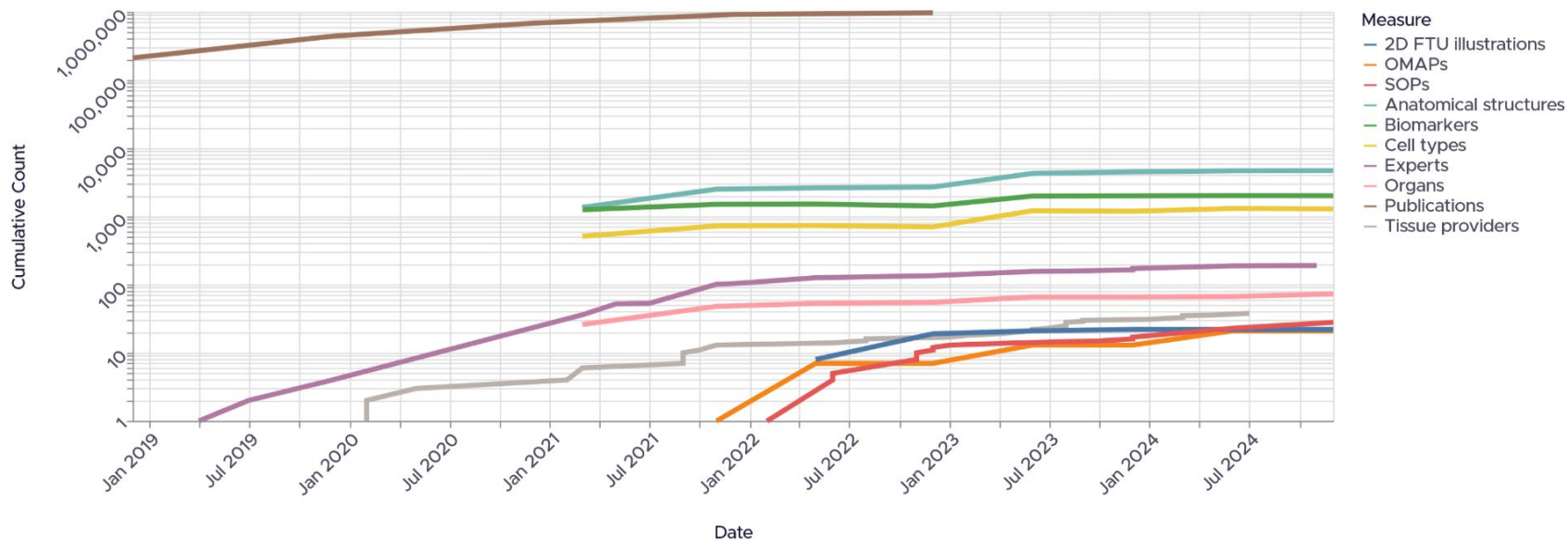
<b>10,064,033</b> Nodes	<b>171,250,177</b> Edges	<b>125,838 MB</b> Size
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## Ontologies Extended ⓘ

<b>126</b> Terms added to Uberon	<b>141</b> Terms added to CL	<b>461</b> Terms added to PCL
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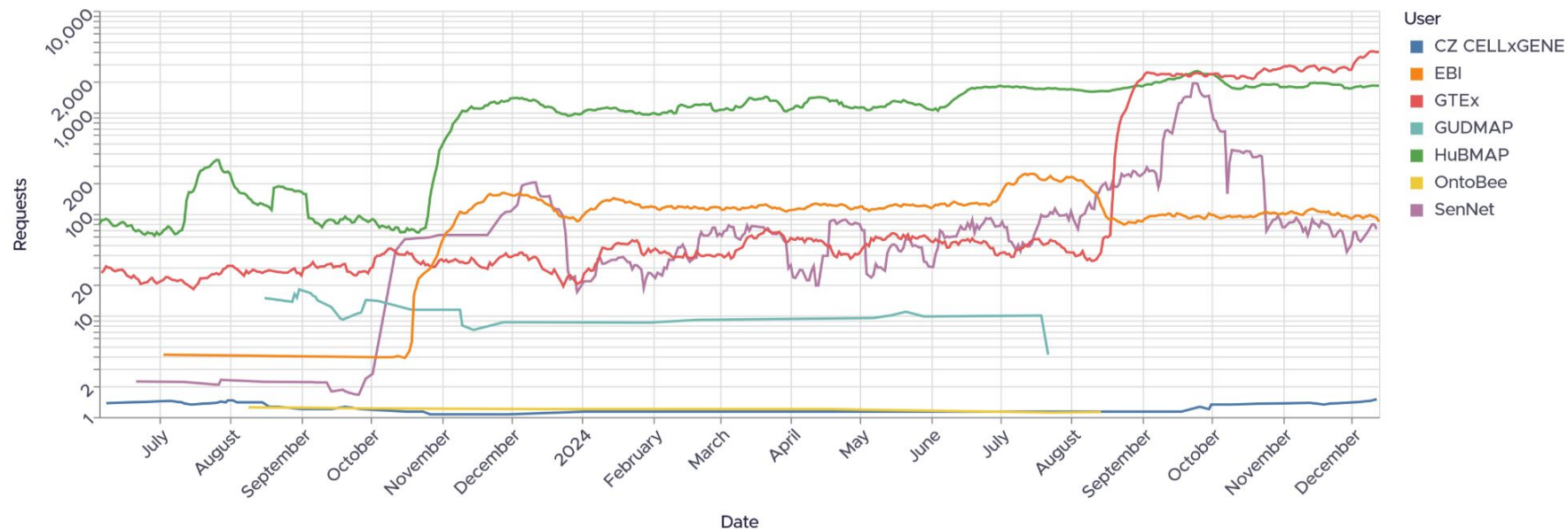
# HRA KG Data

HRA Growth Per Release ⓘ



# HRA KG (and other HRA products) usage

Digital Object Usage ⓘ

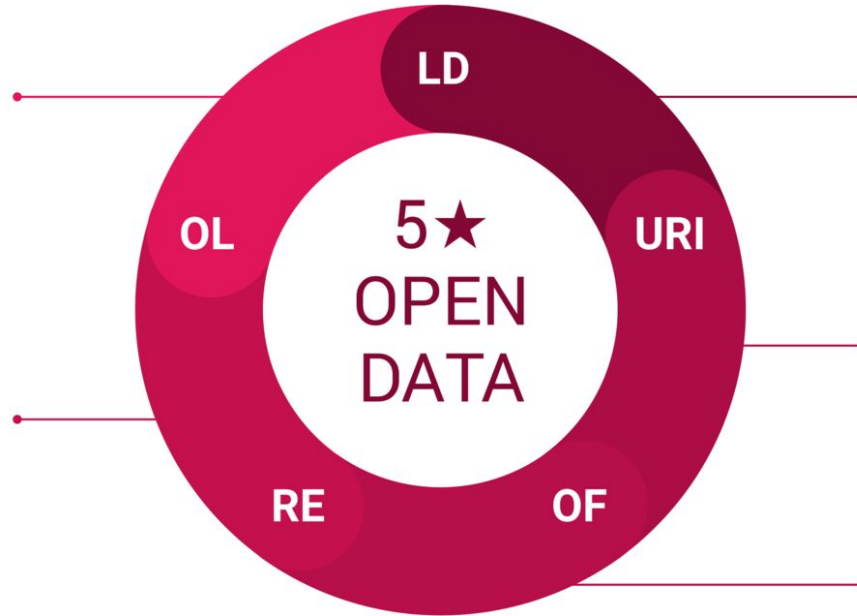


★ **open licence** [PDF]

available on the web (whatever format) but with an open licence, to be Open Data.

★★ **machine readable** [XLS]

available as machine-readable structured data.



★★★★★ **Linked Open Data**

★★★★ plus: link your data to other people's data to provide context.

★★★★★ **use URIs** [RDF]

★★★ plus: use open standards from W3C (RDF and SPARQL) to identify things, so that people can point at your stuff.

★★★ **open format** [CSV]

★★ plus non-proprietary format.

# 5 Star Linked Open Data

URLs:

- **purl**.humanatlas.io (permanent identifiers)
- **lod**.humanatlas.io (hosts digital object metadata as DCAT Datasets)
- **cdn**.humanatlas.io (hosts raw data files)

Content Negotiation:

- **purl** returns the digital object data based on accept header
  - Redirect to **lod** for humans, JSON or RDF Formats for machines
- **lod** returns the digital object metadata based on accept header
  - Return HTML for humans, JSON or RDF Formats for machines



# Infrastructure

- **HRA KG** data is pre-computed and uploaded to *Amazon S3*.
- **purl/lod/cdn.humanatlas.io** is hosted using *Amazon CloudFront* which returns data hosted on *Amazon S3* and implements content negotiation via *Amazon CloudFront Functions*.
- The latest version of each digital object in RDF format is loaded into a *Blazegraph* SPARQL server hosted by *Amazon ECS* and available via *Amazon CloudFront* at <https://lod.humanatlas.io/sparql>. This allows ad-hoc queries of HRA KG data using SPARQL.

