BIRNLex & The NIF Ontology:

Decomposing complex semantic domains to empower ontology-driven data federation

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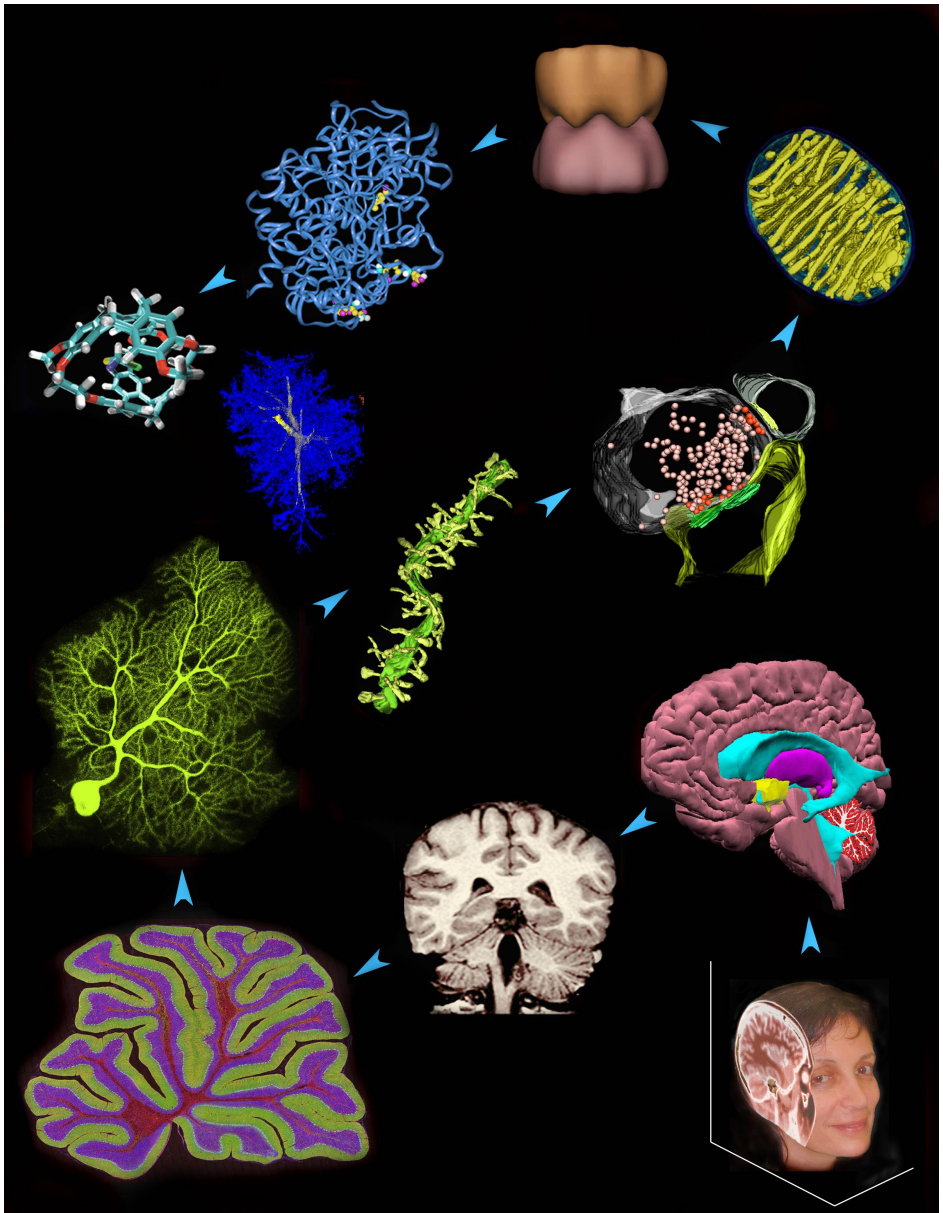
NCMIR BIRN BIOMEDICAL INFORMATICS RESEARCH NETWORK UCSD

