maxo
MEDICAL ACTION ONTOLOGY

monarch
INITIATIVE

Expansion of Medical Action Ontology (MAxO) for COVID-19

2020 October

*Development/Curation

Leigh Carmody (JAX)
Nico Matentzoglu (EBI)
Lauren Chan (OSU)
Nicole Vasilevsky (OHSU)

*Annotation/Text-mining software tools

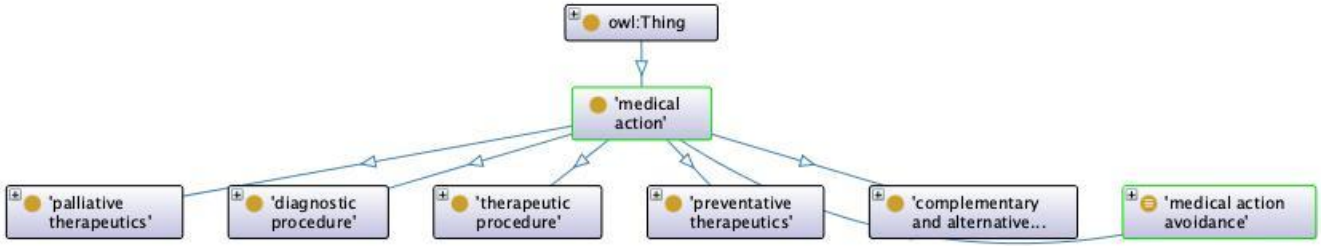
Hannah Blau (JAX)
Michael Gargano (JAX)

Peter Robinson (JAX)

The Jackson Laboratory

Medical Action Ontology (MAxO) overview

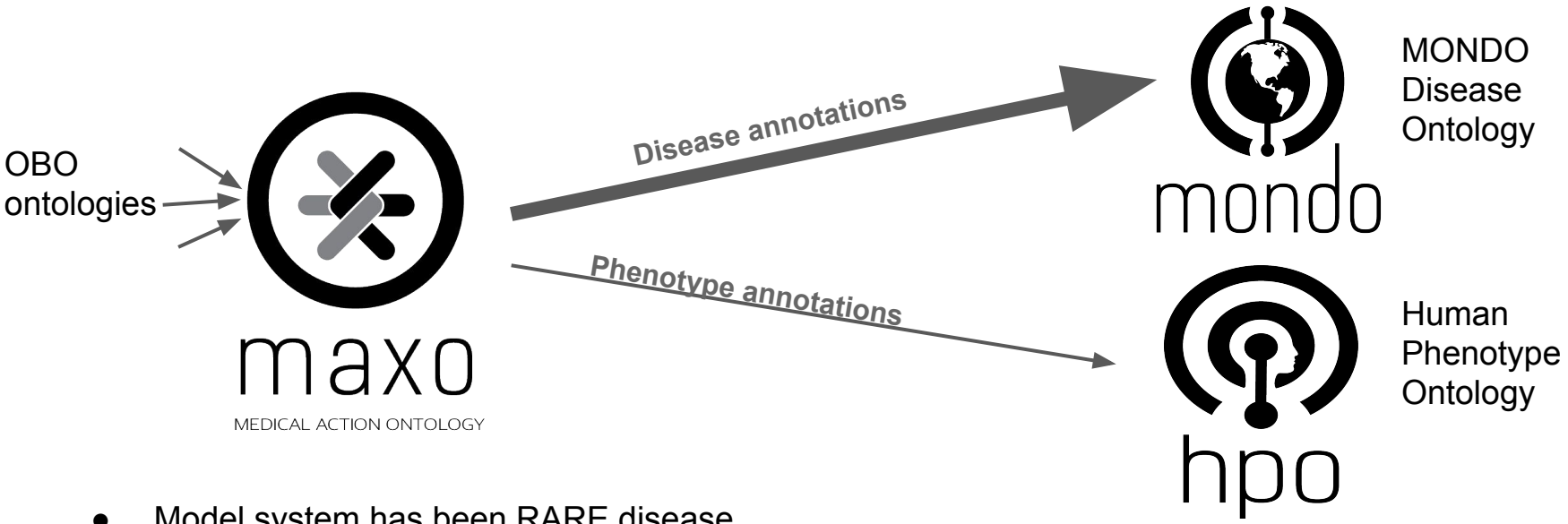
The Medical Action Ontology (MAxO) provides a structured vocabulary for medical procedures, interventions, therapies, and treatments.