# Scalability

- With the use of pre-computed data and hosting them as static files means the solution scales massively with low technical complexity.
- Caching from *Amazon CloudFront* and static files makes it extremely fast.
- *Blazegraph* can scale to billions of edges allowing fast ad-hoc queries.

# FAIRness

- All data is represented in standard RDF format using standard ontologies and terminology where possible.
- Uses a well known Linked Open Data pattern.
- All data is CC BY 4 and code is MIT licensed.



**Jonathan Silverstein, *University of Pittsburgh, HuBMAP and SenNet***

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# 24hr - HuBMAP and SenNet FAIR and Scalable Infrastructures

Presented by

Jonathan Silverstein, Phil Blood, Nils Gehlenborg, Kay Metis

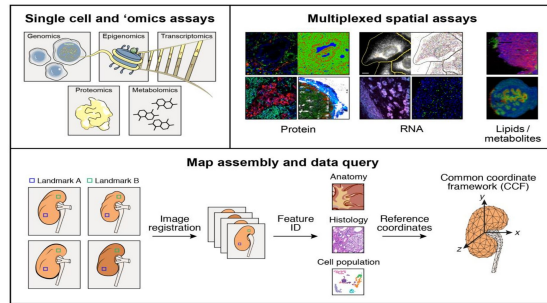
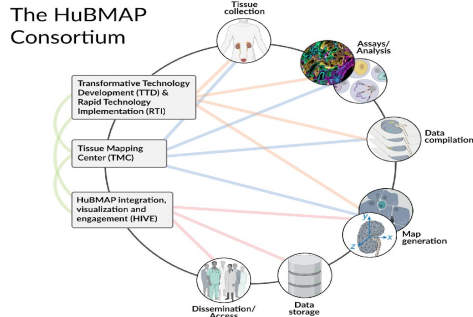
# SenNet (and HuBMAP) DCC Team

Adams, Andrew	Information Security Officer	Maier, Elizabeth	UX Designer
Adams, Etowah	Software Developer	Metis, Kay	Executive Director
Alkathan, Mariam	Project Manager	Misra, Gaurab	Project Manager
<b>Bar-Joseph, Ziv</b>	<b>PD/PI</b>	Moerth, Eric	Software Developer
Betancur, David	Senior Software Engineer	Morgan, Richard	Project Manager
Bidanta, Supriya	Graduate Student (research assistant)	Nguyen, Nam	Postdoctoral Scholar
Bisciotti, Joe	Project Manager	Niculcea, Emily	Staff
<b>Blood, Philip</b>	<b>PD/PI</b>	Puerto, Juan	Cloud Developer
Bolin, Axel	Programmer	Qaurooni, Danial	Postdoctoral Scholar, Fellow,
<b>Borner, Katy</b>	<b>Co-Investigator</b>	Ropelewski, Alexandar J	Senior Scientist
Bueckle, Andreas	Scientist	<b>Ruffalo, Matthew</b>	<b>Co-Investigator</b>
Burke, Christopher	Systems Engineer	Ruschman, Nancy	Project Manager
Burke, Karl	Senior Software Developer	<b>Satija, Rahul</b>	<b>Co-Investigator</b>
Cao-Berg, Ivan	Data Wrangler	Scibek, Robin	Program Management
Chiacchia, Ken	Science Writer	Shirey, Bill	Senior Software Developer
Choudhary, Saket	Postdoctoral Scholar	Sibilla, Max	Software Developer
<b>Gehlengorg, Nils</b>	<b>Co-Investigator</b>	<b>Silverstein, Jonathan C</b>	<b>PD/PI</b>
Ginda, Michael	Research Analyst	Simmons, Joseph Alan	Solutions Architect
Hasanaj, Euxhen	Postdoctoral Scholar	Singh, Raman Kumar	Postdoctoral Scholar
Helfer, Jesse	Project Manager	Smith, Tracey	Project Coordinator
Herr, Bruce William	Systems Architect	Soleiman, Natasha	Project Manager
Honick, Brendan	Data Curator and Metadata Librarian	Song, Qi	Graduate Student (research assistant)
Jain, Yashvardhan	Research Software Developer	Turner, Morgan	R&D Manager
Kenney, Mariah	Data Curator and Metadata Librarian	Uranic, Jackie	Data Curator and Metadata Librarian
Klinesmith, Holly	Communications Coordinator	Varney, Lisa Ann	Software Developer
Lee, Young Je	Postdoctoral Scholar	Yuan, Zhou	Senior Software Developer
Madonna, Tyler	Programmer		

# HuBMAP

Develop an open and global platform to map healthy cells in the human body and determine how relationships among cells affect human health

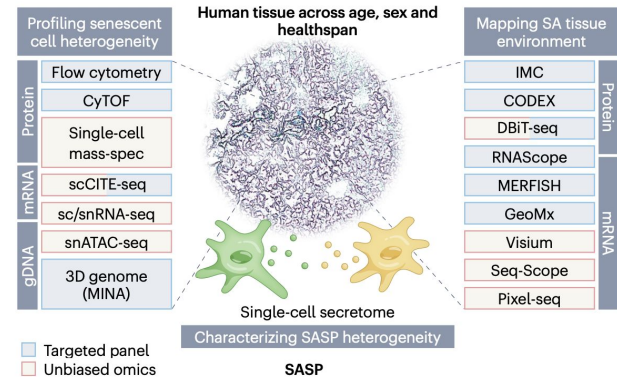
**The Human Body at Cellular Resolution: The NIH Human Biomolecular Atlas Program.**  
Snyder et al. *Nature*. 574, p. 187-192 (2019)



# SenNet

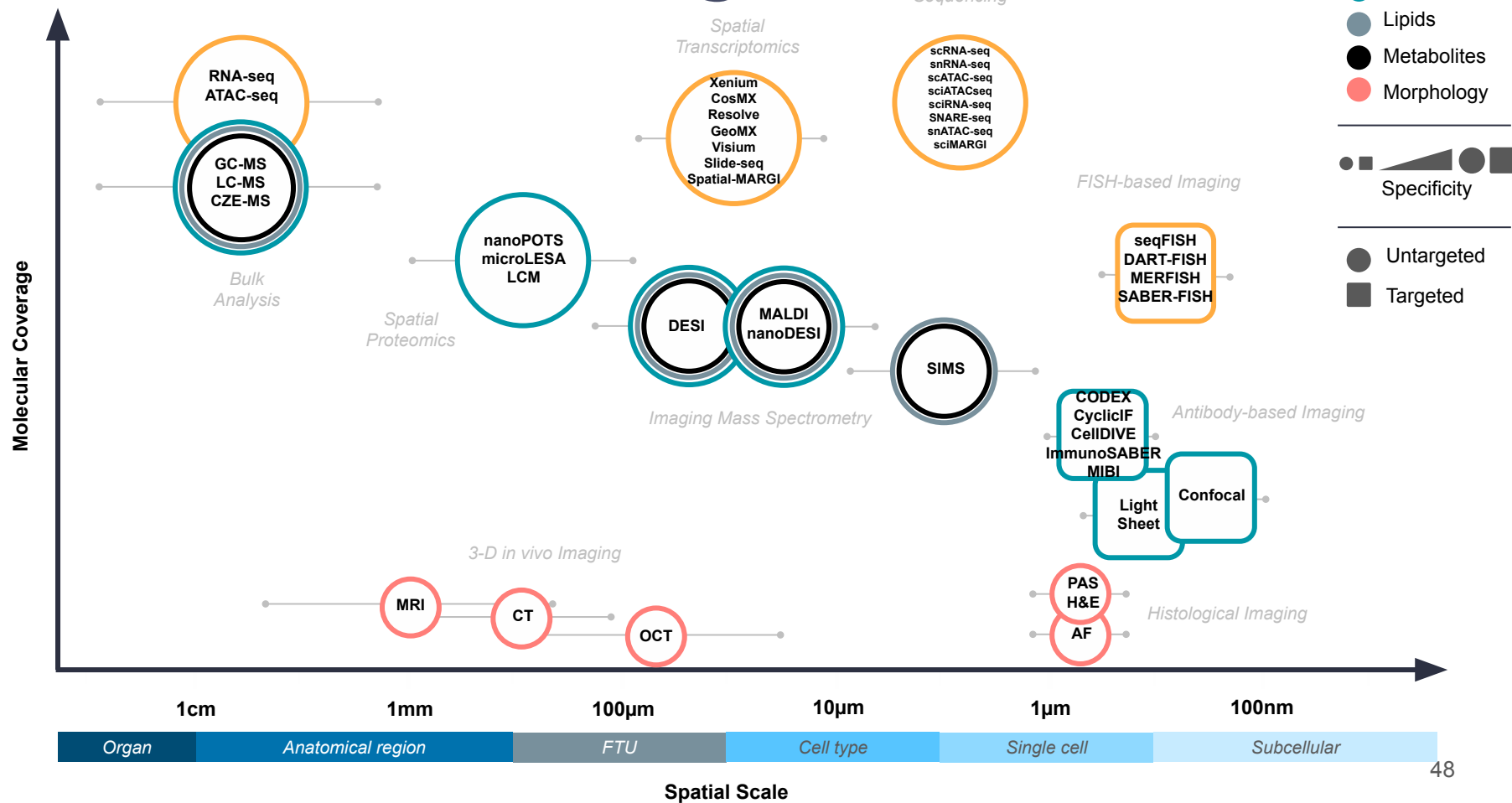
Identify and functionally characterize the heterogeneity of senescent cells across multiple tissues in human health and lifespan at single cell resolution

