

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

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## \*Development/Curation

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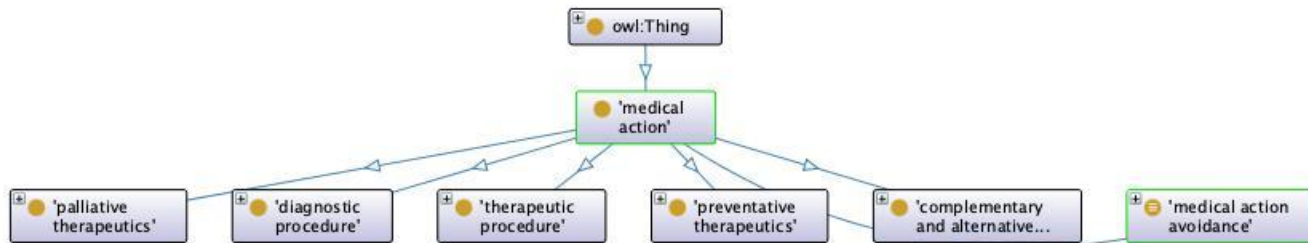
## \*Annotation/Text-mining software tools

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# Medical Action Ontology (MAxO) overview

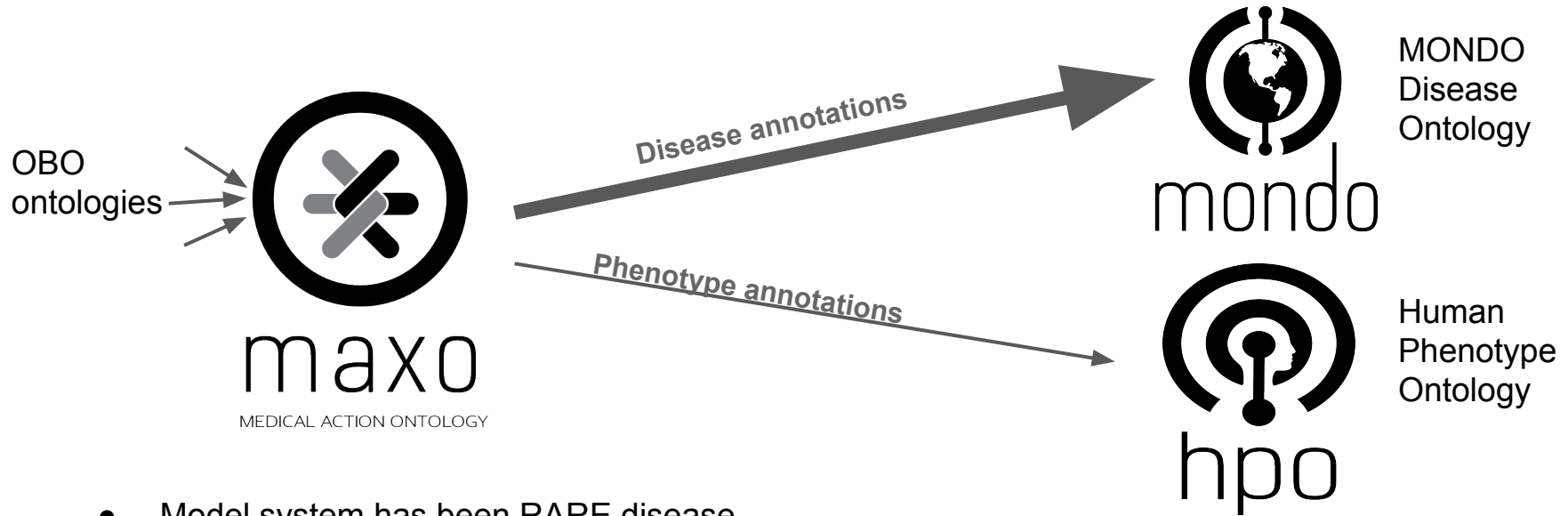
The Medical Action Ontology (MAxO) provides a structured vocabulary for medical procedures, interventions, therapies, and treatments.



- 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.



# USE case: Text-mining GeneReviews

- Goal: Enrich MxO by identifying new terms and extracting MxO-disease links
- Has easy to identify 'management' sections
- Identify new MxO 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.

