BioHackathon 2018 - Paris
http://bh2018paris.info/

Application of RDF-based models and tools for enhancing interoperable use of biomedical resources

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Project links
- http://dbcls.rois.ac.jp/
- http://biohackathon.org/
- http://med2rdf.org/

Background information

Background

Our group is mainly from two national bioinformatics centers in Japan

- National Bioscience Database Center (NBDC)
  - Governmental funding agency
- Database Center for Life Science (DBCLS)
  - National institute for database technologies

Running NBDC/DBCLS BioHackathon meeting over the past 10 years

- Mission: Integration of databases in Life Sciences and Biomedical domains
  - Improvement of Standardization and Interoperability of DBs
  - Currently through the Semantic Web technologies
    - Standard ontologies and identifiers
    - Promoting to provide and use of RDF-based data resources
    - Development of applications on top of the RDF data

RDF resources currently available

- Nucleotide seq & annot
  - INSDC (DDBJ/DBCLS)
- Genome
  - Ensembl (EBI)
  - RefSeq (TogoGenome)
- Protein seq & annot
  - UniProt (SIB)
- Protein structure
  - PDB (PDBj)
  - BMRB (PDBj)
  - FAMSBASE (Chuo U)
- Compounds
  - PubChem (NCBI)
  - ChEMBL (EBI)
  - Nikkaji (JST)
- Gene expression
  - RefEx (DBCLS)
  - ExpressionAtlas (EBI)
- Samples
  - BioSamples (EBI/DDBJ)
  - JCM (RIKEN)

- Biomedical (Med2RDF)
  - ICGC, COSMIC, CIViC
  - DGIdb, OpenTG-Gates
  - ClinVar, dbSNP, dbVar
  - ExAC, gnomAD
  - HiNT, INstruct
- Glycome
  - GlyTouCan, GlycoEpitope, WURCS, GGDonto, PAConto
- Proteome
  - jPOST
  - The Human Protein Atlas
- Pathway
  - Reactome (EBI)
- Others
  - MeSH (NCBI)
  - BioModels (EBI)
  - MBGD (NIBB/DBCLS)
  - Quanto (DBCLS)

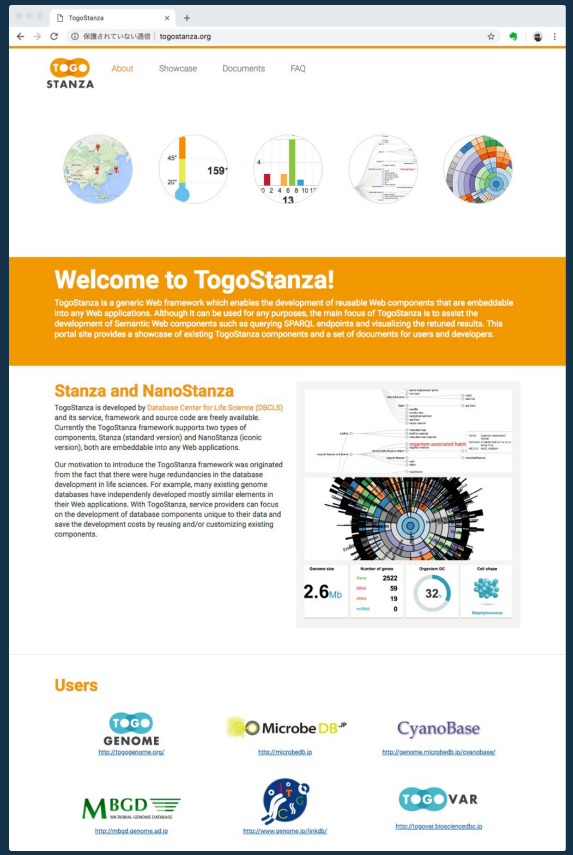
Goals of the hacking project

TogoStanza (TogoGenome)

- **Reusable Web Components for Genome DB**
  - http://togostanza.org/
  - (e.g. http://togogenome.org/gene/9606:APOE)
  - SPARQL back-ended visualization modules (cf BioJS)
  - Already used in TogoGenome, TogoVar, MicrobeDB.jp etc.

