

# FAIRtracks and Omnipy

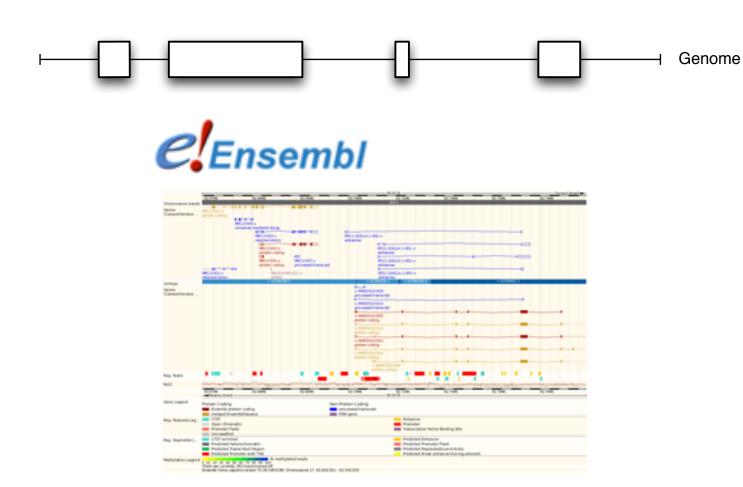
- FAIRtracks interoperability story



March 7, 2023 Sveinung Gundersen

www.elixir-europe.org

### Genomic tracks



Any genomic data file mapped to a reference genome coordinate system!

- Not only for visual analysis!

- Gene regions, repeating elements, conserved regions
- Chromatin accessibility (e.g., DNase I Hypersensitivity)
- Binding of Transcription Factors to DNA
- Histone modifications along DNA
- Gene expression, Gene fusion, Transcription start sites (TSS)
- Cis-regulatory elements (promoters, enhancers...)
- DNA methylation
- 3D genome structure
- GWAS SNPs for disease, SNVs and CNVs in cancer



# Important goals for FAIRtracks

**F**indable

**A**ccessible

Interoperable

Reusable



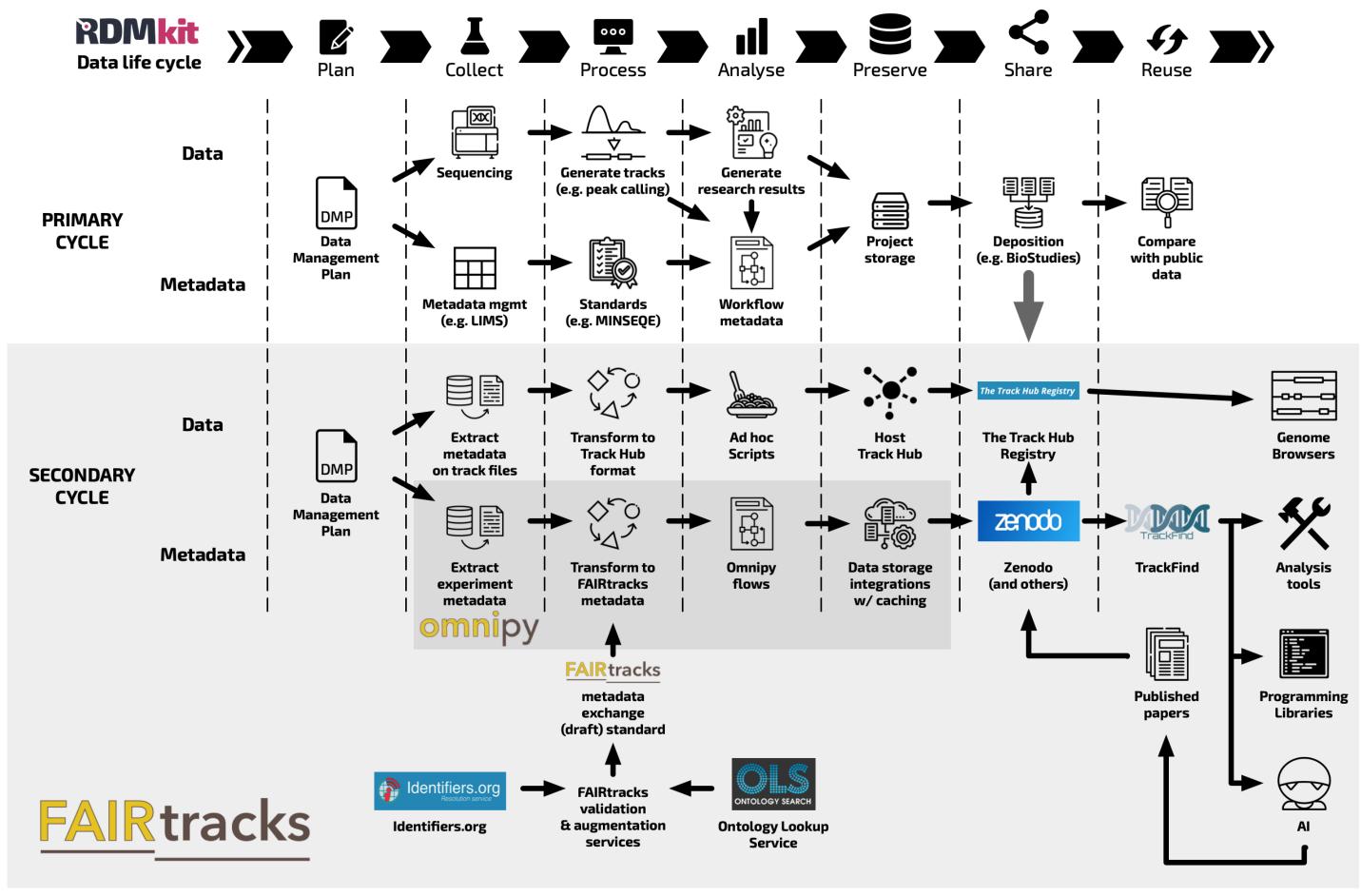
- Global identifiers for track files, as well as track collections, studies, samples, and experiments
- Search and import of individual track files across repositories, also repositories not supported by consortia data portals
- Search using formal (non-free-text) queries
- Easy (automated) retrieval of track data
- Persistence of track data and versioned metadata
- Lack of standard metadata model with practically useful attributes
- Annotation using community-accepted vocabularies/ontologies
- Cross-references to records in relevant (meta)data repositories
- Support for detailed context-specific metadata content together with standardised summary attributes
- Simple process for data providers to submit data and metadata that at the same time accommodates the required stringency for (automated) downstream usability
- Easily available data usage licenses
- Detailed provenance of experimental and in silico analysis steps