Biomedical **I**nformatics **R**esearch **N**etwork

http://www.nbirn.net



The BIRN is a shared biomedical IT infrastructure designed to hasten the derivation of new understanding and treatment of disease through use of distributed knowledge

- ***Enables distant collaboration*** *(technical, scientific, social and political) between disparately located groups with different expertise and resources*
- *Provides **technical infrastructure to support collaboration**, designed to be extensible to other biomedical communities, e.g. bringing transparent GRID Computing to Biomedical Research*
- *Policy: **Open access and dissemination** of data and tools (i.e. Open Source)*

BIRN Testbeds

Currently, three driving biomedical projects focused on *neurodegenerative disease* to guide infrastructure requirements. Development is performed by the **BIRN Coordinating Center (BIRN-CC)** staff

- Morphometry BIRN
  - Promote shared morphometric analysis of human MRI
  - PI: Bruce Rosen, Harvard/MGH, (NCRR award U24RR21382)
- Function BIRN
  - Correlate behavioral and cognitive assessments with functional MRI imaging
  - PI: Steven Potkin, UC Irvine, (NCRR award U24RR21382)
- Mouse BIRN
  - Study mouse models of disease - promote sharing across the model organism community
  - Move basic research to applications in clinical contexts
  - PI: Arthur Toga, UCLA, (NCRR award U24RR21760)

Morphometry BIRN: Overview

- **Scientific Goal**

correlates
human neuroanatomical data ⇔ clinical assessment reports

Diseases: Unipolar Depression, Mild Cognitive Impairment (MCI), Alzheimers Disease

- **Methods**
  - Support multi-site structural MRI clinical studies or trials
  - Multi-site MRI calibration, acquisition and analysis
  - Integrate advanced image analysis and visualization tools
  - Provide shared semantic context for representing both the clinical reports and MRI results (and increasingly gene expression)

- **Sites (9)**
  MGH, BWH, Duke, UCLA, UCSD, UCI, JHU, Wash U, MIT

Morph BIRN Use Case:
Decomposing Assessments and Disease - relate to function

Assessment
- Beck Depression Index
- Abnormal Involuntary Movement Scale
- Verbal Fluency Assessment
- Edinburgh Handness Inventory
- California Verbal Learning Test
- Structured Clinical Interview for DSM IV (SCID)
- Premorbid Adjustment Scale
- Hallucination Assessment
- InterSePT Scale for Suicidal Thinking

Function
- Affective State
- Motivation
- Reading
- Generative speech
- Walking/Gait
- Smoking

Disease
- Unipolar Depression
- MCI
- Alzheimer's Disease
- Nicotine Addiction
- Schizophrenia
- Autism
- Parkinson's Disease
- Huntington's Disease