- 980 MAxO terms
- 3261 synonyms
  - 3156 Exact synonyms
  - 28 Broad synonyms
  - 84 Narrow synonyms

Medical Action Ontology (MAxO) Annotations

MAxO was developed to annotate diseases.



- Model system has been RARE disease.

USE case: Text-mining GeneReviews

- Goal: Enrich MAxO by identifying new terms and extracting MAxO-disease links

- Has easy to identify 'management' sections
- Identify new MAxO terms, synonyms
- Build a better text-mining tool

Fanconi Anemia

Synonym: Fanconi Pancytopenia

Parinda A Mehta, MD and Jakub Tolar, MD, PhD.

▸ Author Information

Initial Posting: February 14, 2002; Last Revision: March 8, 2018.

Estimated reading time: 51 minutes

Summary

Go to:

Clinical characteristics. Fanconi anemia (FA) is characterized by physical abnormalities, bone marrow failure, and increased risk for malignancy. Physical abnormalities, present in approximately 75% of affected individuals, include one or more of the following: short stature, abnormal skin pigmentation, skeletal malformations of the upper and lower limbs, microcephaly, and ophthalmic and genitourinary tract anomalies. Progressive bone marrow failure with pancytopenia typically presents in the first decade, often initially with thrombocytopenia or leukopenia. The incidence of acute myeloid leukemia is 13% by age 50 years. Solid tumors – particularly of the head and neck, skin, gastrointestinal tract, and genitourinary tract – are more common in individuals with FA.

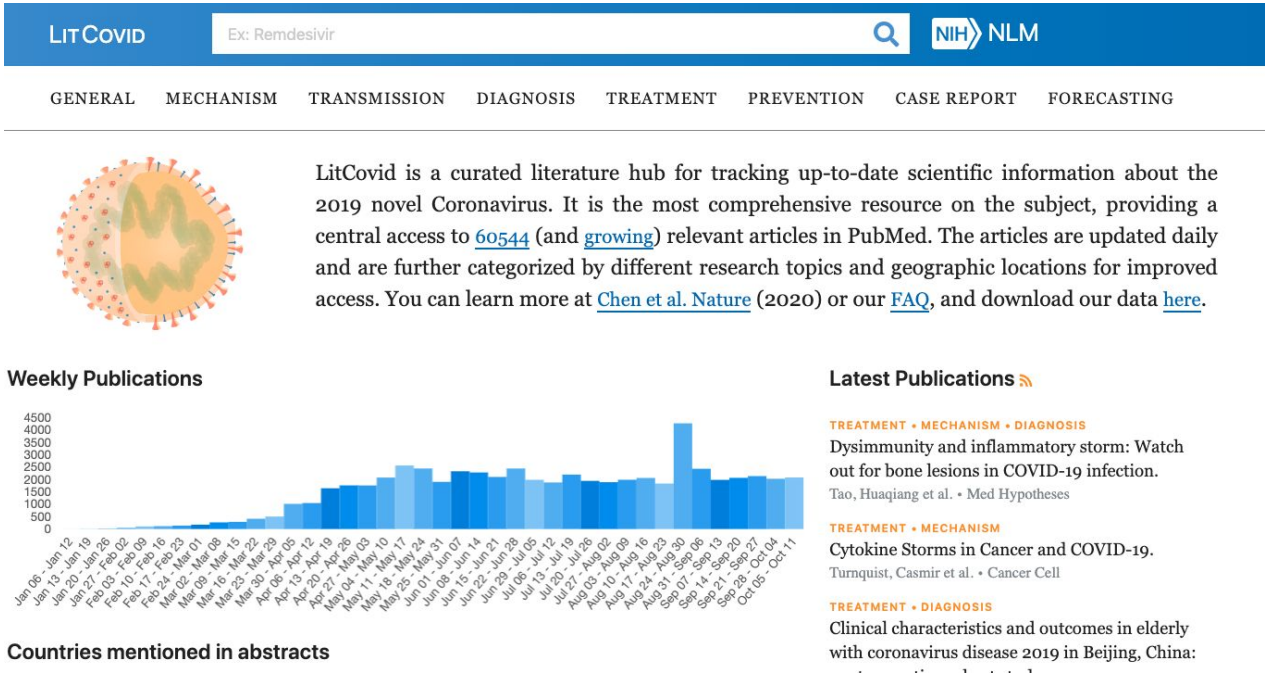
Diagnosis/testing. The diagnosis of FA is established in a proband with increased chromosome breakage and radial forms on cytogenetic testing of lymphocytes with diepoxybutane (DEB) and mitomycin C (MMC). The diagnosis is confirmed by identification of one of the following:

- Biallelic pathogenic variants in one of the 19 genes known to cause autosomal recessive FA
- A heterozygous pathogenic variant in *RAD51*, known to cause autosomal dominant FA
- A hemizygous pathogenic variant in *FANCB*, known to cause X-linked FA

Management. *Treatment of manifestations:* Administration of oral androgens (e.g., oxymetholone) improves blood counts (red cell and platelets) in approximately 50% of individuals with FA; administration of G-CSF improves the neutrophil count in some; hematopoietic stem cell transplantation (HSCT) is the only curative therapy for the hematologic manifestations of FA, but the high risk for solid tumors remains and may even be increased in those undergoing HSCT. All these treatments have potential significant toxicity. Early detection and surgical removal remains the mainstay of therapy for solid tumors.

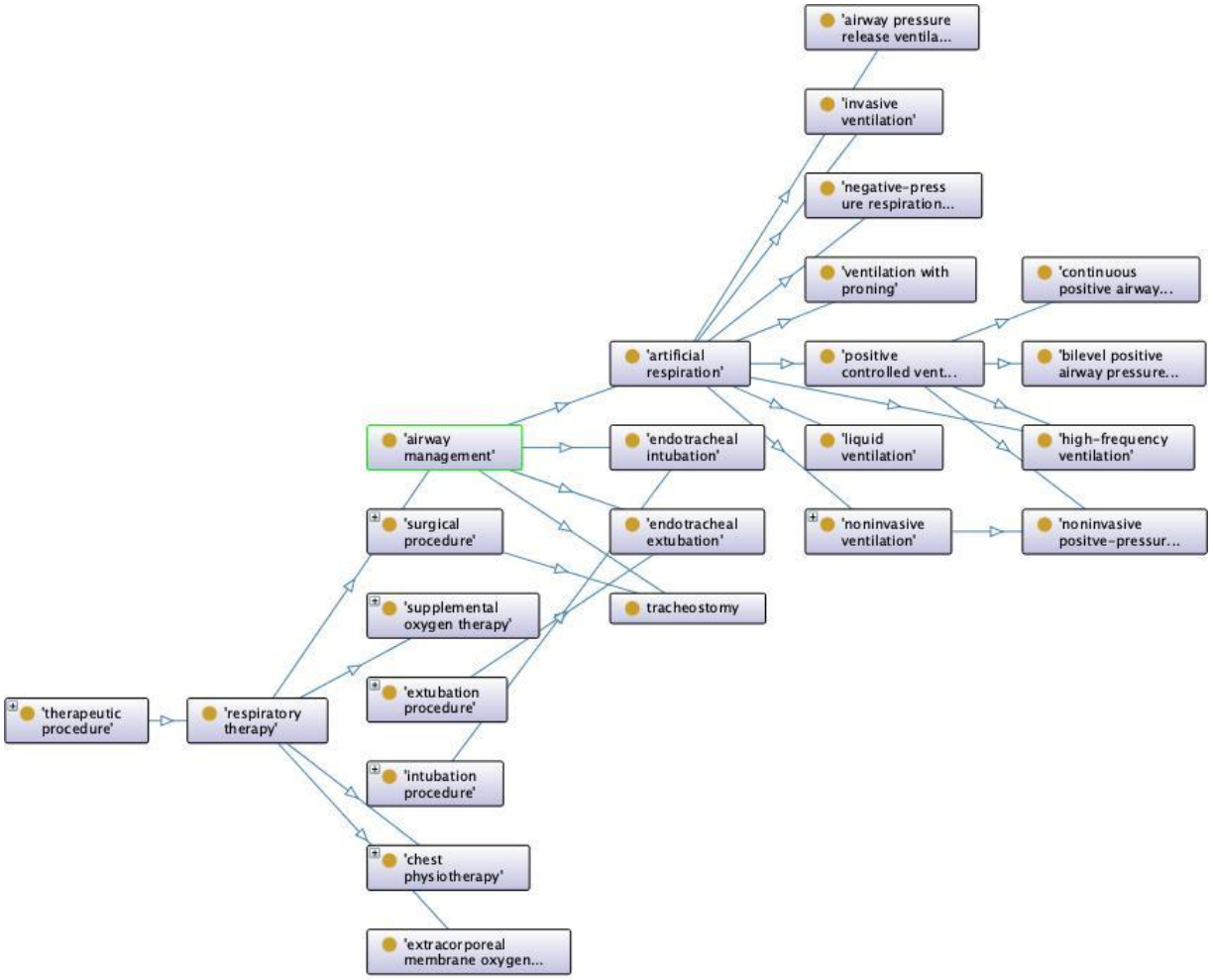
COVID-19 literature resources

CDC, WHO, and PUBMED

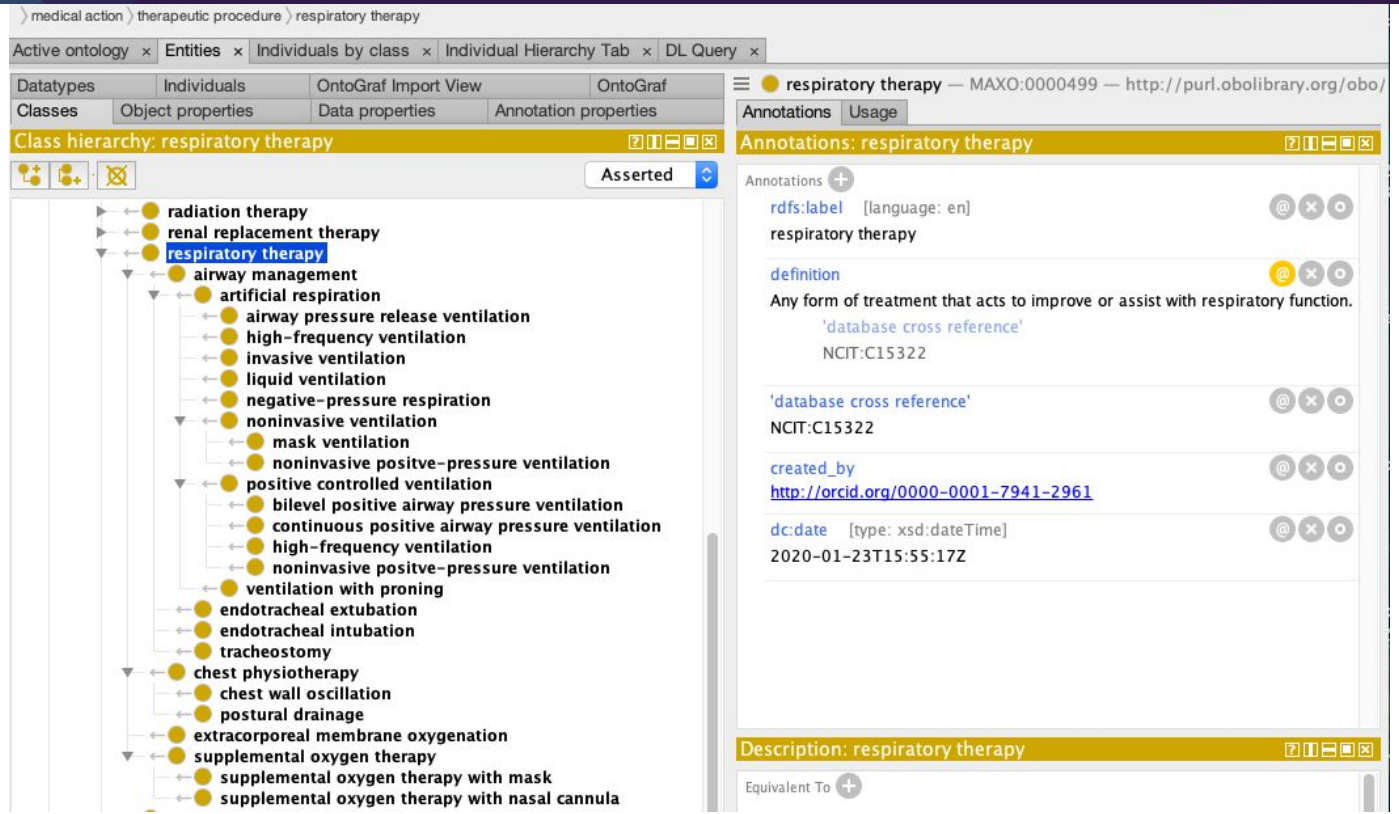


Structure of airway management terms

- Higher level terms there previously, but added many terms to include commonly conducted medical actions