# FAIRtracks (tool assembly)

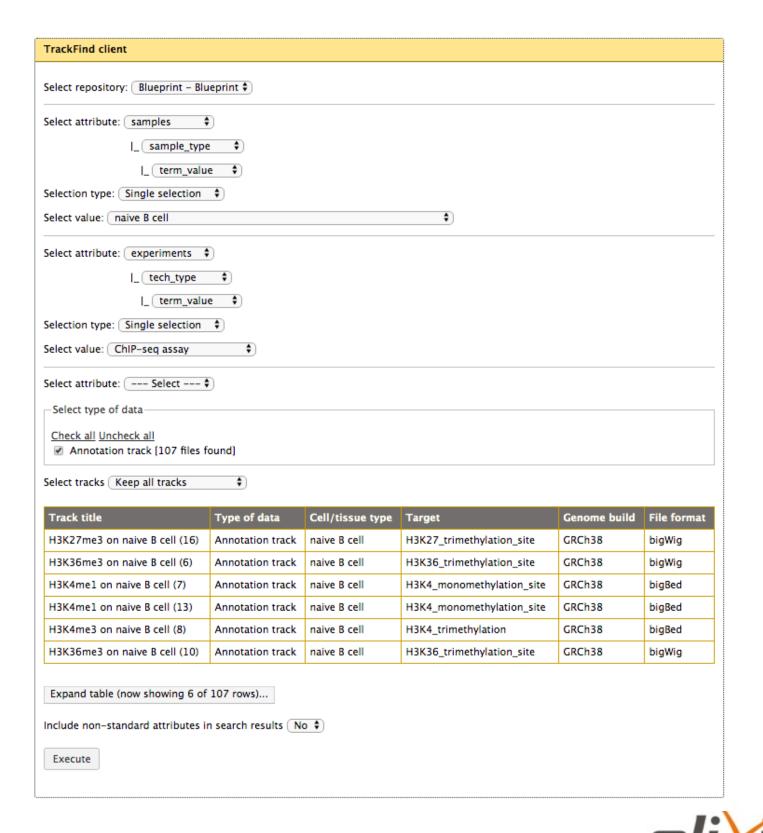
- Represents a secondary iteration of the data life cycle (as defined in RDMkit)
- FAIRtracks

   (more-or-less)
   begins when
   the primary
   data life cycle
   ends



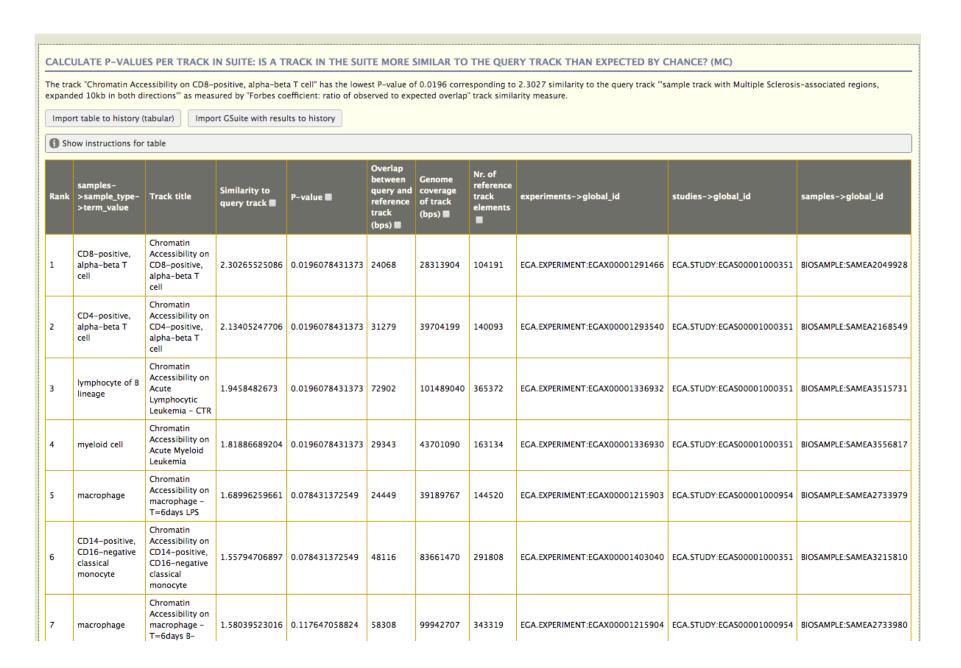
# Tool integration

- TrackFind client implemented in GSuite HyperBrowser:
  - https://hyperbrowser.uio.no/trackfind test (search for tool "trackfind")
- JSON and GSuite (<a href="http://gtrack.no">http://gtrack.no</a>)
   formats as metadata / search result exchange format
- Search results can be transferred to the HyperBrowser server, preprocessed, and used in statistical analyses
- Integration into vanilla Galaxy under development



# Tool integration

- Analysis example
  - Sites of open chromatin for various cell types from BLUEPRINT (DNasel HS)
  - Set of variants associated with Multiple Sclerosis (MS)
  - Which cell types are most relevant to MS? (i.e., where do the variants overlap the most with open chromatin?)





