eagle-i: biomedical research resource datasets

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Abstract. In this paper we present the linked data sets produced by the eagle-i project. We describe the content, the features and some of the applications currently leveraging these datasets.

Keywords: eagle-i, linked data, biomedical ontologies, research resources, research profiles

1. Introduction

An important facet of biomedical research is to access the right tools and research resources needed to answer specific biological questions. Biomedical resources are generated, purchased and used during the course of research. Information about such resources is often sequestered in lab notebooks or a lab’s digital records, making the resources difficult to find, share, and reuse. The goal of eagle-i, (http://www.eagle-i.net) - a two-year project funded by National Center for Research Resources (NCRR) - is to make these “invisible” research resources more discoverable by collecting information about them and making the information available through a semantically enabled, federated search system and as linked data sets.

The eagle-i architecture is composed of four main components: an underlying ontology; data collection tools comprising an ETL toolkit and a web-based, ontology driven Semantic Web Entry and Editing Tool (SWEET); institutional triple-store repositories; and a central web-based search application.

To support structured data collection, retrieval and publication, a modular set of ontologies, collectively known as the eagle-i Resource Ontology (ERO) was developed. The ontology contains domain representation for research resources including organisms, instruments, protocols, constructs, antibodies, biospecimens, human studies and research opportunities.

The data generated through eagle-i’s data collection tools are stored as triples in accordance with the Resource Description Framework (RDF) and made available through SPARQL endpoints (https://www.eagle-i.net/export/sparqlers/) and as linked data sets. In this paper we present the key characteristics of the datasets, provide sample SPARQL queries and discuss some of the known usages of our datasets.

2. Dataset Descriptions

Our datasets cover biomedical resources available at 25 institutions. The main resource types we collect and publish information about are:

- Biological Specimen
- Database
- Document
- Human Study
- Instrument
- Organism or Virus
- Organization
- Person
- Reagent
- Research Opportunity

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- Service
- Software

The datasets can be queried through SPARQL endpoints (https://www.eagle-i.net/export/sparqlers/), can be browsed using classic and linked data browsers and are available via direct RDF download (https://www.eagle-i.net/export/rdf-download/). Full information about each dataset is available at the Data Hub (http://datahub.io/dataset?q=eagle-i). In Table 1, we present a summary of the triples in each dataset; these numbers include only the triples that are directly related to resource instances, that is (a) triples where the subject is a resource instance and (b) triples that provide minimal information (type and label predicates) for the objects in (a). We deliberately exclude from these datasets most ERO ontology triples, as they do not per-se represent biomedical resources. Table 2 gives an overview of the number of triples across all participating institutions devoted to each of the resource types.

A URI for a resource resolves as an HTML page when accessed by a browser. This page displays all the data about the resource and its inferred types as well as a link to the RDF download (shown in Figure 1).