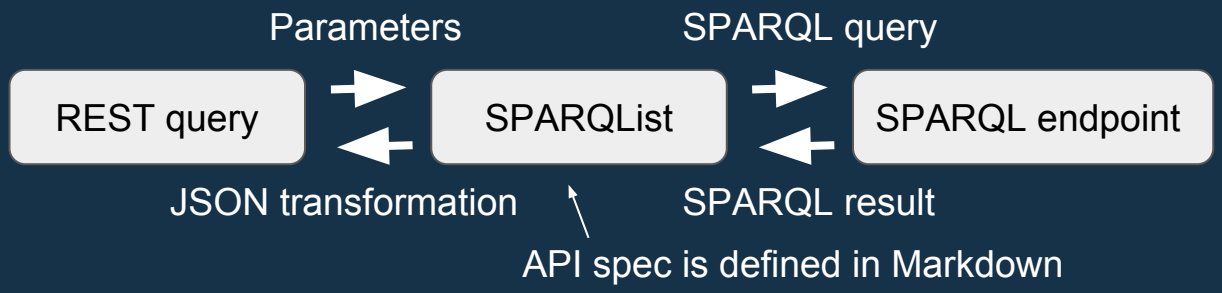
- **General goal of the hacking project**
  - Expose embedded SPARQL queries as REST APIs
  - Potential collaboration with BioJS?

- **Expected results** at the end of the hackathon
  - Data retrieval from REST API through SPARQList
  - Better design ideas for the future

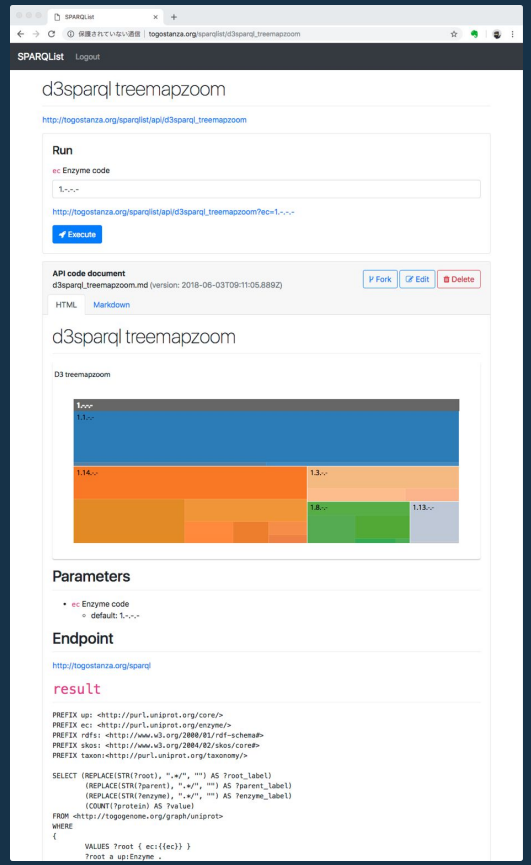


SPARQList

- **REST API repository for SPARQL**
  - http://togostanza.org/sparqlist/
  - https://github.com/dbcls/sparqlist
  - API definition can be written in Markdown
  - Execute SPARQL on-the-fly
  - Transform resulting JSON with JavaScript