# Identifiers for genomic tracks?

- Should there be globally unique, persistent identifiers for genomic track files (e.g. BigBED/BigWIG, VCF, etc)
- No such thing exists, but we highly recommend that they should be created and indexed

- Also, identifiers for track collections would be very useful
  - Would easily FAIRify "Mix-and-match" track file collections analysed in particular research papers, if the track files are already FAIRified



### Updates

- **ELIXIR** Recommended Interoperability Resource 2021
- Manuscript published Apr 2021
- Invited blog post in F1000Research published Dec 2021
- New web site published 2022: https://fairtracks.net



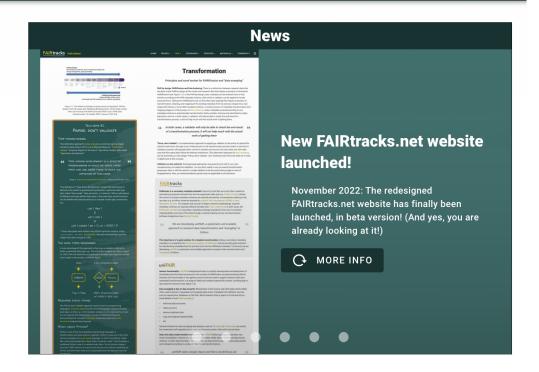


STANDARDS > SERVICES >



- Hoards of genomic track data at your fingertips

In the spirit of Open Science, the FAIRtracks ecosystem provides technical solutions for the abundance of genome browser track files ("genomic tracks") to become "Findable, Accessible, Interoperable, and Reusable (FAIR)" in new research contexts.



#### **COMMUNITY BUILDING**

#### **Bridging the data gaps**

We aim to connect:

- data providers
- biocurators
- tool developers
- the FAIR community
- researchers/data analysts
- ...and other interested parties

Together we can mobilize the power of genomic tracks!

#### **TECHNOLOGY**

#### **Quality metadata and services**

Working in concert with the FAIRtracks draft standard for metadata of genomic tracks, we have built an ecosystem of services to interface with track metadata, including:

- Metadata augmentation
- Metadata validation
- Metadata transformation
- Precision search

You can connect to these core services both upstream (for data providers/biocurators) and downstream (for tool developers/analytical end users).

#### ORGANIZATIONAL BACKING

#### **Endorsed by ELIXIR**

The FAIRtracks ecosystem is developed and provided as part of the national Service Delivery Plans by ELIXIR Norway and ELIXIR Spain, and is supported by the Track Hub Registry group at EMBL-EBI.

FAIRtracks is endorsed by ELIXIR Europe as a Recommended Interoperability Resource (RIR).

But we need more partners:

Please help us bring the wealth of available track data and metadata to the fingertips of researchers and bioinformaticians everywhere!



### Projects / collaborations

- Galaxy implementation study (2021-2023)
  - Task 1.4. Enhanced metadata functionality in Galaxy to support querying and importing of FAIRtracks-annotated genomic track data
    - Implement <u>TrackFind</u> client as a Galaxy tool
    - Idea: associate FAIRtracks metadata with data as a Galaxy dataset/collection, keeping this relation through downstream analysis steps
    - Investigate RO-Crate integration
- RDA TIGER (HORIZON-INFRA-2022-EOSC-01-04)
  - RDA/EOSC project started Jan 2023
  - Demonstrator Working Group for RDA TIGER (pre-approved, but still need to apply)
    - Goal is to integrate metadata from (at least) Functional Annotation of ANimal Genomes (FAANG) into FAIRtracks, and also expand tool interoperability
    - Some funding available
    - More importantly: door-opener into both RDA and EOSC



### Literary digression: omnify

So that the Church of England, in these manners of dispensing the power of the keys, does cut off all disputings and impertinent wranglings, whether the priest's power were judicial or declarative; for possibly it is both, and it is optative too, and something else yet; for it is an emanation from all the parts of his ministry, and he never absolves, but he preaches or prays, or administers a sacrament; for this power of remission is a transcendent, passing through all the parts of the priestly offices. For the keys of the kingdom of heaven are the promises and the threatenings of the Scripture, and the prayers of the Church, and the Word, and the Sacraments, and all these are to be dispensed by the priest, and these keys are committed to his ministry, and by the operation of them all he opens and shuts heaven's gates ministerially.

No more ingenious way of making nothing of a thing than by making it every thing. Omnify the disputed point into a transcendent, and you may defy the opponent to lay hold of it. He might as well attempt to grasp an aura electrica.

 One of the most obscure words in the English language:

# omnify transitive verb

om·ni·fy 'ämnəˌfī

-ed/-ing/-es

: to make universal: ENLARGE

"Notes on Jeremy Taylor", The Literary Remains Of Samuel Taylor Coleridge, by Samuel Taylor Coleridge Publication date: 1836-1839



#### Programming environment PyCharm VSCode Jupyter Notebook Command line **Omnipy** Generic tasks & flows Task and flow specification (in Python) Flattening **Abstraction layer** Mapping TaskTemplate Ontology LinearFlowTemplate Cleaning DagFlowTemplate Validation FuncFlowTemplate Workflow & (Extract, Transform, Lower-level data manipulation **Data repositories** Load) ETL systems Pandas **FAIR** services ENCODE Nextflow R (standard library) TCGA Snakemake Frictionless (Meta)data schemas ENA CWL MS Excel / Google Sheets Óntologies GitHub Galaxy Python standard library Validation **REST APIs** Airflow SQL Prefect (note: draft image! 😄) Compute/storage infrastructures Kubernetes Cloud compute (Amazon WS, Azure, Google Cloud) Cloud storage (S3) HPC