<table>
<thead>
<tr>
<th>Data set</th>
<th>Published triples</th>
</tr>
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<tbody>
<tr>
<td>University of Pennsylvania</td>
<td>21866</td>
</tr>
<tr>
<td>Vanderbilt University</td>
<td>19536</td>
</tr>
<tr>
<td>Oregon Health &amp; Science University*</td>
<td>76818</td>
</tr>
<tr>
<td>Harvard University*</td>
<td>372288</td>
</tr>
<tr>
<td>University of Alaska Fairbanks</td>
<td>15446</td>
</tr>
<tr>
<td>University of Hawai`i Manoa</td>
<td>347219</td>
</tr>
<tr>
<td>Jackson State University*</td>
<td>10501</td>
</tr>
<tr>
<td>Montana State University*</td>
<td>24737</td>
</tr>
<tr>
<td>Morehouse School of Medicine*</td>
<td>8763</td>
</tr>
<tr>
<td>Dartmouth College*</td>
<td>372288</td>
</tr>
<tr>
<td>University of Puerto Rico*</td>
<td>35733</td>
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<tr>
<td>Clark Atlanta University</td>
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</tr>
<tr>
<td>Charles Drew University</td>
<td>3940</td>
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<tr>
<td>The City College of New York, CUNY</td>
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</tr>
<tr>
<td>Florida Agricultural and Mechanical University</td>
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<tr>
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<tr>
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<tr>
<td>Meharry Medical College</td>
<td>1805</td>
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<tr>
<td>Ponce School of Medicine</td>
<td>2001</td>
</tr>
<tr>
<td>Texas Southern University</td>
<td>1474</td>
</tr>
<tr>
<td>Tuskegee University</td>
<td>927</td>
</tr>
<tr>
<td>Universidad Central del Caribe</td>
<td>5477</td>
</tr>
<tr>
<td>University of Texas at El Paso</td>
<td>2773</td>
</tr>
<tr>
<td>University of Texas at San Antonio</td>
<td>4050</td>
</tr>
<tr>
<td>Xavier University of Louisiana</td>
<td>9395</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td><strong>1345484</strong></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Resource type</th>
<th>Published triples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Biological specimens</td>
<td>48317</td>
</tr>
<tr>
<td>Databases</td>
<td>421</td>
</tr>
<tr>
<td>Documents</td>
<td>68890</td>
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<tr>
<td>Human studies</td>
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<td>Instruments</td>
<td>125696</td>
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<td>Organisms or viruses</td>
<td>456107</td>
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<td>Organizations</td>
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<td>People</td>
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<tr>
<td>Reagents</td>
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<tr>
<td>Research opportunities</td>
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</tr>
<tr>
<td>Services</td>
<td>62501</td>
</tr>
<tr>
<td>Software</td>
<td>27291</td>
</tr>
</tbody>
</table>

Number of triples published per site (Table 1) and per resource type (combined for all institutions) (Table 2) as of March 5, 2013.

* - Denotes status as member institution of original eagle-i network  
  " - Denotes status as currently funded, CTSA eagle-i member institution
3. Domain modeling

Our approach to domain modeling was driven by the following goals:

1. Reuse existing ontologies as much as possible to reduce the modeling burden and to maximize future data integration
2. Identify design patterns and ontology engineering solutions that would allow a set of ontologies to drive the eagle-i user interfaces while at the same time remaining of general use to the biomedical community at-large.

Regarding the first point, because most of our domain coverage was biomedical in nature, we referred to principles and existing ontologies within the OBO Foundry [1]. Conformance with OBO Foundry standards fixed the following design choices:

- Use of the Basic Formal Ontology [2] as the upper level ontology
- Predominant utilization of ontologies in the OBO Foundry constellation due to their quality, extensive usage and common design principles
- Application of the MIREOT principle [3] for referencing entities in external ontologies

So far as the second goal is concerned, we developed a design pattern approach to separate, within our ontology suite, the application-specific portion from the “core” content that was worth sharing with the community. Our approach, described extensively in [4], has been generalized and reused in other efforts such as the Reagent Ontology (ReO https://code.google.com/p/reagent-ontology/) and the Agent, Resource and Grant ontology (ARG https://code.google.com/p/connect-isf/). It has also led to a set of recommendations for implementing a maintenance and release pipeline using available tools and service [5].

Another key element of our ontology development process has been the coordination of efforts within the Biomedical Ontology Community. These include active collaboration and discussion with other ontology development groups (through tracker term requests, developer call participation, etc.). Although time consuming, these efforts allow easy reuse of portions of other ontologies and help achieve better data integration and interoperability.
Fig. 2. Classes can be searched for using the autocomplete feature (A). In this case, we see information about ‘transgenic organism’. All of the properties for this class are shown. Clicking the property name displays its definition, URI and annotations as shown by feature (B). Referenced taxonomies are sets of terms used as ranges for some properties (such as the Disease taxonomy for the related disease property) while embedded types denote classes for which instances can be only created in the context of another instance. For example, a construct insert can only be created in conjunction with its containing construct.
4. Sample SPARQL queries

We present some sample queries that illustrate interesting usages of the eagle-i datasets. For each one, we specify the particular SPARQL endpoint used such that query results can be reproduced.