Curation tools: Protege 5.5.0



Curation tools: Ontology development kit (ODK) & Dead Simple Ontology Design Patterns (DOSDP)

- Bundles tools for releases
- Allows for customizable QC procedures
- Once DOSDP patterns are established, terms are added using spreadsheets.

https://github.com/INCATools/ontology-development-kit

DOSDP pattern: swab by (UBERON) location

```
pattern_iri: http://purl.obolibrary.org/obo/maxo/patterns/swab_by_location.yaml
description: "Collection of biological material using a swab"

contributors:
classes:
  swab: MAXO:0000548
  location: owl:Thing

relations:
  has_input: RO:0002233
  part_of: BFO:0000050

annotationProperties:
  exact_synonym: oio:hasExactSynonym
  created_by: oio:created_by
  date: dc:date
  comment: rdfs:comment
  xref: oio:hasDbXref

vars:
data_vars:
data_list_vars:
  syns: xsd:string
name:
  text: "%s swab collection"
  vars:
   - location

annotations:
  - annotationProperty: exact_synonym
    text: "swab collection of %s"
    vars:
     - location

  - annotationProperty: date
  - annotationProperty: created_by
  - annotationProperty: comment
  - annotationProperty: xref

exact_synonym:
def:
  text: "Collection biological material from within the %s using a swab."
  vars:
    - location

equivalentTo:
  text: "'swab' and 'has_input' some ('part_of' some %s)"
  vars:
    - location
```

‘Swab by location’ pattern

defined_class	defined_class_name	location	location_label
MAXO:0010301	rectal swab collection	UBERON:0001052	rectum
MAXO:0010302	vaginal swab collection	UBERON:0000996	vagina
MAXO:0010303	buccal swab collection	UBERON:0006956	buccal mucosa
MAXO:0010304		UBERON:0035612	nasal turbinate
MAXO:0010305	nasopharyngeal swab collection	UBERON:0001728	nasopharynx
MAXO:0010306		UBERON:0000402	nasal vestibule
MAXO:0010307	skin swab collection	UBERON:0000014	zone of skin
MAXO:0010308	oropharyngeal swab collection	UBERON:0001729	oropharynx
MAXO:0010309	anterior nares swab collection	UBERON:0005928	external naris
MAXO:0010310		UBERON:0000341	throat

```
vars:
data_vars:
data_list_vars:
  syns: xsd:string
name:
  text: "%s swab collection"
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annotations:
  - annotationProperty: exact_synonym
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annotations:
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     - location
```

Ontology imports

Currently Imported

- Relation Ontology (RO)
- Information Artifact Ontology (IAO)
- Food Ontology (FoodOn)*
- Chemical Entities of Biological Interest (CHEBI)
- Human Phenotype Ontology (HP)*+
- Uber-Anatomy Ontology (UBERON)
- Gene Ontology (GO)
- Neuro Behavior Ontology (NBO)*+