Local compute & storage



#### Data portals

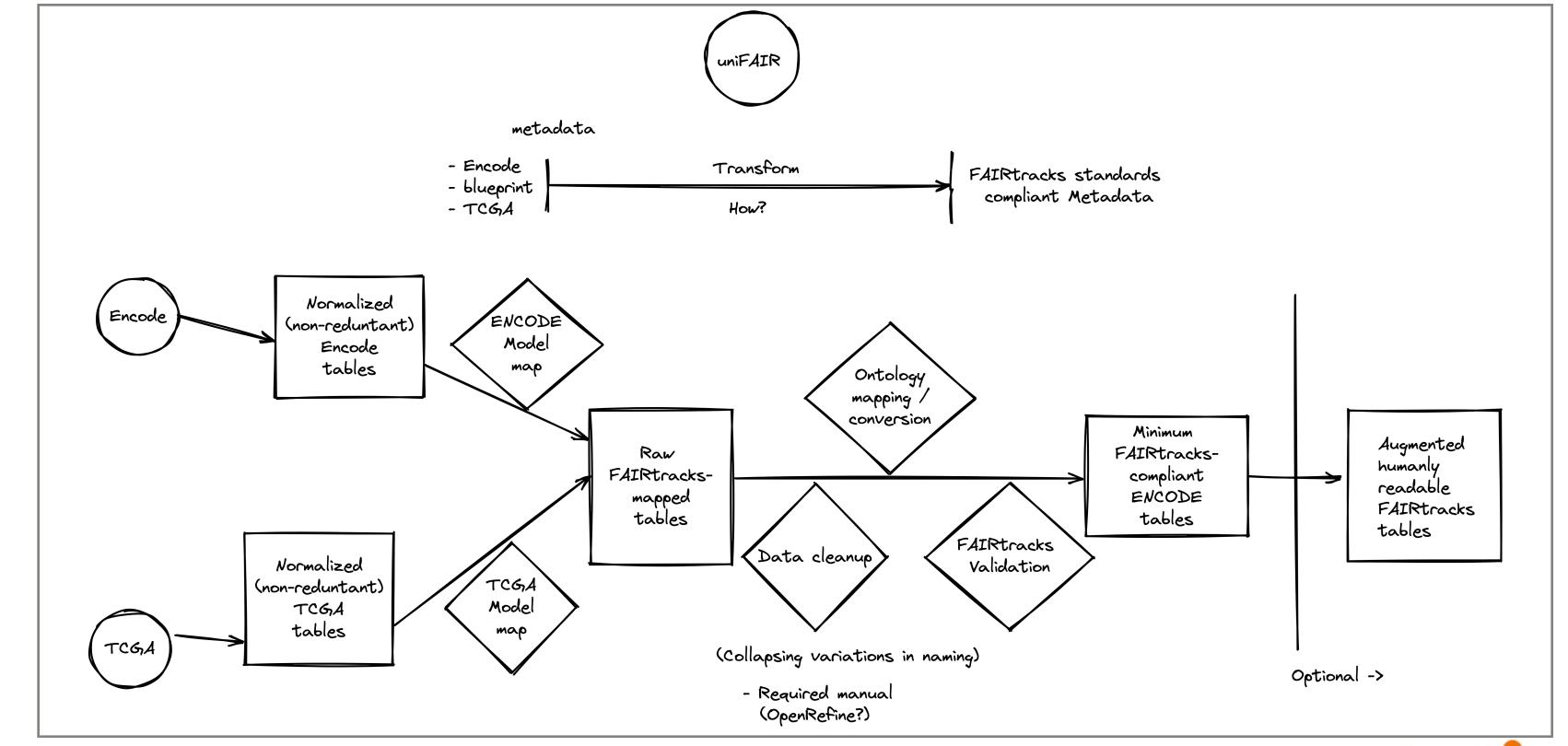
Custom metadata schemas:

ENCODE FAANG TCGA ICSC IHEC











#### Data portals

Custom metadata schemas:

ENCODE FAANG TCGA ICSC IHEC metadata

- Encode
- blueprint
- TCGA

Normalized
(non-reduntant)
Encode
tables

Normalized
(non-reduntant)
FAIRtracks
map
conversion

FAIRtracks

rapped
tables

Normalized
(non-reduntant)
FAIRtracks
map
conversion

FAIRtracks

rapped
tables

(collapsing variations in naming)