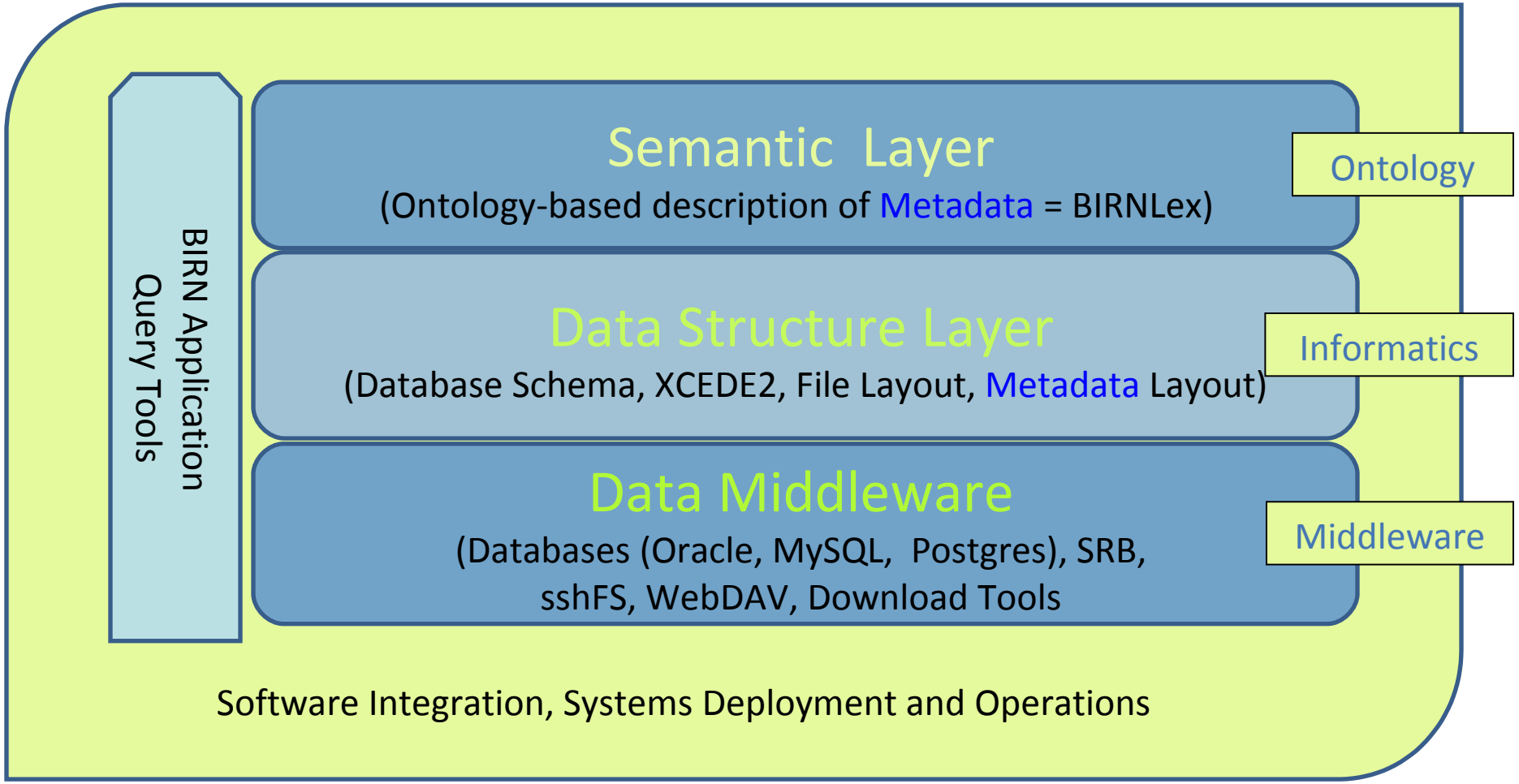
Morph BIRN Use Case: Long-term effects of Depression - link to AD?

- Find volumetric data/metadata from MRIs of humans with specific diagnosis(es)
  - Which regions are decreased/increased in size relative to normal controls?
  - Which regions show structural differences across a variety of diagnoses?
  - Which regions show connectivity changes across a variety of diagnoses?
  - Which regions show structures or connectivity differences across a variety of pharm Rx?