**NIH SenNet Consortium to map senescent cells throughout the human lifespan to understand physiological health.** SenNet Consortium.  
*Nature Aging* 2, 1090–1100 (2022)



**NIH Common Fund Programs:** The NIH Common Fund supports bold scientific programs that catalyze discovery across all biomedical and behavioral research. These programs enable NIH Institutes and Centers to collaborate on innovative research expected to address high priority challenges for the NIH as a whole. Each Common Fund Program typically supports dozens of centers with hundreds of millions of dollars in funding and are up to ten years in length. We lead the Infrastructure, Engagement, and Data Processing/Coordination for the two Common Fund programs HuBMAP and SenNet.

# HuBMAP Technologies





# PSC: A place for getting research done...

## National service provider



## Collaborative Research



## Education and training



PSC IS A JOINT  
COMPUTATIONAL RESEARCH  
CENTER BETWEEN **CARNEGIE  
MELLON UNIVERSITY** AND  
THE **UNIVERSITY OF  
PITTSBURGH**.

*39 years of leadership*

*23 HPC systems*

(11 of which were the first or  
unique)

*Pioneering Artificial Intelligence  
and Research Computing.*

## CMU/Pitt Community



## Networking and security



## Industry Partners



## Pitt's high-performance computing upgrade signals accelerated translational research

By **Brian Buntz** | March 28, 2024



The University of Pittsburgh's recent, significant expansion of its high-performance computing capabilities, courtesy of a gift from Dell Technologies, indicates a strategic commitment to using large-scale data analysis for faster translational research. The additional 9.672 gigaFLOPS of computational power – translating to nearly 9.7 trillion additional computations per second – could help Pitt's Innovation Hub for Health Science Medical Research attract research funding in areas such as precision medicine, drug discovery, and disease modeling. Interest in **high-performance computing in drug discovery** has ramped up in recent years.



*Jonathan Silverstein, professor and chief research informatics officer, and Kay Métis, Executive Director Programs and Strategic Initiatives, in the Network Operations Center (NOC) with the systems that Dell awarded to the University.  
(Tom Altany/University of Pittsburgh)*

"These large collaborative projects, which involve hundreds of investigators, utilize a unique computational architecture we've installed on the Dell system," said Jonathan C. Silverstein, MD, professor and chief research informatics officer at the University of Pittsburgh. The new system will enable secure, large-scale processing of image, genomic, and proteomic data to generate cellular and molecular maps for research.



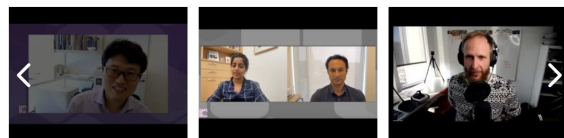
## Consortium Undergraduate Student Program

The [Consortium Undergraduate Student Program](#) is a summer research internship program for undergraduate students hosted by laboratories participating in the NIH Common Fund's Cellular Senescence (SenNet) Program.

Earlier this summer, SenNet welcomed the 2024 CUSP student cohort, all of whom interned at SenNet research sites. Get to know our 2024 CUSP students [here](#).

If you're interested in applying to a future CUSP cohort, please view our [CUSP overview video](#), which features interviews with our very first CUSP cohort as they detail their experiences in the program.

For more information on CUSP, either visit the [CUSP homepage](#) or contact [culp@sennetconsortium.org](mailto:culp@sennetconsortium.org).



## The SenNet Interview Series

The SenNet Interview Series is an anthology of video interviews profiling the researchers contributing to the SenNet Consortium. In these interviews, scientists offer candid insights into their ongoing senescence research, initiatives within the Consortium, and the ways in which SenNet functions as both a community and a resource for senescence researchers across the U.S.

The scientists interviewed discuss their backgrounds and what initially drew them to the field of senescence research. They also share their learned experience for junior investigators and students interested in this field.

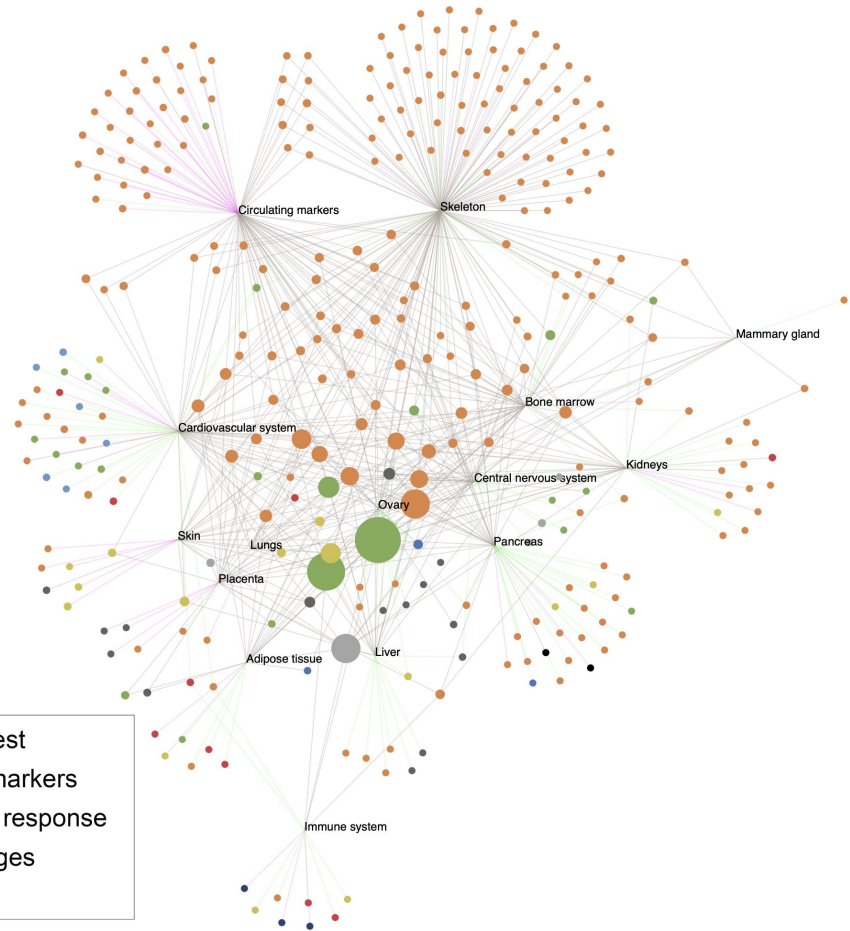
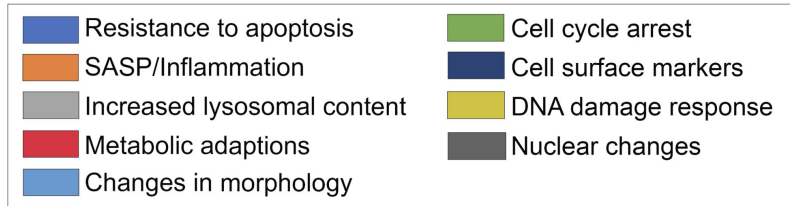
[All published interviews can be watched here.](#)

[Consider subscribing to our YouTube page](#) to be notified as soon as new interviews go live.