Optional ->

Optional ->

 Required manual (OpenRefine?) Metadata exchange standard

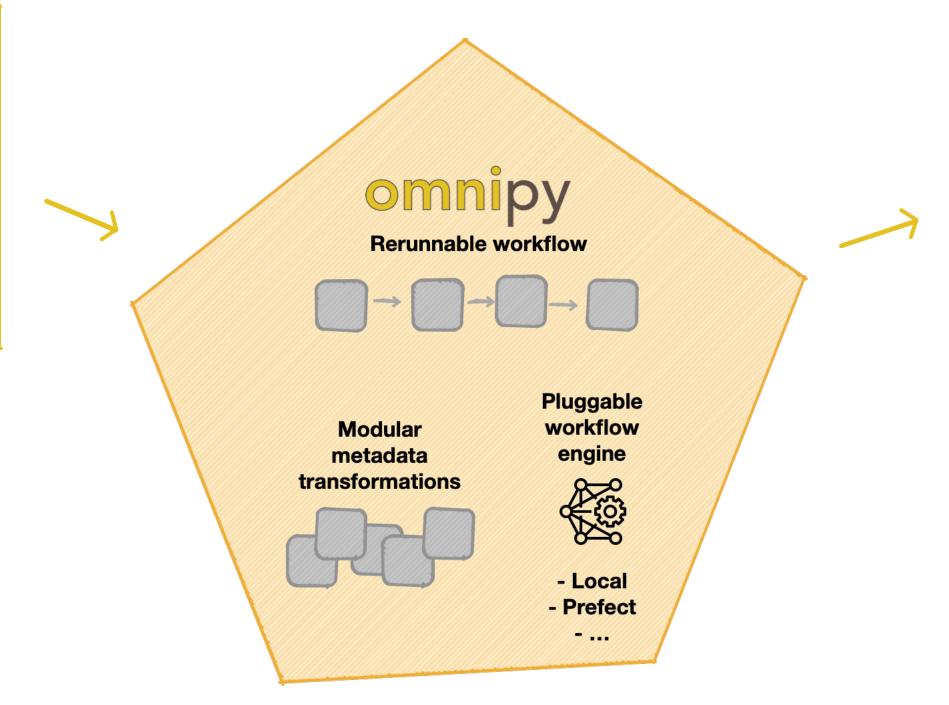
**FAIR** tracks



#### Data portals

Custom metadata schemas:

ENCODE FAANG TCGA ICSC IHEC



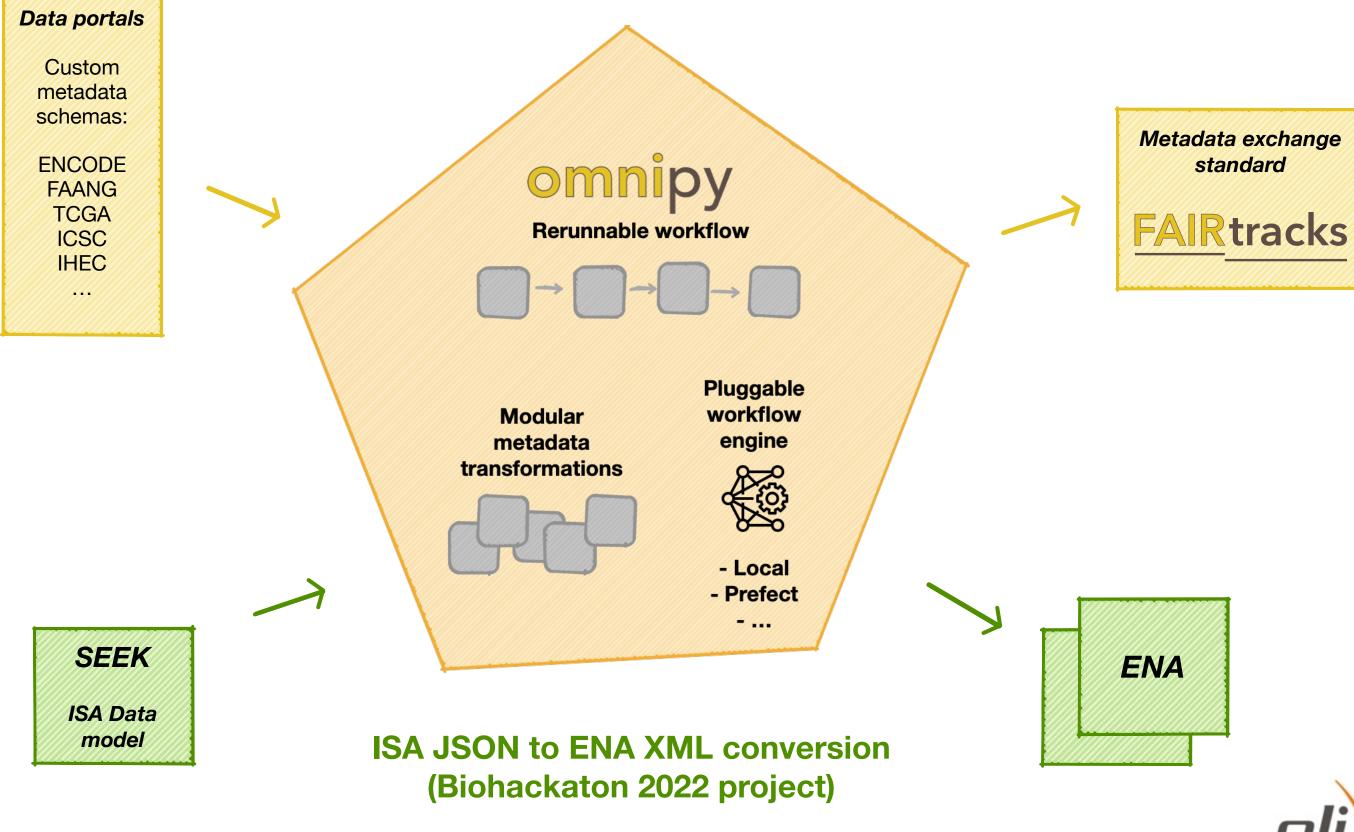
Metadata exchange standard

**FAIR** tracks

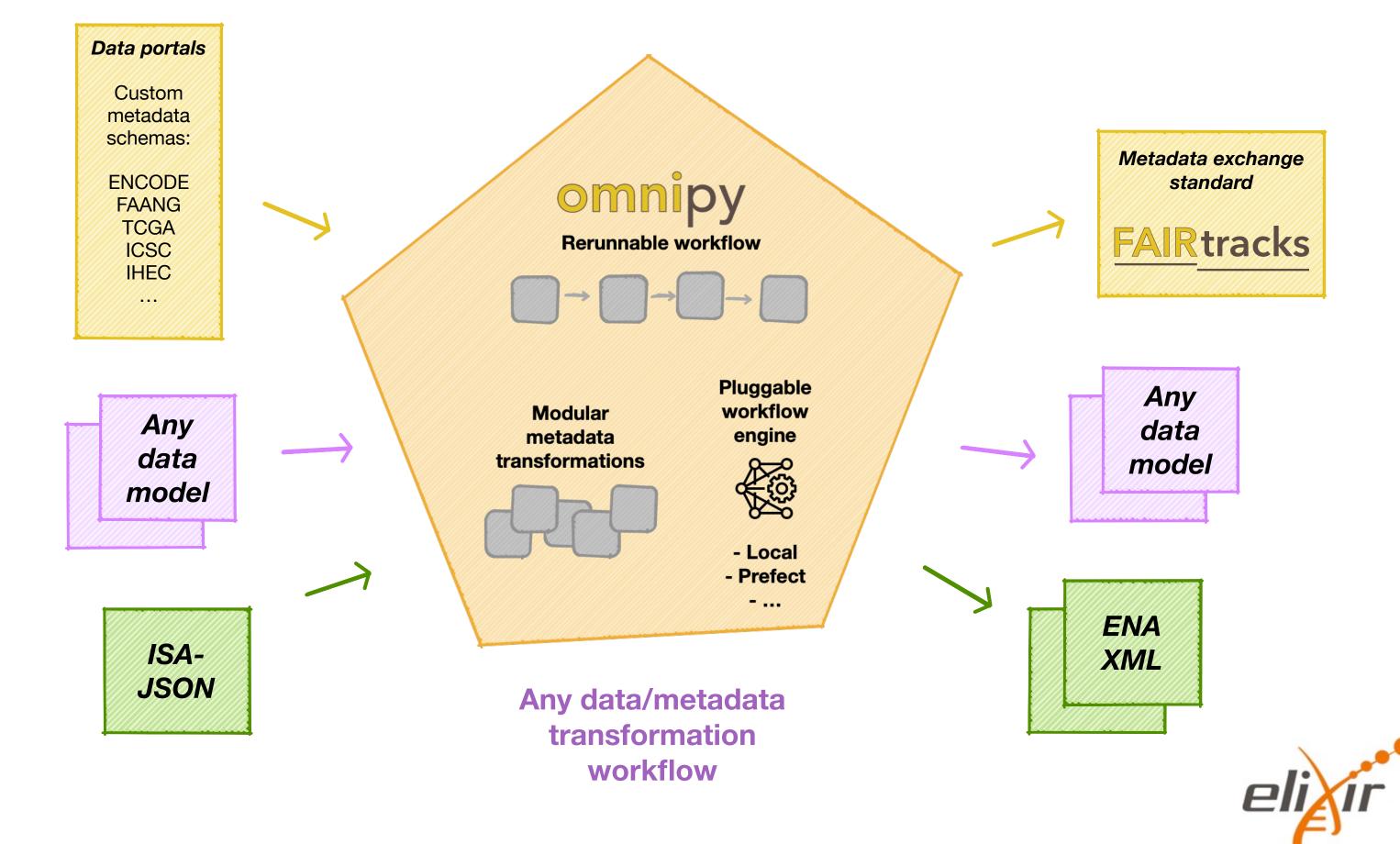
**FAIR** tracks

metadata ETL workflows

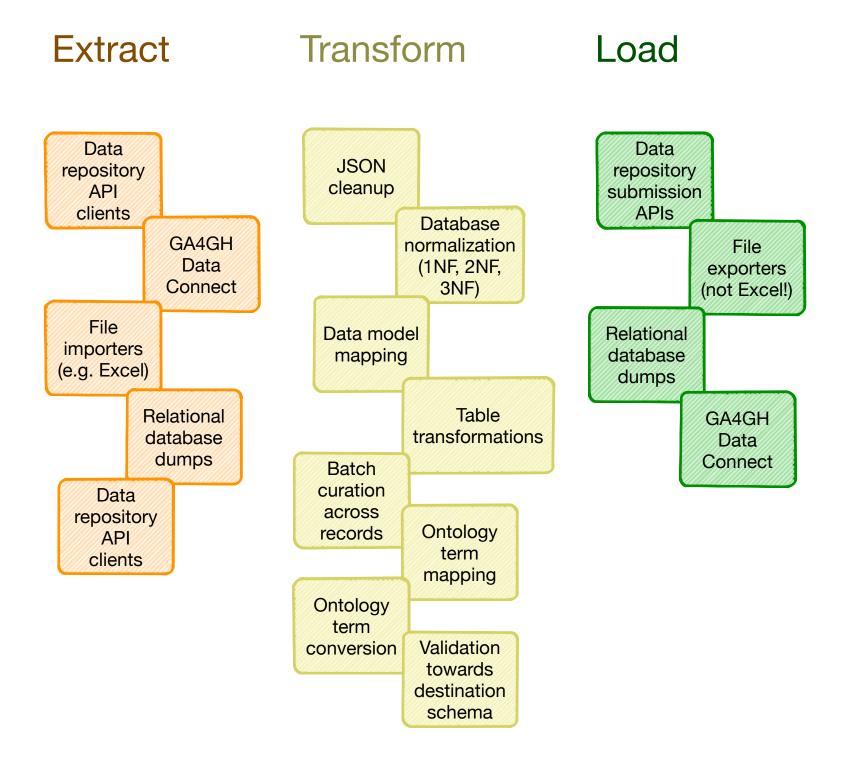








### Reusable metadata ETL modules



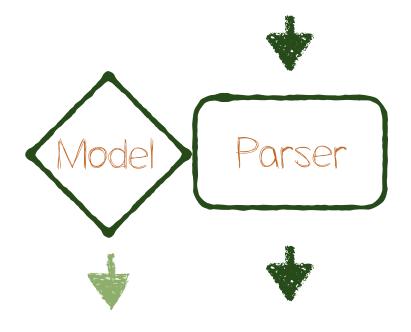


### Parse, don't validate!



- Alexis King: "Parse, don't validate" (2019 blog post)
  - Blog: https://lexi-lambda.github.io/blog/2019/11/05/ parse-don-t-validate/
- Step-wise parsers munge the data to conform to schema/data model restrictions
- Allow for some variation in data input, with automatic type conversion
- Gradually improve structure of data