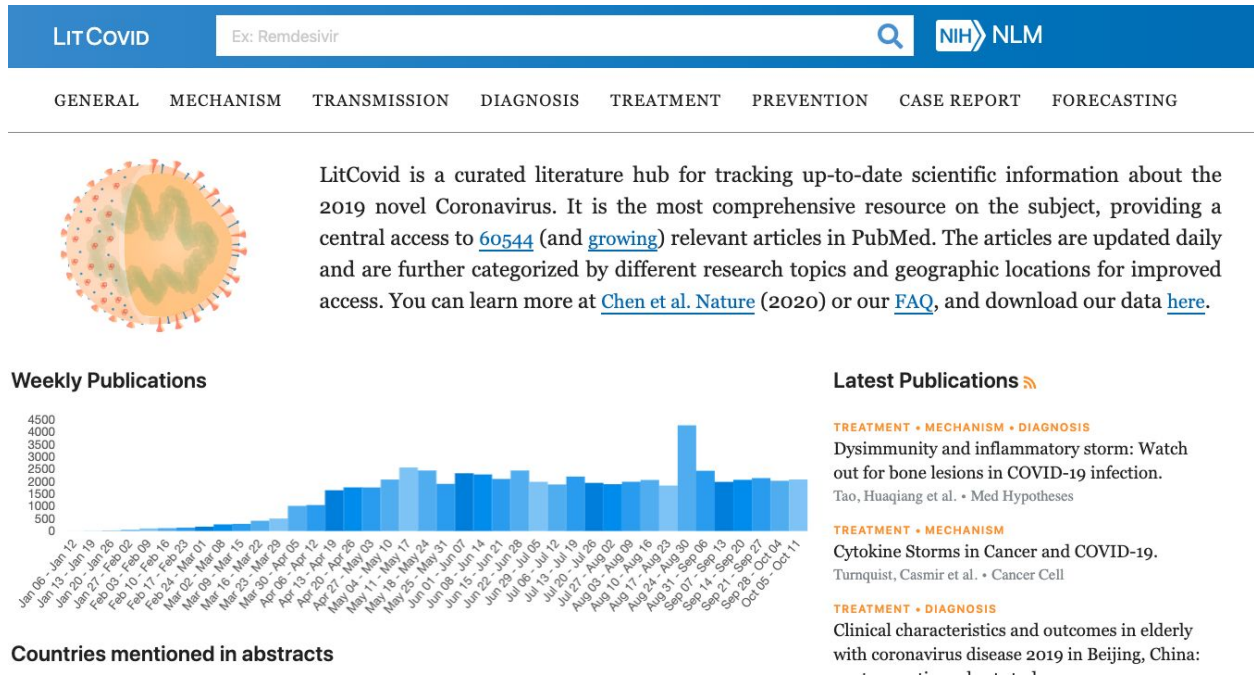
**Diagnosis/testing.** The diagnosis of FA is established in a [proband](#) with increased [chromosome](#) breakage and radial forms on [cytogenetic](#) testing of lymphocytes with diepoxybutane (DEB) and mitomycin C (MMC). The diagnosis is confirmed by identification of one of the following:

- 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.