# SenNet Biomarkers

<https://docs.sennetconsortium.org/biomarkers/>

Suryadevara, V., Hudgins, A.D., Rajesh, A. *et al.* SenNet recommendations for detecting senescent cells in different tissues. *Nat Rev Mol Cell Biol* (2024).  
<https://doi.org/10.1038/s41580-024-00738-8>



# HuBMAP and SenNet Features

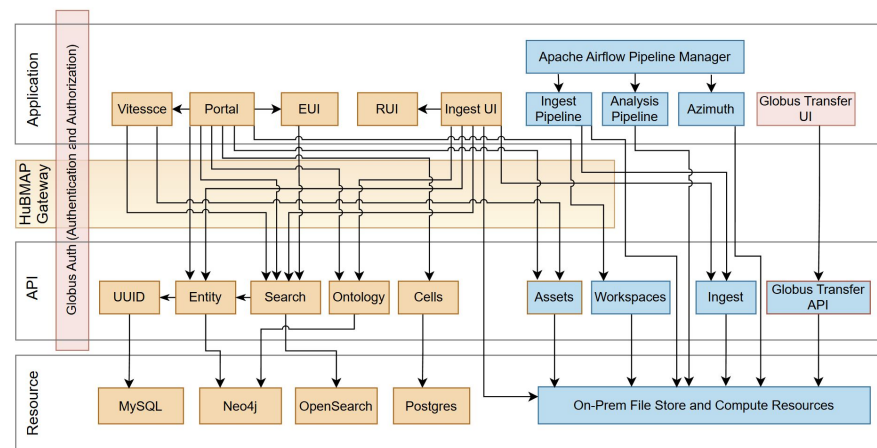
## Designed for FAIRness of Data and Software

- **Flexible hybrid cloud microservices architecture** for efficient data storage, easy software integration, low barriers to computing on data (**free**, leveraging PSC national resources)
- **Portable/interoperable/reusable/reproducible pipelines** with harmonized processing
- **Robust provenance** with unique identifiers
- **Ontologies and standardized and detailed metadata** with CEDAR templates
- **Community standards** for publishing findable and accessible APIs and data
- **Open software, data, and publication policies**

**Deeply collaborative:** PSC and Pitt jointly designed the architecture and build and run all the backend data infrastructure and collaborate with CMU CompBio, Indiana U., Harvard, New York Genome Center and U. Florida on tools.

**Software resources for atlas building:** dozens of github repositories and API endpoints, reused and updated across consortia

## Flexible Hybrid Cloud Microservices Architecture



HuBMAP Flexible Hybrid Cloud Microservices Architecture: AWS PSC Globus

**Human BioMolecular Atlas Program (HuBMAP): 3D Human Reference Atlas Construction and Usage.**  
Borner, et al. bioRxiv 2024.03.27.587041; doi: <https://doi.org/10.1101/2024.03.27.587041>

### Efficient data access and processing at scale:

thousands of datasets on-premises, leveraging investments by NSF and others to provide **free access** national scientific community

**Easy software integration:** Tools wrapped and integrated as-is using CWL and Docker, e.g. Azimuth

# HuBMAP Analysis Pipelines

- **sc/snRNA-seq:** Salmon quantification, Scanpy + scVelo downstream analysis
- **sc/snATAC-seq:** HISAT2 + ArchR
- **Sequencing spatial transcriptomics:** Salmon quantification, Scanpy + scVelo downstream analysis
  - Visium (without probes), Slide-seq
  - Harmonized with sc/snRNA-seq outputs, including output file format (addition of spatial coordinates for each capture bead)
- **Multiome RNA-seq & ATAC-seq:** subsets of RNA-seq and ATAC-seq pipelines used directly, embedded in multiome pipeline
- **Imaging (not currently represented in ATLAS-D2K):**
  - Multichannel imaging with segmentation masks: SPRM
  - CODEX (v1): Cytokit (+ SPRM)
  - PhenoCycler (CODEX v2), CellDIVE, MIBI: DeepCell (+ SPRM)
  - FISH (merFISH, DARTFISH): PIPEFISH pipeline
  - 3D segmentation and analysis: IMC complete, CODEX under development

The screenshot displays the HuBMAP data portal interface. At the top, it lists three data entries: HBM749.MT (snRNA-seq [Salmon] | Lung), HBM993.ZNNJ.876 (snRNA-seq (SNARE-seq2) [Salmon] | Kidney (Le...)), and HBM476.VHDB.268 (Slide-seq [Salmon] | Kidney (Right)). The HBM476.VHDB.268 entry is highlighted, showing its publication date (2022-02-14) and modification date (2022-02-14). Below this, the 'Visualization' section is active, displaying a 'Scatterplot (UMAP) 1,588' for 'Fallopian Tube (Right)' and a 'Spatial 1,952 spots, 1 i...' visualization. The main visualization area shows three side-by-side UMAP scatterplots: 'Scatterplot (UMAP - RNA)', 'Scatterplot (UMAP - ATAC)', and 'Scatterplot (UMAP - Multiome)'. Each plot shows a complex, multi-colored distribution of points. Below the plots, there are sections for 'Genomic Profiles' and 'Peak List 24,919 peaks'. A search bar is visible at the bottom right, with 'A1BG' and 'A1BG-AS1' listed as results.



## The CEDAR Workbench is an essential component of open science, ensuring FAIR data and enhancing scientific reproducibility

[Launch Workbench](#)

CEDAR is making data submission smarter and faster, so that scientific researchers and analysts can create and use better metadata. Through better interfaces, terminology, metadata practices, and analytics, CEDAR improves metadata from provider to end user.

2000  
users

51,875  
resources



120,829  
metadata

FAIR principles: Acronym for Findable, Accessible, Interoperable, and Reusable, which is a way of sharing data to maximize its utility

# 10x Multiome

Prepare your metadata based on the latest metadata schema using one of the template files below. See the instructions in the [Metadata Validation Workflow](#) document for more information on preparing and validating your metadata.tsv file prior to submission.

Related files:

-  [Excel template](#): For metadata entry.
-  [TSV template](#): Alternative for metadata entry.

REQUIRED - For this assay, you must also prepare and submit two additional metadata.tsv files following the metadata schemas linked here for [RNAseq](#) and [ATACseq](#). For additional documentation on this dataset type, please visit [here](#).

## Metadata schema

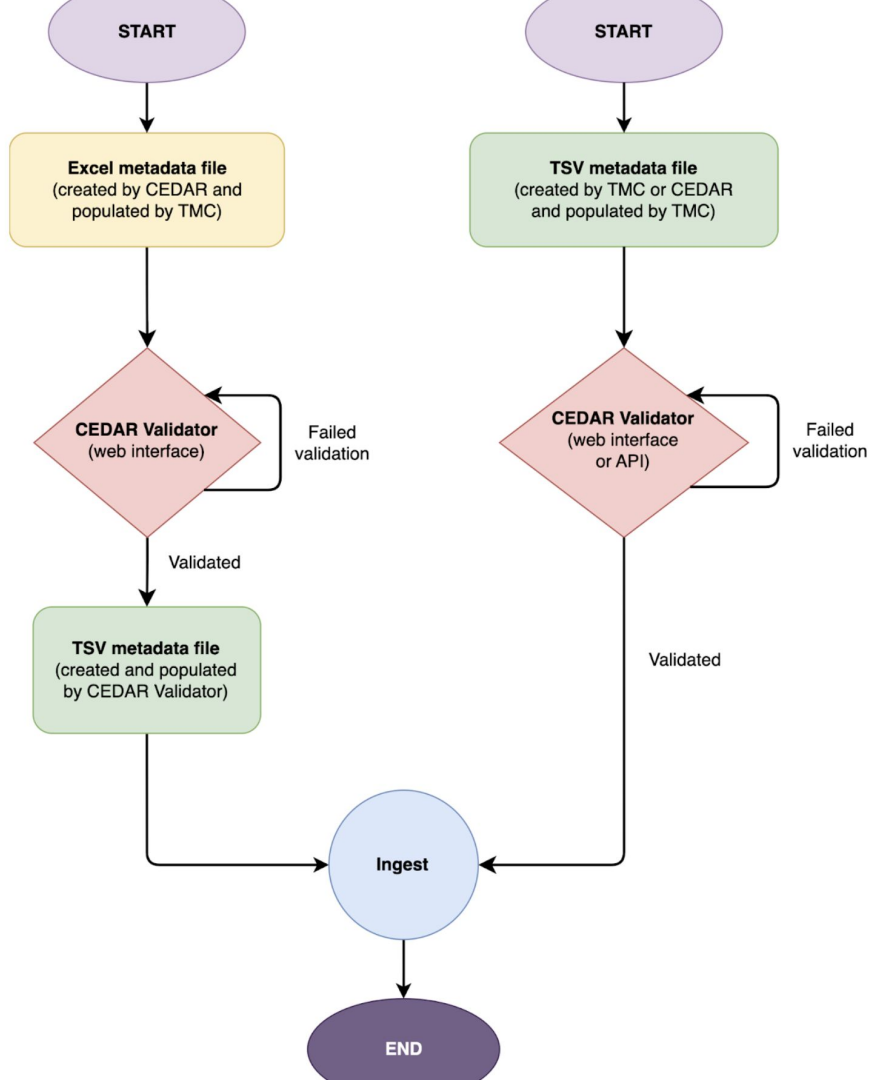
[Version 2 \(use this one\)](#)

## Directory schemas

**Version 2.0 (use this one)**

pattern	required?	description
<code>extras\/*.*</code>	✓	Folder for general lab-specific files related to the dataset.
<code>extras\expected_cell_count\.txt</code>		The expected cell count for the RNA sequencing dataset. This is an optional file that, if present, will be used by the HIVE's RNA sequencing analysis pipeline. With some datasets, knowing the expected cell count has improved the output of the HIVE analysis pipeline.
<code>raw\/*.*</code>	✓	All raw data files for the experiment.