### Less structured data



More structured data w/ model in data type





- Python-based dynamic orchestration engine
- Fully Open Source: Apache 2.0 license
- Dynamically registered, DAG-free workflows:
  - Supports if/for/while statements
  - Dynamic branching logic depending on runtime conditions
- Code as workflows:
  - No need to specify task and workflow parameters outside of normal code
  - Debug locally, run remotely using the same source code
  - Use tools you might already know, including:
    - Pydantic, FastAPI, RRule, Sqlite, asyncio, Dask, ...
- Rules engine:
  - State-based orchestration engine, with API separation
  - GUI Dashboard to administrer workflow runs
- A bunch of pre-built integrations:
  - data sources/destinations (GitHub, PostgreSQL, Jupyter notebooks, etc.)
  - deployment options (Docker, Kubernetes, cloud storage, etc.)

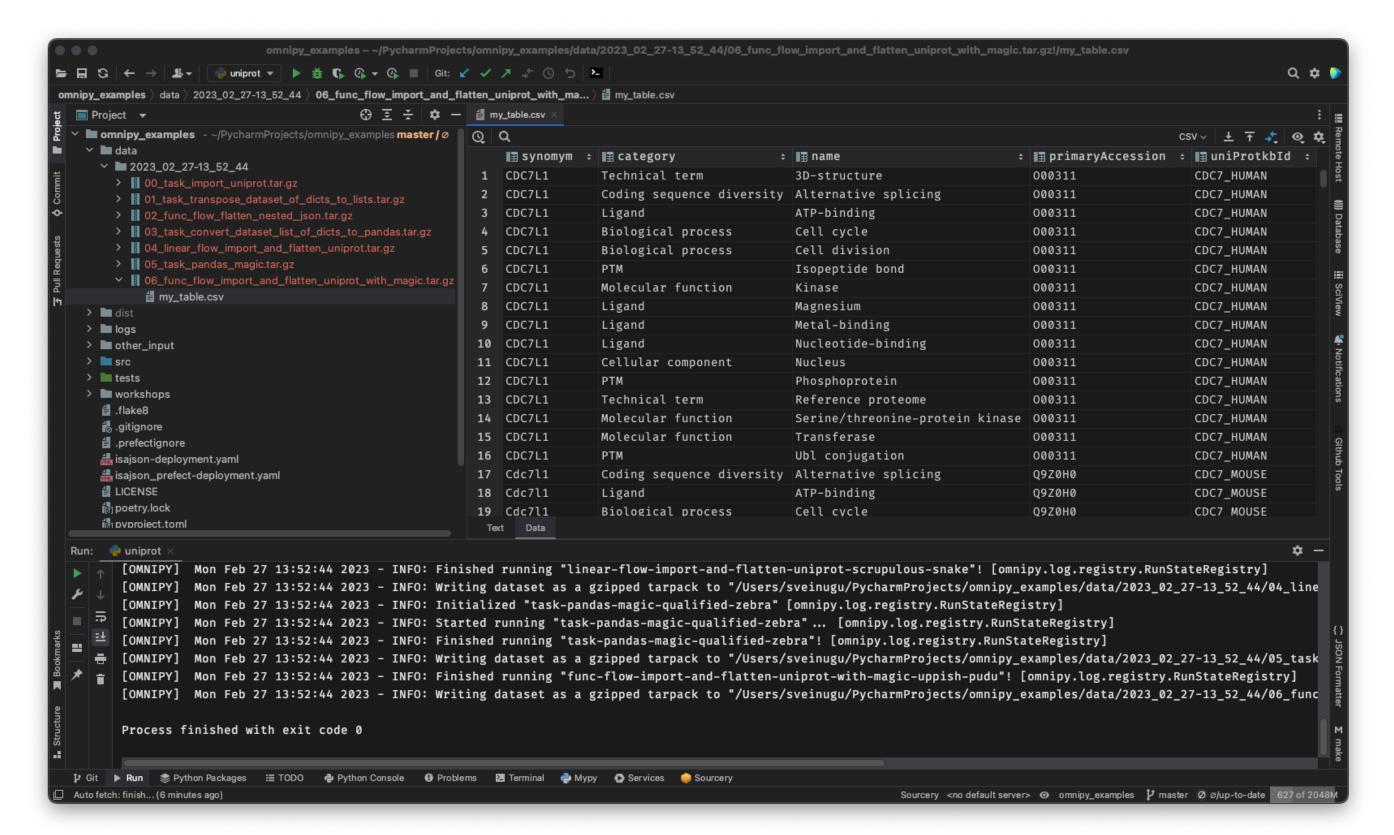
#### Simple Prefect workflow with two tasks

(from https://orion-docs.prefect.io/tutorials/first-steps/#run-a-basic-flow-with-tasks)