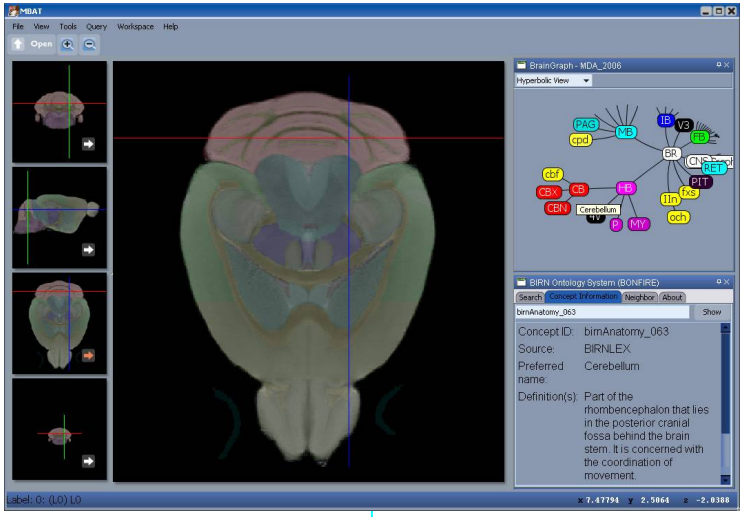
- Given a brain region which shows structural differences
  - Do these changed regions have commonalities (i.e. cell types, neurotransmitters, other afferent/efferent connections)?
  - What difference measures (size, transmitter, connections via DTI, etc.) between or within altered regions are correlated with one another? X-correlate with connectivity change?
  - Do certain changes correlate with specific diagnosis profile or pharm Rx for depression?

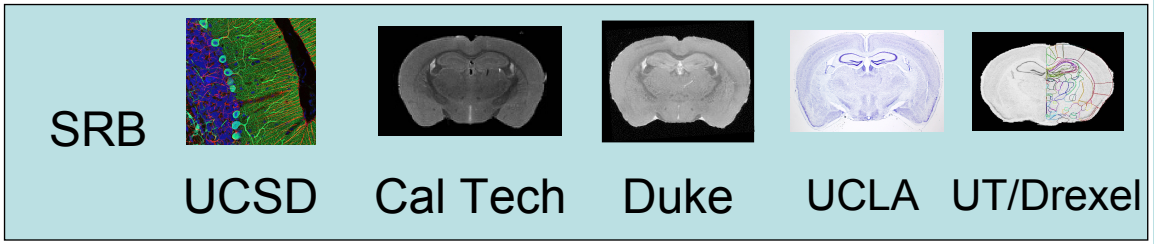
- Examine volumetric and connectivity (DTI) changes in humans with genotyping data for APOE status
  - Do altered brain regions or connectivity patterns correlate with APOE status? How does this change with age?
  - Do any changes correlate with the severity of depression diagnosed earlier?
  - How do these changes correlate with anti-depressant Rx?

BIRN Data Repository



The BIRN Mediator enables semantic integration across image repositories





Mediation Requirements:

- A rose is a rose
- A S. nigra is a S. nigra

Implementation Solution:

- infrastructure is ontology-centric
- Data Federation based on *shared* semantic framework
  - Source database data models must be mapped ontologically:
    - To determine equivalent fields
    - To search content in appropriate, shared semantic context
- Heavy-weight solution
  - Requires commitment
  - BIRN researchers funded to do this work