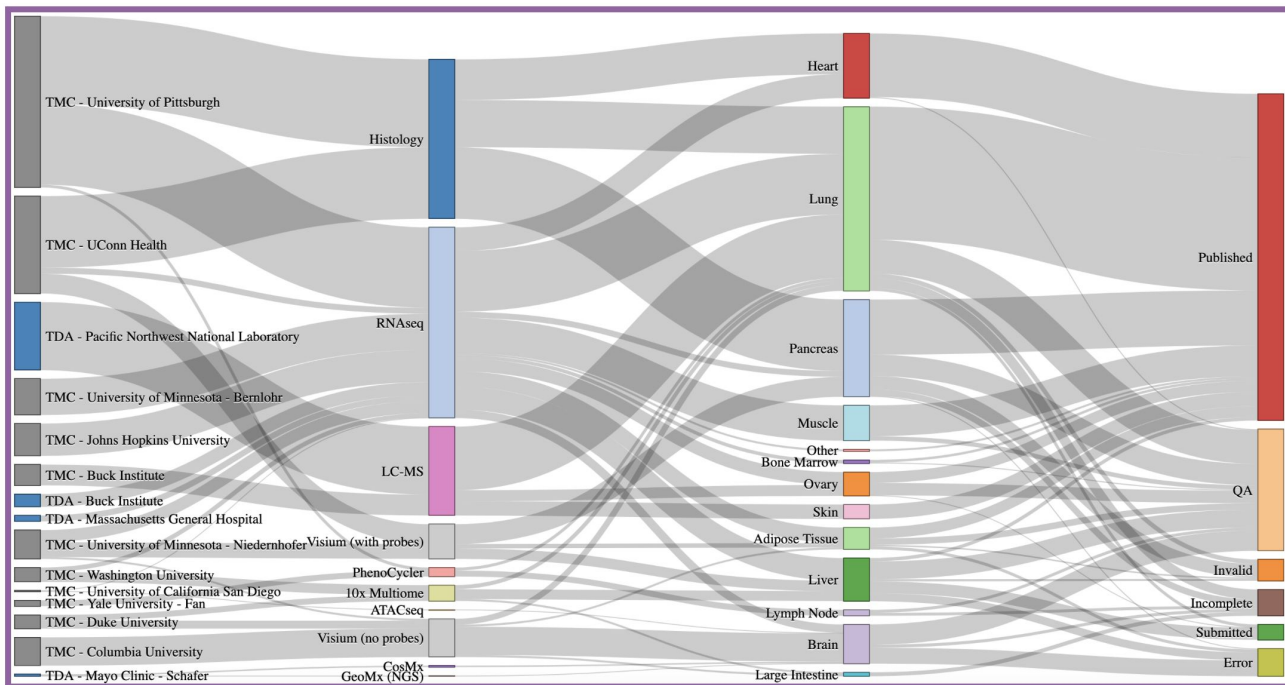






## Data Previews

Welcome to the SenNet data preview page where you can explore regular updates and visualizations of data being processed and published by the SenNet Consortium. This page acts as an overview of current Consortium data and a look ahead at data that is yet to be published.



This October 2024 SenNet Sankey diagram tracks how data has been uploaded and published through a variety of categories:

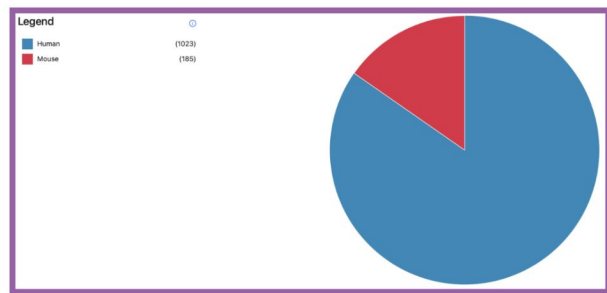
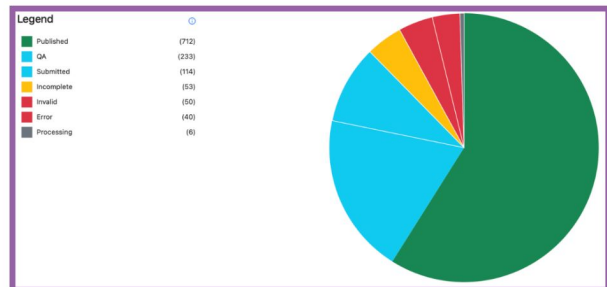
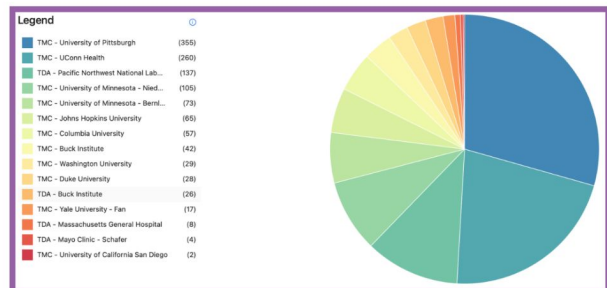
1. Which Tissue Mapping Center (TMC) or Technology Development and Application (TDA) processes the tissue.
2. Which technology is being used to process tissue.
3. The human or murine organ from which the senescent cell tissue was taken.
4. The status of that data in October 2024.

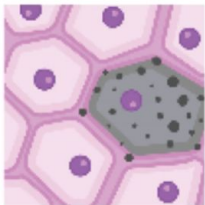
<https://docs.sennetconsortium.org/data-sankey/>



The Sankey visualization above and the graph and table below are created directly from the data dashboard, which is only accessible to Consortium members, and which details operational information about primary datasets at all statuses, enabling a preview of upcoming data and detailed tracking by Consortium members. The public sees only published datasets in the [data portal](#) whereas all data submitted is shared in the portal by Consortium members.

	10x Multiome	ATACseq	CosMx	GeoMx (NGS)	Histology	LC-MS	PhenoCycler	RNASeq	V/Isium (no probes)	V/Isium (with probes)	Xenium	Grand Total
<b>Adipose Tissue</b>								31	4	9		44
Human								4	4	9		17
Mouse								27				27
<b>Bone Marrow</b>								7				7
Human								7				7
<b>Brain</b>	1	3	1					17	57			79
Human									57			57
Mouse	1	3	1					17				22
<b>Heart</b>				82				48				130
Human				82				48				130
<b>Kidney (Left)</b>				20								20
Human				20								20
<b>Large Intestine</b>	4								4			8
Human	4								4			8
<b>Liver</b>	20			47				46		21		134
Human	20			47				22		21		110
Mouse								24				24
<b>Lung (Left)</b>	5			25	72			49	7			158
Human	5			25	72			45	7			154
Mouse								4				4
<b>Lung (Right)</b>	3			70	56	6	79	5				219
Human	3			70	56	6	75	5				215
Mouse							4					4
<b>Lymph Node</b>							17					17
Human							17					17
<b>Muscle</b>							71					71
Mouse							71					71
<b>Other</b>							4					4
Mouse							4					4
<b>Ovary (Left)</b>				22			22					44
Human				22			22					44
<b>Ovary (Right)</b>							4					4
Human							4					4
<b>Pancreas</b>				156			12		40	32		240
Human				156			12		40	32		240
<b>Skin</b>					29							29
Mouse					29							29
<b>Grand Total</b>	32	1	3	48	353	179	23	390	77	70	32	1208





## Cellular Senescence Network (SenNet) Method Development Community

MEMBERS 116

MORE ↓

Timeline Publications 161 Members 116 Discussions Resources News

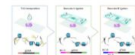
SEARCH

CATEGORY: ALL PUBLICATIONS ↕

SORT BY: DATE ↕

### Yale University\_Spatial ATAC Sequencing for Fixed Fresh Frozen Human Lymph node Tissue via DBiT-seq

Oct 22, 2024 PUBLICATION Negin Farzad



Negin Farzad<sup>1</sup>, Yanxiang Deng<sup>1</sup>, Archibald Enniful<sup>1</sup>, yao.lu.yl<sup>1</sup>, Rong Fan<sup>1</sup>

<sup>1</sup>Yale University

Views 38

Workspaces:





# SenNet Consortium

Software used for the Cellular Senescence Network (SenNet)

- Overview
- Repositories 21
- Projects 2
- Packages
- People 4

## Popular repositories

**documentation** Public

SenNet Documentation

JavaScript ☆ 1

**ingest-validation-tools** Public

Forked from [hubmapconsortium/ingest-validation-tools](#)

SenNet data submission guidelines, and tools which check that submissions adhere to those guidelines.