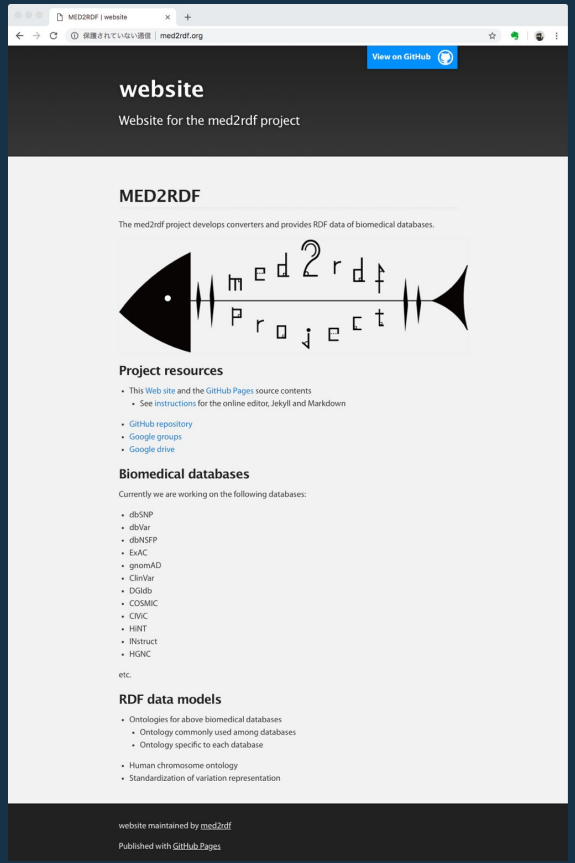
- **General goal of the hacking project**
  - Develop REST APIs for SPARQL embedded in TogoStanzas
- **Expected results** at the end of the hackathon
  - Data retrieval from REST API through SPARQList



Med2RDF

- **Reusable Biomedical RDF datasets**
  - http://med2rdf.org/
  - Provides RDF version of biomedical datasets for
    - ClinVar, dbSNP, dbVar, ExAC, gnomAD
    - ICGC, COSMIC, CIViC, HiNT, INstruct
    - Ontologies for Human Chromosome, Variation model
  - In collaboration with Kyoto University under the AMED project

- **General goal of the hacking project**
  - Develop Ontologies & TogoStanza for Med2RDF data
  - Pull more attention from international collaborators
- **Expected results** at the end of the hackathon
  - Completion and deployment of HCO and IDO at Id.org

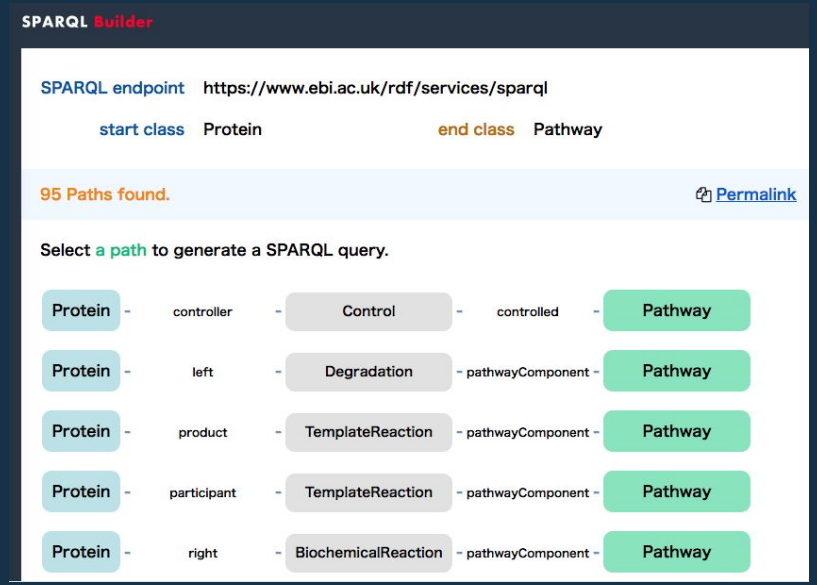


SPARQ Builder / LOD Surfer

- **A search system based on class-class relationships**
  - Using metadata extracted from SPARQL endpoint, SPARQL Builder supports in writing SPARQL.
  - Based on the metadata for SPARQL Builder, LOD Surfer can extract data from multiple endpoints.