Pending

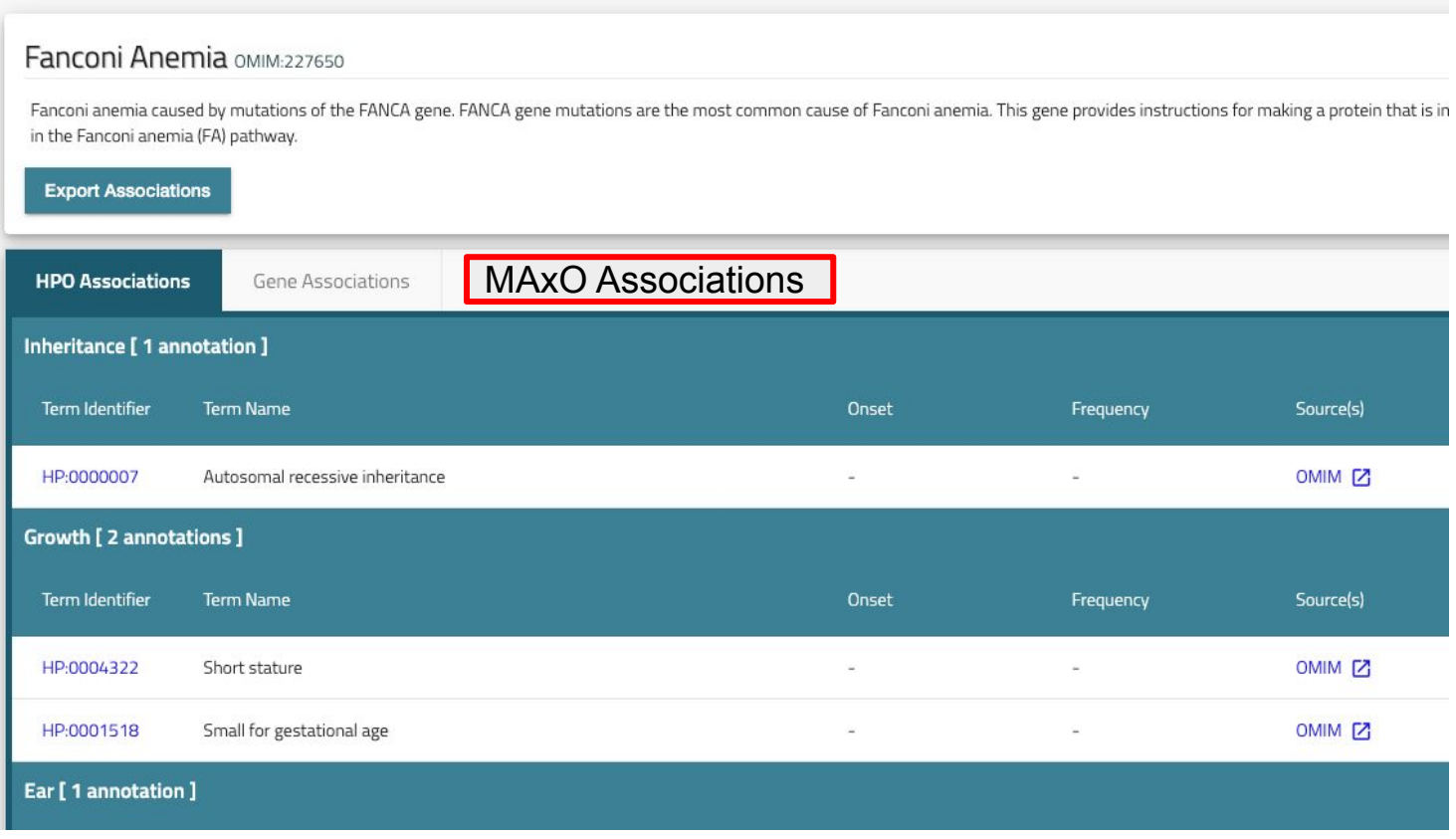
- Ontology for Biomedical Investigations (OBI)*
- Protein Ontology (PR)*
- *Ontology for Nutritional Studies (ONS)**
- Ontology for Medically Related Social Entities (OMRSE)*
- Cell Ontology (CL)

‘*’ Requested new terms
‘+’ Editor

COVID-19 related treatment terms

- Airway management terms (i.e, airway pressure release ventilation)
- CT pattern and terms (i.e., lung CT)
- Radiography pattern and terms (i.e., CT pulmonary angiogram)
- Clinical chemistry measurement terms (i.e., D-dimer/procalcitonin measurements)
- Swab collection pattern and terms
- Biomarker measurements (i.e., C-reactive protein biomarker assessment)
- Blood cell measurements (i.e., lymphocyte count, eosinophil count)
- Patient assessments (i.e., patient demographic characteristics assessment, self body temperature measurement)
- Viral detection assays (i.e., clinical RNA detection test)

Hpo.jax.org: Search disease, gene, or phenotype



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*Thank you and please contribute and collaborate!
https://github.com/monarch-initiative/MAxO/issues

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CD2H - National COVID Cohort Collaborative [310132-0621-02]

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