Python ☆ 1

**ccf-preview** Public

SenNet HRA User Interfaces (Preview)

HTML

**aws-workflow** Public

Python

**portal-ui** Public

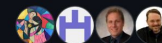
JavaScript 🗨️ 1

**entity-api** Public

A set of web service calls to return information about SenNet entities

Python

## People



## Top languages

- Python
- JavaScript
- Shell
- HTML
- Dockerfile

## Repositories

Find a repository...


Type

Language

Sort

**portal-ui** Public

JavaScript ☆ 0 🗨️ 1 🔄 21 📄 0 Updated 14 hours ago



**entity-api** Public

A set of web service calls to return information about SenNet entities



## 2,430 Works

### Creators & Contributors ?

<input type="checkbox"/>	Börner, Katy	173
<input type="checkbox"/>	Quardokus, Ellen M.	106
<input type="checkbox"/>	Radtke, Andrea J.	55
<input type="checkbox"/>	Germain, Ronald	32
<input type="checkbox"/>	Kelleher, Neil	29
<input type="checkbox"/>	Saunders, Diane	24
<input type="checkbox"/>	Browne, Kristen	21
<input type="checkbox"/>	Weber, Griffin	19
<input type="checkbox"/>	Beuschel, Rebecca T.	14
<input type="checkbox"/>	Gustilo, Katherine	11

### Publication Year

<input type="checkbox"/>	2024	611
<input type="checkbox"/>	2023	762
<input type="checkbox"/>	2022	556
<input type="checkbox"/>	2021	188
<input type="checkbox"/>	2020	313

### Work Type

<input checked="" type="checkbox"/>	Dataset	2,430
-------------------------------------	---------	-------

## Anatomical Structures, Cell Types, plus Biomarkers (ASCT+B) table for Bone Marrow and Blood/Pelvis v1.0

Muzlifah A. Haniffa, Laura Jardine & Sarah A. Teichmann

Text File published 2021 in [The Human BioMolecular Atlas Program \(HuBMAP\) Reference Repository](#)

Anatomical Structures, Cell Types, plus Biomarkers (ASCT+B) tables aim to capture the nested part\_of structure of anatomical human body parts, the typology of cells, and biomarkers used to identify cell types. The tables are authored and reviewed by an international team of experts.

DOI registered March 29, 2021 via DataCite.



Dataset

 <https://doi.org/10.48539/hbm963.tbfp.428>

## Anatomical Structures, Cell Types, plus Biomarkers (ASCT+B) table for Brain v1.0

Song-Lin Ding, Jeremy A. Miller & Amy Bernard

Text File published 2021 in [The Human BioMolecular Atlas Program \(HuBMAP\) Reference Repository](#)

Anatomical Structures, Cell Types, plus Biomarkers (ASCT+B) tables aim to capture the nested part\_of structure of anatomical human body parts, the typology of cells, and biomarkers used to identify cell types. The tables are authored and reviewed by an international team of experts.

### Publication Year

2024 687

### Work Type

Dataset 687

### Registration Agency

DataCite 687

## 687 Works

### Histology data from the pancreas of a 39-year-old white male

Paul Robson, Anne Marchini & Tim Adams

Content published 2024 in [Cellular Senescence Network \(SenNet\)](#)

DOI registered March 7, 2024 via DataCite.

[Dataset](#)

 <https://doi.org/10.60586/snt899.hzkd.452>

### Histology data from the pancreas of a 39-year-old white male

Paul Robson, Anne Marchini & Tim Adams

Content published 2024 in [Cellular Senescence Network \(SenNet\)](#)

DOI registered March 7, 2024 via DataCite.

[Dataset](#)

 <https://doi.org/10.60586/snt759.kjjz.639>

Advanced Search

Search for datasets Search

Showing all results

Clear All Sources: HuBMAP

### Search Filters

Clear All

Date

Start date: 2020 End date: 2024  
 01/01/2020 12/31/2024

Submit Reset

Include resources with no date information.

Type

Sources

Search sources filters

IID Repositories

AccessClinicalData@NIAID 0

2,766 Results

Best Match 10 results Download Metadata

<< < 275 276 277 > >>

### DATASET

[HBM622.JXWQ.554](#)

2023-11-03

CODEX [Cytokit + SPRM] data from the lymph node of a 10-year-old white male

Metadata Completeness ⓘ

Measurement Technique Pathogen Species Health Condition Variable Measured Funding License Usage Info Show metadata +

Topic Categories ⓘ: [Cell biology](#) [Microbiology](#)

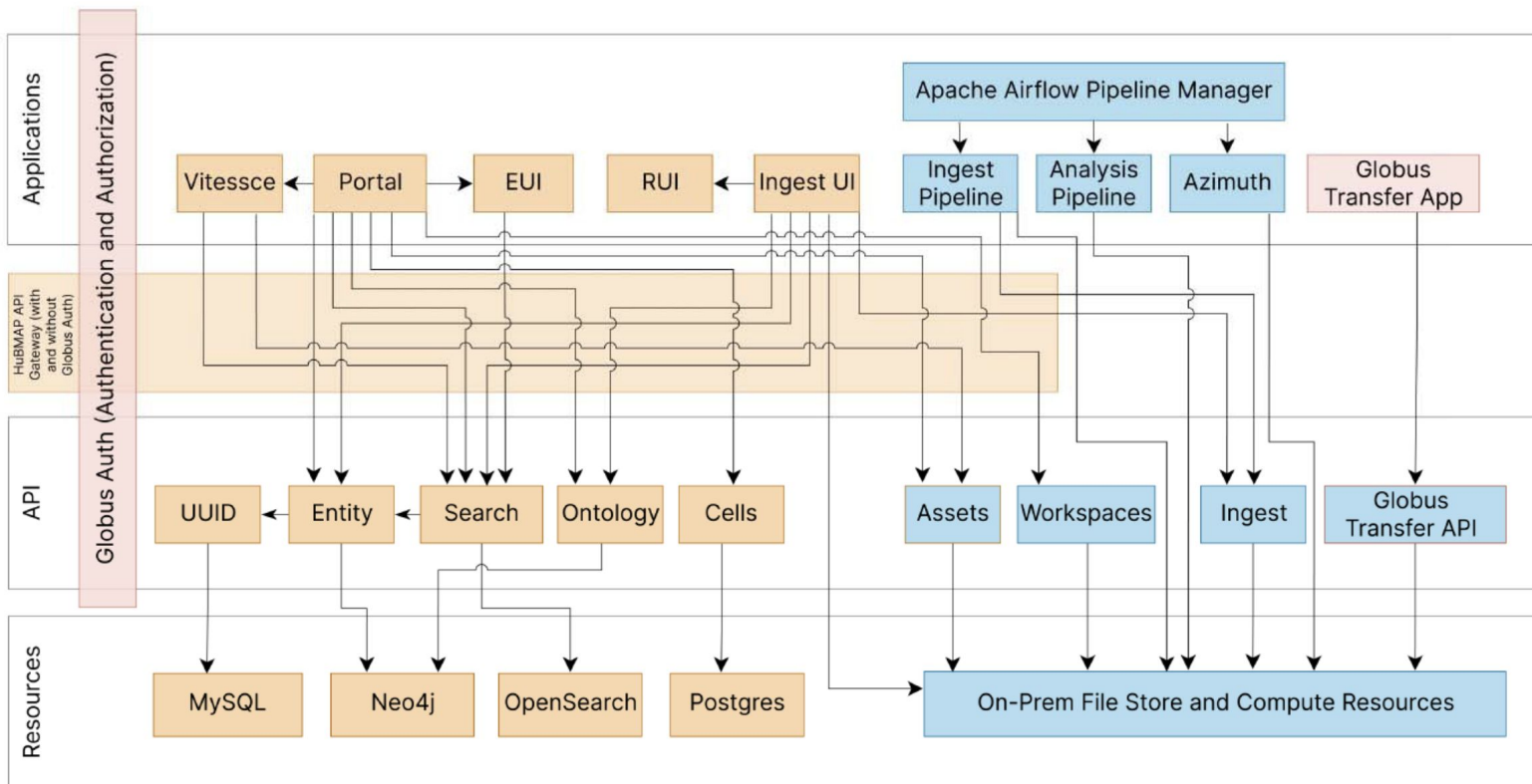
Provided by HuBMAP View resource



# Flexible hybrid cloud infrastructure -> atlas

- 1) **Curation and Ingestion:** Semi-automated data ingestion currently from data providers, and in the future from community partners, and the general research community, to maximize efficiency and usefulness for building the HRA
- 2) **Integration:** Automated analysis and annotation of ingested data and alignment of these annotations to the HRA via the UBKG
- 3) **Findability and Accessibility:** Manifestation of backend resources in the modular architecture of APIs and containers, services, and documentation that currently minimizes user friction in integrated searching, querying, analyzing, and viewing of data, and in the future of tissue maps at multiple spatial scales and among multiple layers of information
- 4) **Interoperability:** Use of the deployment of the UBKG to translate data, HRA assets, and community data among one another via ontologies
- 5) **Analysis:** Infrastructure support to currently enable users with interactive analyses of HuBMAP data via Jupyter notebooks, and in the future, batch workflows among both HuBMAP and user-contributed data and tools, including integration and mapping against the HRA
- 6) **Sustainability:** HuBMAP's flexible hybrid cloud infrastructure—efficiently leveraging on-premises resources at PSC for services that would incur significant public cloud charges, such as data storage, processing, analysis, and download facilitates sustainment of open tools, data, and infrastructure beyond the end of the HuBMAP program.