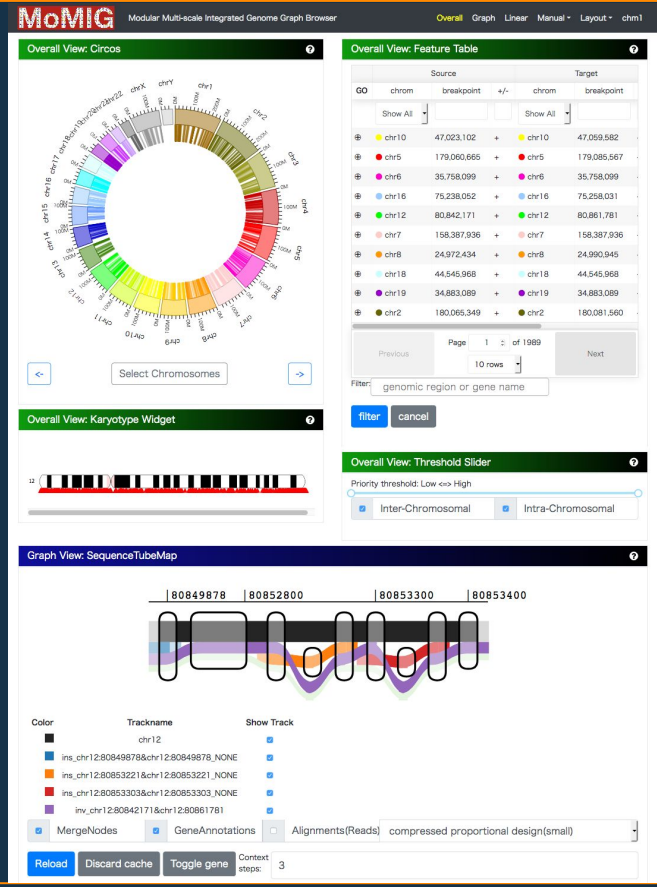
- **General goal of the hacking project**
  - Develop an efficient federated search system using the metadata and class-class relationships.
  - Find an application to these systems.

- **Expected results** at the end of the hackathon
  - Include more SPARQL endpoints especially for large datasets.



Graph-based Japanese Reference Genome

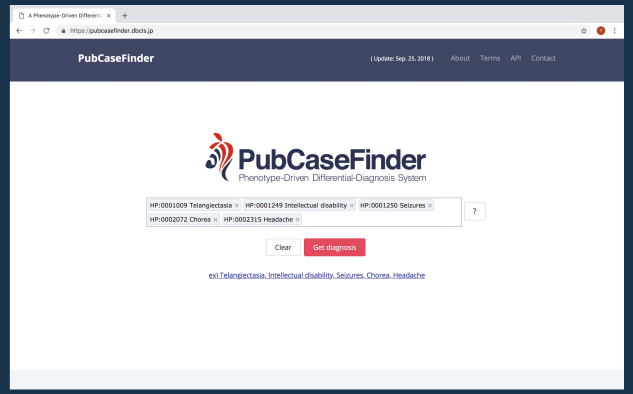
- **Graph-based Japanese Reference Genome**
  - Improves mapping analysis of Japanese genome
    - Including SNPs and SVs observed in Japanese as branching of mathematical graph
  - Cooperates with related projects
    - Genome graph workshops: http://genomegraph.jp
    - MoMIG, a graph genome browser (right figure)
      - http://demo.momig.tokyo/
      - https://github.com/MoMI-G/MoMI-G/