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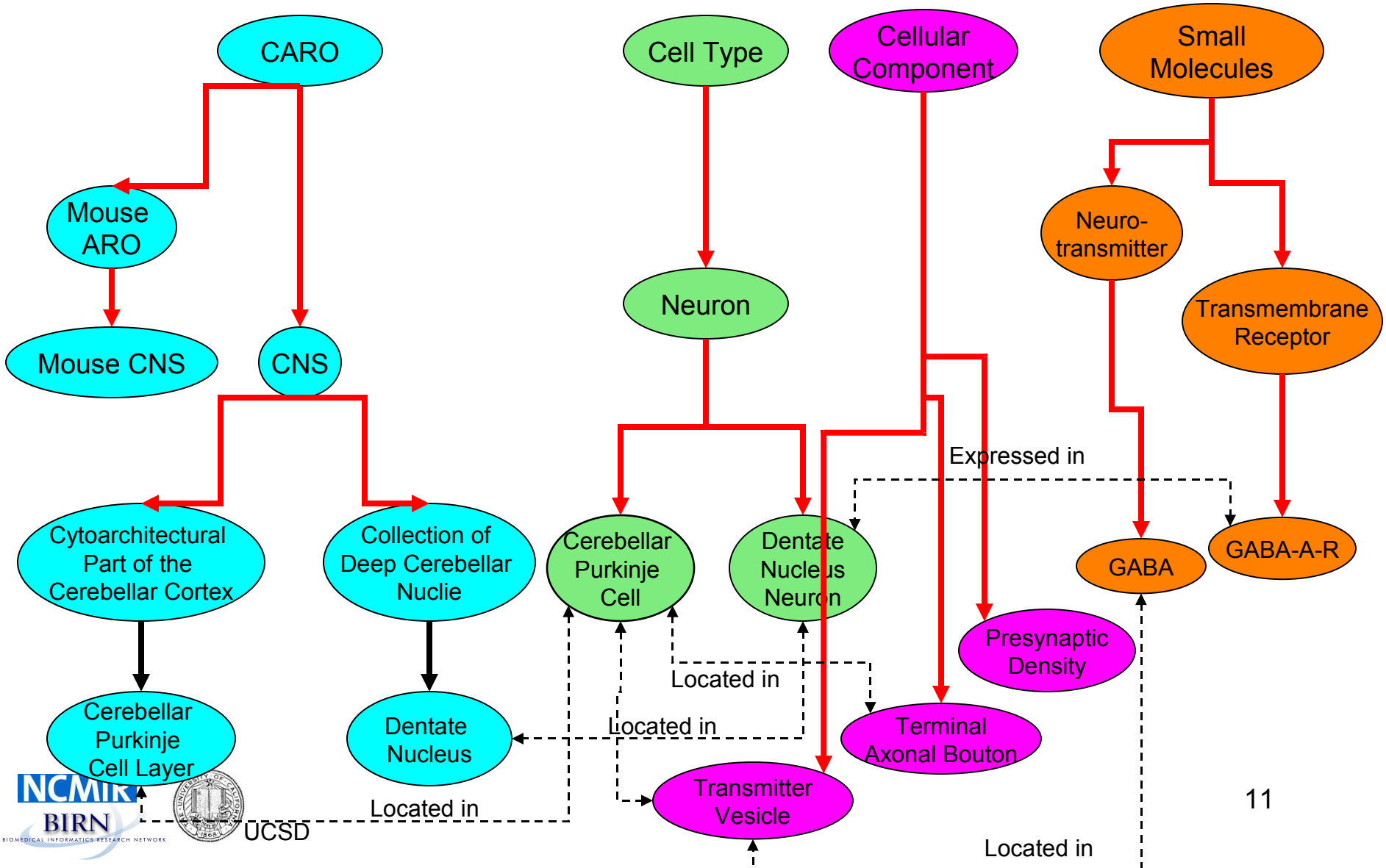
BIRNLex - The common language

- BIRNLex is an ontology-based lexicon
  - Curated by cross-BIRN Ontology Task Force
- It re-uses relevant terminologies, taxonomies, and ontologies, when they exist
- We add entities and relations as needed by BIRN researchers
- All organized and combined using current biomedical ontology development *best practices*
- Represented using standard formalism: OWL/RDF

BIRN Ontology TF

- Maryann Martone (chair) - UCSD/mBIRN
- Jeff Grethe - UCSD/BIRN-CC
- Amarnath Gupta - UCSD/SDSC/BIRN-CC
- Christine Fennema-Notestine - UCSD/Morph BIRN
- Jessica Turner - UCI/FBIRN
- Daniel Rubin (advisor) - NCBO
- David Kennedy(MGH/Harvard) - Morph BIRN
- Bill Bug (UCSD) - mBIRN & BIRN-CC

Objective: represent complex neuroscience domains Using shared community ontologies