# Flexible hybrid cloud infrastructure -> atlas



HuBMAP Flexible Hybrid Cloud Microservices Architecture:

AWS    PSC    Globus

# Human BioMolecular Atlas Program Data Portal

An open platform to discover, visualize and download standardized healthy single-cell tissue data

**HBM969.MLWK.466**  
Kidney (Left)  
PAS Stained Microscopy  
Modified 2022-03-01

**Dataset Metadata**

**Dataset Type**

- 2D Imaging Mass Cytometry 13
- 3D Imaging Mass Cytometry 3
- ATACseq 96
- Auto-fluorescence 140
- CODEX 128
- Cell DIVE 12

HubMAP ID	Organ	Data Type	Status	Last Modified
<input type="checkbox"/> HBM978.COGC.988	Kidney (Left)	PAS Stained Microscopy		
<input type="checkbox"/> HBM223.DJGM.264	Kidney (Right)	Auto-fluorescence Microscopy		
<input type="checkbox"/> HBM422.KHRJ.366	Brain	MUSIC		
<input type="checkbox"/> HBM666.FFW.363	Heart	sciATACseq		
<input type="checkbox"/> HBM672.TRVG.967	Lung (Right)	snRNAseq (10x Genomics v3)		
<input type="checkbox"/> HBM655.RVNL.232	Large Intestine	snRNAseq (10x Genomics v3)		
<input type="checkbox"/> HBM596.PZBR.726	Small Intestine	Whole Genome Sequencing		
<input type="checkbox"/> HBM596.PZBR.726	Kidney (Right)	PAS Stained Microscopy		

**Gene Expression (per Distribution)**

**Cluster Membership**

**Discover**

Find data with our faceted search or explore by biological entities of organs, molecules or cell types.

[Explore datasets](#)

[Explore molecules/cell types](#)

**Visualize**

Explore spatial and single-cell data through powerful visualizations to gain deeper insights for your research.

[Visualize data with Workspaces](#)

**Download**

Preview files with our built-in file browser and download datasets from Globus or dbGaP straight to your device.

[Find datasets to download](#)

**What's New?**

Stay up to date with the latest HuBMAP Data Portal developments.

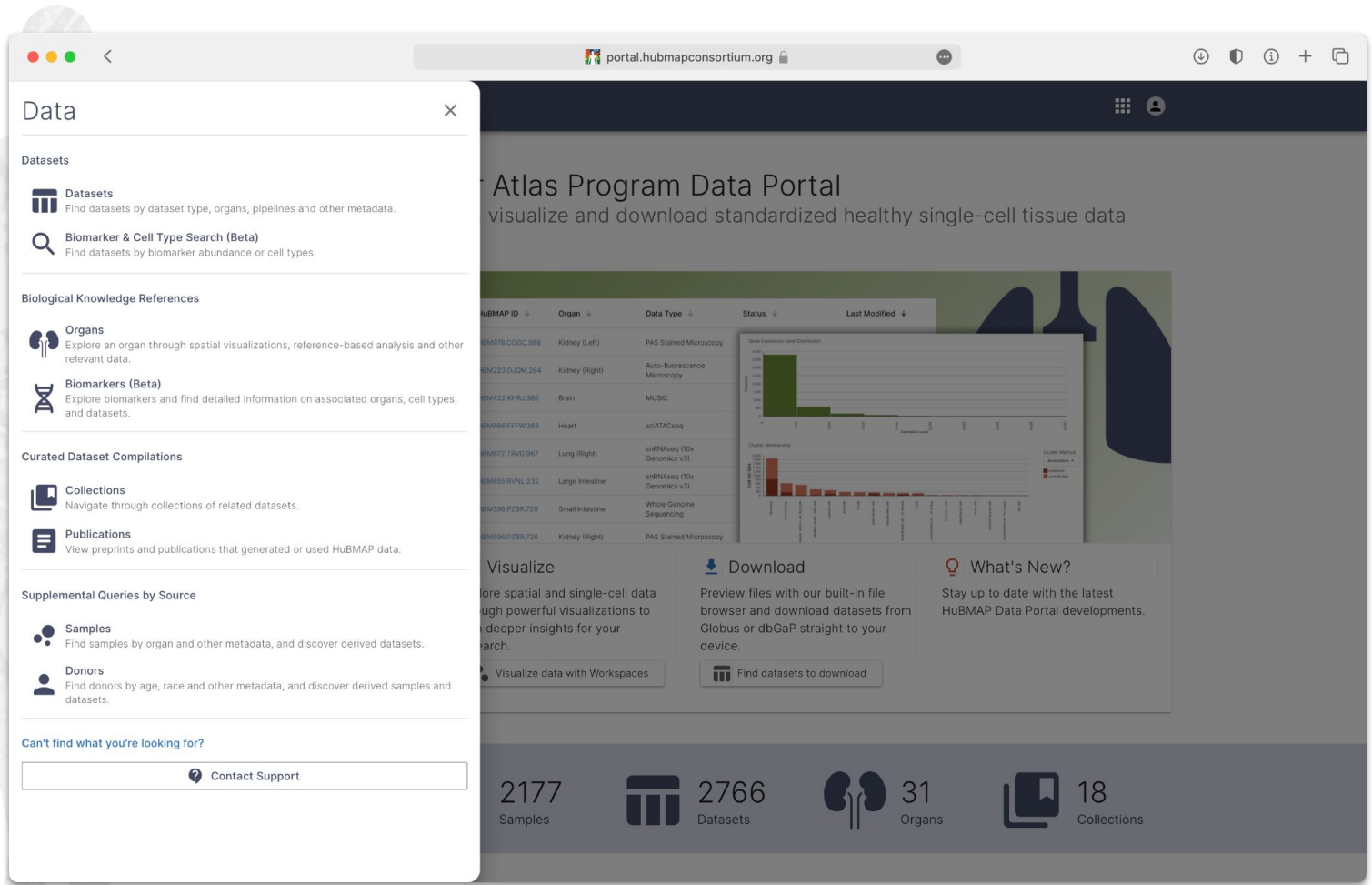
233  
Donors

2177  
Samples

2766  
Datasets

31  
Organs

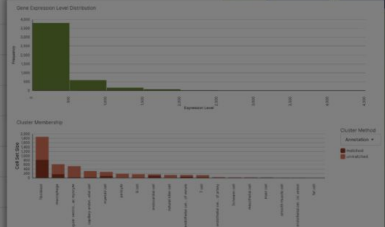
18  
Collections



# Atlas Program Data Portal

visualize and download standardized healthy single-cell tissue data

HUBMAP ID	Organ	Data Type	Status	Last Modified
BM978.C0GC.888	Kidney (Left)	PAS Stained Microscopy		
BM223.DJOM.264	Kidney (Right)	Auto-fluorescence Microscopy		
BM422.KHRL366	Brain	MUSIC		
BM696.FFW3K3	Heart	sciATACseq		
BM672.TAVG.967	Lung (Right)	snRNAseq (10x Genomics v3)		
BM855.RVNL.232	Large Intestine	snRNAseq (10x Genomics v3)		
BM596.PZBR.726	Small Intestine	Whole Genome Sequencing		
BM596.PZBR.726	Kidney (Right)	PAS Stained Microscopy		



### Visualize

Explore spatial and single-cell data through powerful visualizations to gain deeper insights for your research.

Visualize data with Workspaces

### Download

Preview files with our built-in file browser and download datasets from Globus or dbGaP straight to your device.

Find datasets to download

### What's New?

Stay up to date with the latest HuBMAP Data Portal developments.

2177 Samples   2766 Datasets   31 Organs   18 Collections

<https://portal.hubmapconsortium.org/>

# Datasets

**Getting Started**

Welcome to the HuBMAP Data Portal. Get a quick tour of different sections of the dataset search page.