In order to better understand the classes and relationships used in the following queries we suggest referring to the eagle-i ontology browser (http://search.eagle-i.net/model/), a screen capture from which can be seen in Figure 2.

URIs of eagle-i classes and properties can also be found using Ontobee [6] (http://www.ontobee.org/browser/index.php?o=ERO). The queries can be executed through the SPARQL interfaces (Fig.3) or passed programmatically to the endpoint.

In the following examples, we omit for brevity the declaration of the following prefixes:

```
PREFIX rdfs: <http://www.w3.org/2000/01/rdf-schema#>
PREFIX obo: <http://purl.obolibrary.org/obo/>
PREFIX mesh: <http://purl.bioontology.org/ontology/MSH/>
```

4.1. Query for researcher expertise

The eagle-i datasets allow identification of individual expertise by leveraging the connection between resources and related techniques, diseases and instruments as well as the linkage between resources and people. As an example, the query below identifies likely experts in radioimmunoassay techniques (obo:ERO_0000652) by connecting the nodes between the technique and individuals. In this case, collecting all resources that reference the technique (obo:ERO_0000543) and returning the person(s) indicated as contact (obo:ERO_000021) for that resource.

```
SELECT DISTINCT? person WHERE {
    ## Select Resources that have related ## technique radioimmunoassay
    ## Select the contact person for ## the resource
}
```

If the query is executed against the OHSU endpoint (http://ohsu.eagle-i.net/sparqler/query/) it will return a contact for a set of services that involve radioimmunoassay like the one represented in Fig. 1.

4.2. Query for animal models relevant for a particular disease

Another interesting query is related to the identification of animal models used in the research of autoimmune diseases. The query is reported below.

```
SELECT ?resource WHERE {
    ## Select Organism Resources
    ?resource a obo:OBI_0100026.
    ## That are model of some disease
    ## A and the disease is an autoimmune ## disease
    ?disease rdfs:subClassOf mesh:D001327.
}
```

It is interesting to note that the results returned for this query, when executed against the Harvard endpoint (https://harvard.eagle-i.net/sparqler/query/), include animal models related to Diabetes Mellitus, Type I (see for example: http://harvard.eagle-i.net/0000012a-25bf-7988-f5ed-943080000005) and Sjogren's Syndrome (http://harvard.eagle-i.net/0000012a-25bf-7988-f5ed-943080000003) because both are subsumed in the MeSH Hierarchy for Autoimmune disease (mesh:D001327).
4.3. Query for resources across datasets

For several resources in eagle-i (such as animal models) we collect information on related genes via Entrez gene IDs. This is a useful entry point for connecting non-eagle-i datasets. In the query below for instance, we probe data at the University of Puerto Rico (UPR) for resources relevant to Stony Brook investigators based on the genes they have published about. This query, when executed against the UPR endpoint (http://upr.eagle-i.net/sparqler/query/) return 4 authors related to an ErbB2 construct insert used in a particular plasmid (http://upr.eagle-i.net/i/0000012b-8c1f-c389-3bbe-1c0980000000).

```sparql
SELECT ?entrezgeneid ?author ?resource WHERE {
    ## Query the SPARQL endpoint at Stony Brook
    SERVICE <http://link.informatics.stonybrook.edu/sparql1/>{
        ## Get the AUIs and the CUIs related to the ## entrez gene ids
        ?au
        ## Select the papers that have as subject ## the gene identified by the Entrez ID
        ## Select the author of the paper
        ?paper <http://vivoweb.org/ontology/core#informati
        nResourceInAuthorship> ?authorship.
        ?authorship ?author
        <http://vivoweb.org/ontology/core#linkedAuthor
        or> ?author.
        ?author
        <http://vivoweb.org/ontology/core#hasMemberR
        ole> ?membership.}
        ## Bind the Entrez gene IDs
        ## to eagle-i resources
    }
}
```