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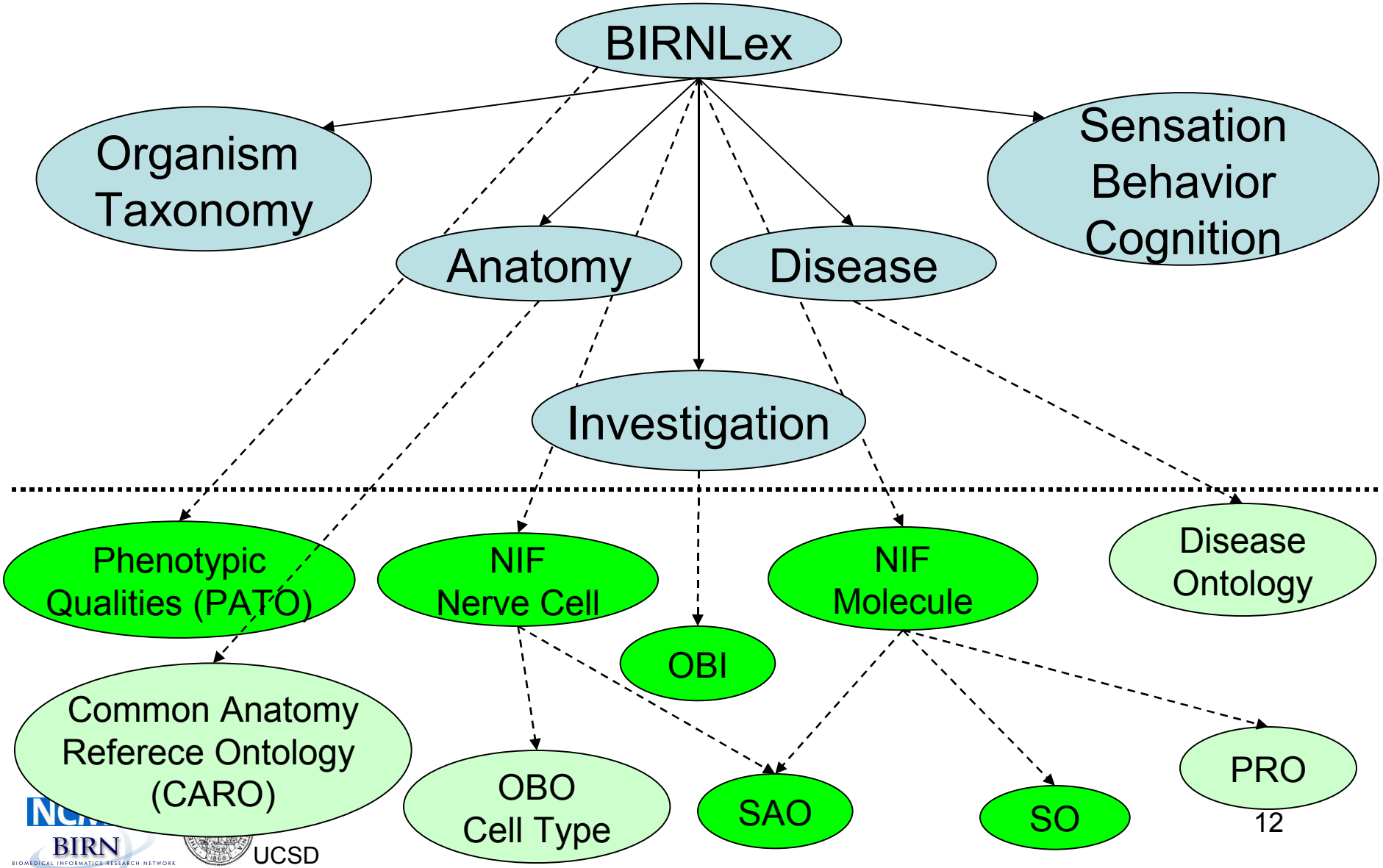
BIRNLex Components

http://purl.org/nbirn/birnlex/ontology/birnlex.owl



Neuroscience Information Framework (NIF)

- Large scoped neuroscience information management project designed to provide an ontology-driven search system

- Uses federated query mediation to make disparate data repositories, web-based resources, and a literature corpus collectively searchable via the NIF ontology (in OWL)

- http://nif.nih.gov/