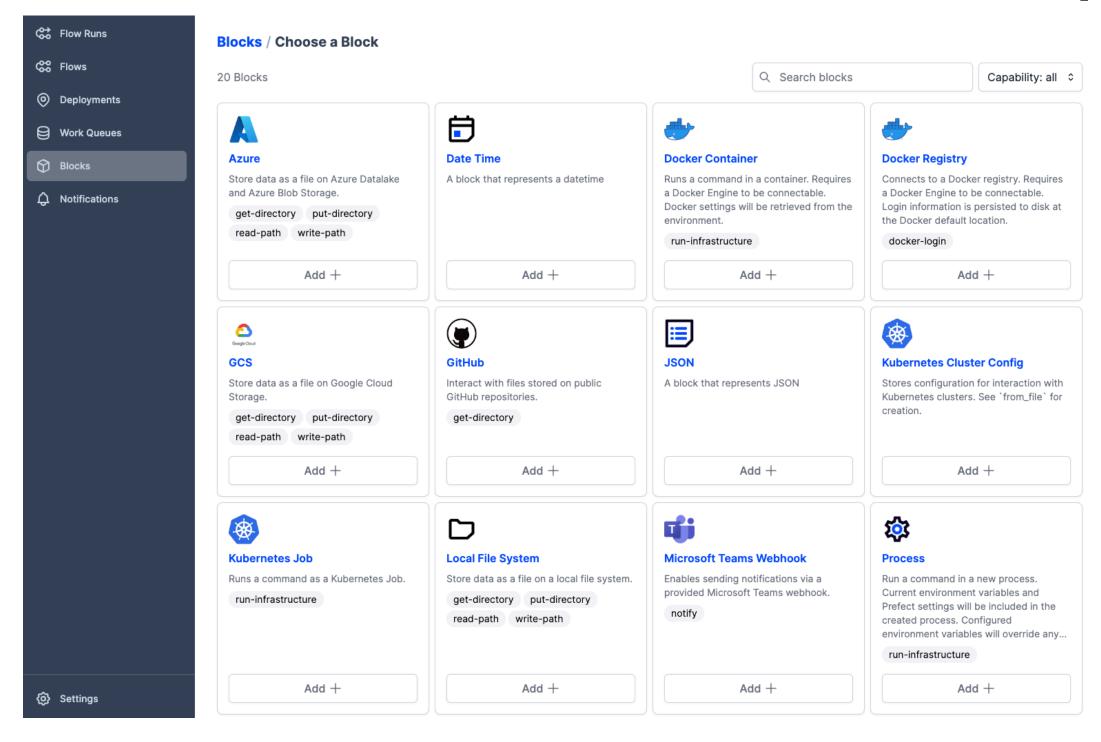
```
import requests
from prefect import flow, task
@task
def call_api(url):
    response = requests.get(url)
    print(response.status_code)
    return response.json()
@task
def parse_fact(response):
    print(response["fact"])
    return
@flow
def api_flow(url):
    fact_json = call_api(url)
    parse_fact(fact_json)
    return
```



# Develop, inspect and deploy directly from IDE

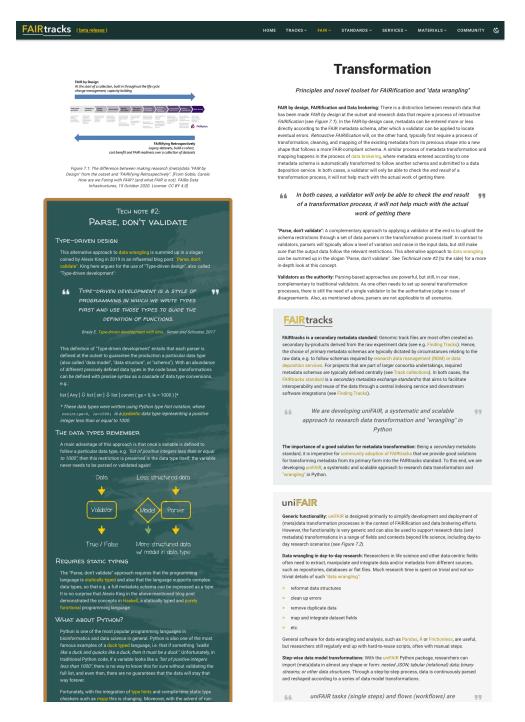


### Prefect orchestration GUI for local and remote deployment





### More information on Omnipy



- Check out the newly launched redesign of the FAIRtracks website:
  - https://fairtracks.net
- Specifically:
  - https://fairtracks.net/fair/#fair-07-transformation
- GitHub repo:
  - https://github.com/fairtracks/omnipy



### Collaboration

We have limited development resources and are aiming for FAIRtracks to become a community endeavor!

We are planning a BioHackathon project on expanding to Omnipy and are looking for collaborators, e.g. to integrate other resources or standards into Omnipy or to use Omnipy to set up metadata transformation.

We are interested in any types of contributions & collaborations!

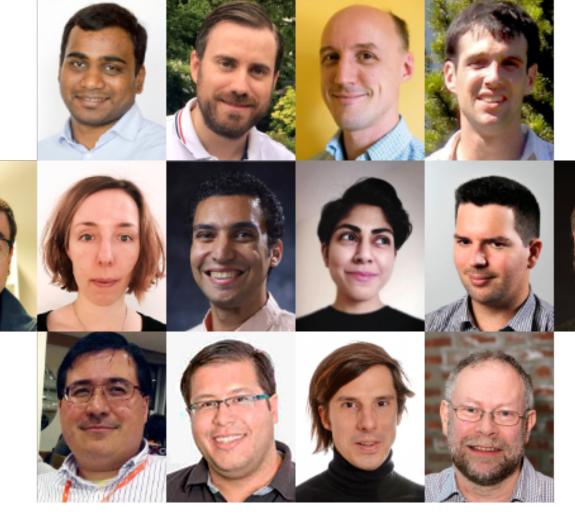
Please contact us at: fairtracks@elixir.no



# Acknowledgements

# FAIRtracks

### S. Gundersen and the FAIRtracks team:



**EMBL-EBI, Hinxton, UK** 

**ELIXIR Norway, Centre for Bioinformatics, UiO** 

+ Jeanne Cheneby, Pável Vázquez

**ELIXIR Spain, Barcelona Supercomputing Centre** 

**ELIXIR Norway, NTNU & UiB** 

Acknowledgements

WP3: Central track search service

WP4: Tool side implementation and testing

WP5: To