5. Dataset Usage

A number of groups have begun to make use of eagle-i-produced data by implementing search and visualization tools that reuse the RDF and that complement the functionality provided by eagle-i applications. The Harvard Catalyst Core Facilities Portal (http://cbmi.catalyst.harvard.edu/cores/index.html) was an early adopter on this front. This portal component generates HTML pages from the core facilities data stored at Harvard University’s eagle-i repository. The automated production of these pages ensure that the service offerings as well as the contact information for each core are standardized, centralized and current, as they are maintained through the eagle-i SWEET.

Another interesting reuse of eagle-i RDF data is the CoreSearch service at Oregon Health & Science University (http://www.ohsu.edu/research/coresearch/).

Leveraging the Plumage tool (http://chtsiatucsf.github.com/plumage/), developed by the Clinical & Translational Science Institute at the University of California San Francisco, CoreSearch allows for visualization and search of OHSU core laboratories, their service offerings and their instruments by converting the RDF data accessed through the eagle-i SPARQL endpoints to static HTML pages that can be optimally indexed by Google and other search engines. Eagle-i datasets are also used in the context of CTSACConnect project (http://www.ctsacconnect.org/) to link clinicians to basic researchers through publications and research resources.

6. Updating maintenance and scalability

The creation of the eagle-i datasets over the three years of the project was possible as a result of the dedicated work of resource navigators (Ph.D. level scientists contacting laboratories and collecting resource information) and curators responsible for the data entry and quality control of collected data. After the grant ends, each institution in the network will be in charge of maintaining and updating their local datasets with the help of detailed guidelines and tools (https://open.med.harvard.edu/display/eaglei/Training). The eagle-i soft stackware, which is available as open source, will continue to be maintained and enhanced by the development team at Harvard with contributions from the open source community. The
Harvard team will also operate central components that tie the eagle-i network together (node registry, central search, global instances repository). The eagle-i ontology has been and will remain an open source community resource (https://code.google.com/p/eagle-i/) that is updated through tracker requests. Each new eagle-i software release incorporates the latest release of the eagle-i Resource Ontology. To assure data alignment in accordance with these regular ontology changes, the developers concurrently release data migration scripts to update each of the triple stores. Other sustainability efforts for data collection are related to the integration of the eagle-i backend with laboratory inventory management systems such as iLab (http://www.ilabsolutions.com/) and billing systems such as Vanderbilt CORES (http://www.mc.vanderbilt.edu/root/vumc.php?site=CORES).

7. Discussion

The eagle-i linked data is a suite of different datasets: one for each institution participating to the network. This choice was motivated by a desire to allow each institution to control their own search and data entry applications as well as to assign their own URIs for their respective published instance data.

The lack of a single SPARQL query interface to search over all of the eagle-i datasets at once, but is easily overcome using programmatic access. As an internal response, for curation purposes we have developed a simple web application that allows eagle-i curators to select multiple SPARQL endpoints and issue queries against them in bulk.

Another characteristic of our datasets is related to the usage of numeric URIs for most of the classes and properties. This choice was driven by the decision to adhere to the OBO Foundry Principles. From the perspective of ontology development, it makes sense to have the semantics of a particular resource be conveyed by its textual and logical definition rather than by a human readable URI or rdf:label. This is convenient to avoid misuse of entities when, for example, a label of a particular entity changes. This makes writing SPARQL queries less straightforward but we have found that good documentation of the ontology through the ontology browser and Ontobee are of great help for our end users.

Another problem we had to face while creating the dataset was related to particular “instances” that did not belong to any institution but were supposed to be “global” (i.e. used form each institution). Examples of this kind of instances are Organizations, Manufactures or any kind of resource that are not tied to particular institutions in our network. For these kinds of resources we use a particular name space and we store them in a dedicated repository.

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References