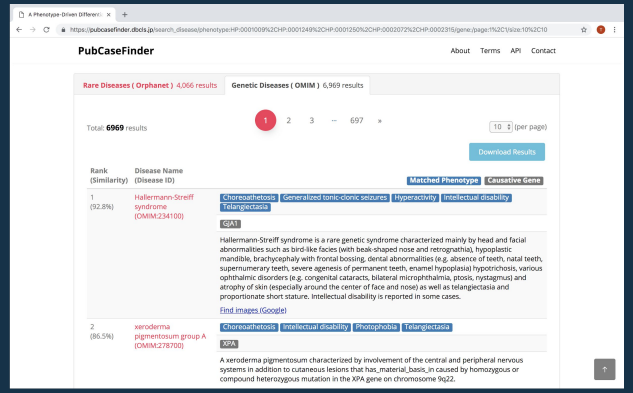
- **General goal of the hacking project**
  - Publish graph-based Japanese reference genome
- **Expected results** at the end of the hackathon
  - Evaluation of graph-based Japanese reference genome

PubCaseFinder

- **A phenotype-driven differential-diagnosis system**
  - https://pubcasefinder.dbcls.jp/
  - Helps clinicians make a diagnosis for rare diseases and genetic diseases defined in Orphanet and OMIM



- **General goal of the hacking project**
  - Develop a REST API for providing a ranked list of genetic diseases based on phenotypic similarity



- **Expected results** at the end of the hackathon
  - Better design ideas for the future

Other tools we developed (to make use of RDF data)

Semantic Web tools developed in DBCLS

- NBDC RDF Portal: RDF data repository with summaries and SPARQL endopoints
- TogoWS: search, retrieval, parse, and convert entries into RDF
- TogoDB: deploy your datasets as a Web database and generate RDF
- D3SPARQL.js: D3.js based JS library to visualize SPARQL results
- SPARQL-proxy: safely deploy a SPARQL endpoint with cache and job management
- SPANG: easy-to-use command-line SPARQL client
- D2RQ Mapper: configure RDB as a SPARQL endpoint w/ intuitive GUI
- Umaka-Yummy: monitor and evaluate the quality of SPARQL endpoints

  :
- Also developing tools for NGS, Web apps, Text-mining, QA system, ML & AI

Hack organisation

Organisation of the hacking project

- TogoGenome/TogoStanza & SPARQList
  - Refactoring to extract embedded SPARQL queries as public REST APIs
  - Explore collaboration with BioJS
    - Toshiaki Katayama, Shuichi Kawashima

- Human Chromosome Ontology & Identifiers.org Ontology
  - Provide canonical URIs for human chromosomes
  - Provide semantics to the Identifiers.org databases
  - Update DBCLS RDFizing database guidelines according to the results
    - https://github.com/med2rdf/hco
    - https://github.com/ktym/idorg-ontology
    - https://github.com/dbcls/rdfizing-db-guidelines
    - Toshiaki Katayama, Shuichi Kawashima

- Japanese reference genome graph
  - Develop initial version of Japanese genome graph
    - Toshiyuki Yokoyama, Toshiaki Katayama

Organisation of the hacking project

- SPARQL Builder / LOD Surfer
  - Include more SPARQL endpoints especially for large datasets.
    - https://github.com/sparqlbuilder
    - https://github.com/LODSurfer
    - Atsuko Yamaguchi

- PubCaseFinder
  - Develop a REST API for providing a ranked list of genetic diseases defined in OMIM
    - https://pubcasefinder.dbcls.jp/mme
    - Toyofumi Fujiwara

- SPANG
  - Facilitate database integration through SPARQL
    - https://github.com/dbcls/spang
    - Hirokazu Chiba

Organisation of the hacking project

- Public data & workflows
  - Get data from public repo, then deploy CWL workflows on clouds and run
    - https://github.com/pitagora-galaxy/cwl
    - Manabu Ishii, Ryota Yamanaka, Tazro Ohta

Post-biohackathon perspectives

- More interoperable data
- More data science
- More applications
- More international collaborations
- Continued in the NBDC/DBCLS BioHackathon in Matsue this December :)