[Begin The Dataset Search Tutorial](#)

Search:  Metadata

- Dataset Metadata
- Dataset Type
- 10X Multiome 132
  - 2D Imaging Mass Cytometry 13
  - 3D Imaging Mass Cytometry 3
  - ATACseq 467
  - Auto-fluorescence 140
  - CODEX 255
  - Cell DIVE 12
  - DESI 15
  - Histology 170
  - LC-MS 287
  - Light Sheet 3
  - MALDI 89
  - MIBI 211
  - MUSIC 14
  - RNAseq 708
  - Slide-seq 84
  - Visium (no probes) 148
  - WGS 17
  - seqFish 18
- Organ

<input type="checkbox"/>	HubMAP ID	Group	Data Types	Organ	Status	Last Modified
<input type="checkbox"/>	HBM648.BZL.557	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Right)	Published	2024-09-19 20:00:48
<input type="checkbox"/>	HBM896.TKHM.928	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Right)	Published	2024-09-19 20:00:45
<input type="checkbox"/>	HBM826.XPQJ.825	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Left)	Published	2024-09-19 20:00:42
<input type="checkbox"/>	HBM253.XDVK.565	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Right)	Published	2024-09-19 20:00:40
<input type="checkbox"/>	HBM852.RFCW.549	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Left)	Published	2024-09-19 20:00:37
<input type="checkbox"/>	HBM762.QGCB.476	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Left)	Published	2024-09-19 20:00:35
<input type="checkbox"/>	HBM483.LBWF.659	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Right)	Published	2024-09-19 20:00:32
<input type="checkbox"/>	HBM365.XQL.727	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Left)	Published	2024-09-19 20:00:29
<input type="checkbox"/>	HBM789.DPKN.375	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Left)	Published	2024-09-19 20:00:27

portal.hubmapconsortium.org/search?entity\_type[0]=Dataset

**HuBMAP** Data Resources

Suspension 200  
 organ 4  
**Status** ^  
 Published 2766 v

**Dataset Processing** ^  
**Dataset Category** ^  
 Raw 2030  
 Processed 736

**Pipeline** ^  
 Salmon 306  
 Cytokit + SPRM 127  
 SnapATAC 107  
 Salmon + Scanpy 74  
 Salmon + ArchR + Muon 66  
[View all](#)

**Visualization Available** ^  
 False 1692  
 True 1074

**Processing Type** ^  
 HuBMAP 723  
 Lab 13

**Assay Modalities** ^  
 Single 2354  
 Multiple 412

**Component Dataset** ^  
 False 280  
 True 132

Donor Metadata v  
 Affiliation v

<input type="checkbox"/>	HBM854.CKSB.432	TMC	Segmentation]	(Left)	Published	20:00:07
<input type="checkbox"/>	HBM858.PFX.737	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Right)	Published	2024-09-19 20:00:04
<input type="checkbox"/>	HBM564.NQJK.936	Vanderbilt TMC	PAS Stained Microscopy [Kaggle-1 Glomerulus Segmentation]	Kidney (Left)	Published	2024-09-19 20:00:02

[Previous](#)
1
2
3
4
...
[Next](#)

2766 Results Found

**HuBMAP**

[About](#)  
[Project Website](#)  
[Documentation](#)

[Software](#)  
[GitHub](#) [Services](#)

[Policies](#)  
[Overview](#)  
[Data Sharing Policy](#)

[Funding](#)  
[NIH Common Fund](#)

[https://portal.hubmapconsortium.org/search?entity\\_type%5B0%5D=Dataset](https://portal.hubmapconsortium.org/search?entity_type%5B0%5D=Dataset)

# Donors


Search Metadata

Donor Metadata

**Sex** ^

- Female 141
- Male 92

**Age** ^




**Race** ^

- White 152
- Black or African American 41
- Unknown 27
- Asian 11
- American Indian or Alaska native 1

[View all](#)

**BMI** ^



**Affiliation** v

<input type="checkbox"/>	HubMAP ID ↓	Group ↓	Age ↓	BMI ↓	Sex ↓	Race ↓	Last Modified ↓
<input type="checkbox"/>	<a href="#">HBM534.PKFT.943</a>	TTD - University of San Diego and City of Hope	90		Female	White	2024-04-03 03:56:03
<input type="checkbox"/>	<a href="#">HBM358.KMTL.795</a>	TTD - University of San Diego and City of Hope	90		Male	White	2024-04-03 03:54:38
<input type="checkbox"/>	<a href="#">HBM639.MJWP.689</a>	TTD - University of San Diego and City of Hope	59		Female	White	2024-04-03 03:52:39
<input type="checkbox"/>	<a href="#">HBM674.FJGN.569</a>	TTD - University of San Diego and City of Hope	60		Female	White	2024-04-03 03:50:51
<input type="checkbox"/>	<a href="#">HBM379.BFST.275</a>	TTD - University of San Diego and City of Hope	79		Male	White	2024-04-03 03:49:17
<input type="checkbox"/>	<a href="#">HBM543.JFMX.339</a>	TTD - University of San Diego and City of Hope	71		Male	White	2024-04-03 03:47:53
<input type="checkbox"/>	<a href="#">HBM448.VMXG.726</a>	TTD - University of San Diego and City of Hope	82		Female	White	2024-04-03 03:46:31
<input type="checkbox"/>	<a href="#">HBM668.XRTH.688</a>	TTD - University of San Diego and City of Hope	82		Female	White	2024-04-03 03:44:33
<input type="checkbox"/>	<a href="#">HBM263.SKPP.554</a>	TTD - University of San Diego and City of Hope	75		Female	White	2024-04-03 03:42:48
<input type="checkbox"/>	<a href="#">HBM993.RDQV.873</a>	TTD - University of San Diego and City of Hope	75		Female	White	2024-04-03 03:40:45
<input type="checkbox"/>	<a href="#">HBM763.FTCX.279</a>	TTD - University of San Diego and City of Hope	71		Male	White	2024-04-03 03:39:14

HBM275.QBCZ.562 Female Black or African American 54 year

Summary View

Narrow View

### Contents

- Summary
- Metadata
- Derived Data
- Provenance**
- Protocols
- Attribution

Key	Value
abo_blood_group_system	Blood Type A
age_unit	year
age_value	54
body_mass_index_unit	kg/m2
body_mass_index_value	20.5
cause_of_death	Anoxia

### Derived Data

View Data On Search Page

Samples (18)

Datasets (56)

HuBMAP ID	Organ	Sample Category	Derived Dataset Count	Last Modified
HBM892.NGDZ.434		block	8	2022-04-12
HBM335.KFLW.753		organ	32	2022-04-11
HBM742.FKCG.594		block	8	2022-06-08
HBM786.SSGC.992		block	8	2022-04-12

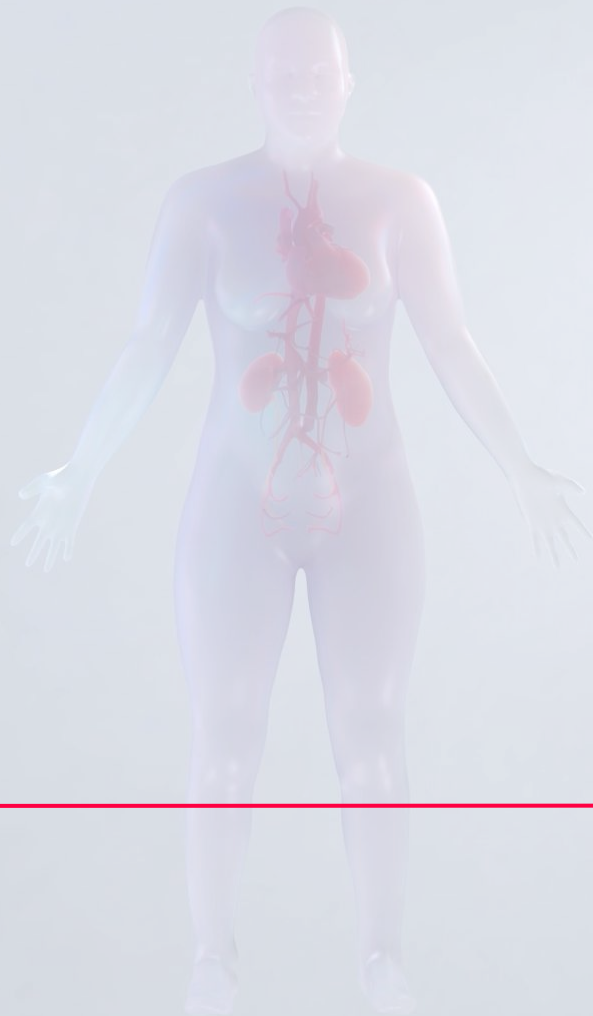
### Provenance

The provenance displays the sequence of events and actions that led to the creation of this donor.



# Q&A

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<https://humanatlas.io/events/2024-24h>

# Questions

How do we best capture data for a Multiscale Human?

How do we map a Multiscale Human?

How do we model a Multiscale Human?

How can LLMs or RAGs be used to advance science and clinical practice?

How to safeguard privacy?

**Thank you**

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