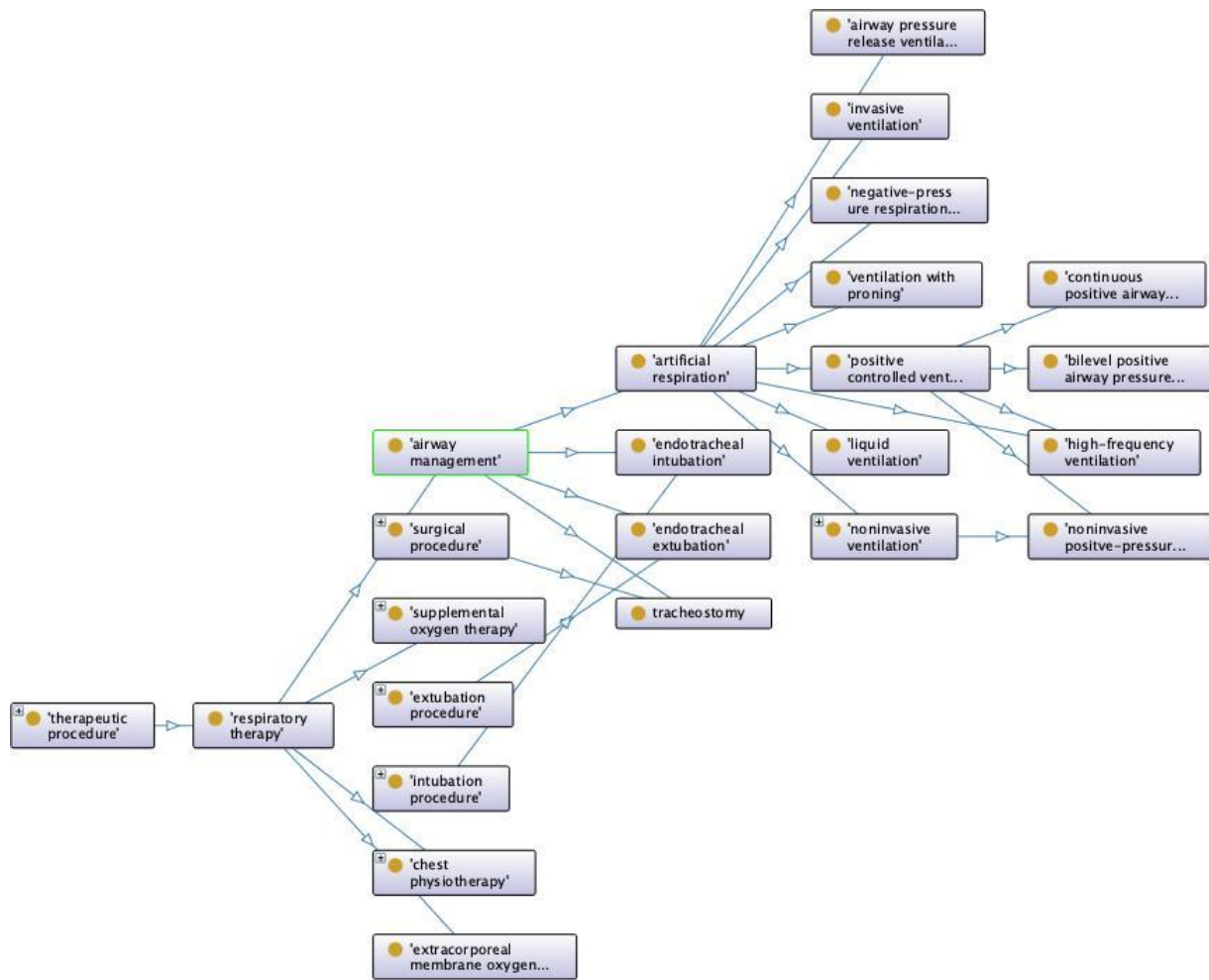
# COVID-19 literature resources

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

medical action > therapeutic procedure > respiratory therapy

Active ontology x Entities x Individuals by class x Individual Hierarchy Tab x DL Query x

Datatypes Individuals OntoGraf Import View OntoGraf

Classes Object properties Data properties Annotation properties

Class hierarchy: respiratory therapy

Annotations: respiratory therapy

radiation therapy  
renal replacement therapy  
respiratory therapy  
airway management  
artificial respiration  
airway pressure release ventilation  
high-frequency ventilation  
invasive ventilation  
liquid ventilation  
negative-pressure respiration  
noninvasive ventilation  
mask ventilation  
noninvasive positive-pressure ventilation  
positive controlled ventilation  
bilevel positive airway pressure ventilation  
continuous positive airway pressure ventilation  
high-frequency ventilation  
noninvasive positive-pressure ventilation  
ventilation with proning  
endotracheal extubation  
endotracheal intubation  
tracheostomy  
chest physiotherapy  
chest wall oscillation  
postural drainage  
extracorporeal membrane oxygenation  
supplemental oxygen therapy  
supplemental oxygen therapy with mask  
supplemental oxygen therapy with nasal cannula

Annotations: respiratory therapy

Annotations +

rdfs:label [language: en]  
respiratory therapy

definition  
Any form of treatment that acts to improve or assist with respiratory function.  
'database cross reference'  
NCIT:C15322

'database cross reference'  
NCIT:C15322

created\_by  
<http://orcid.org/0000-0001-7941-2961>

dc:date [type: xsd:dateTime]  
2020-01-23T15:55:17Z

Description: respiratory therapy

Equivalent To +

# 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"
  vars:
    - location
+annotations:
  - annotationProperty: exact_synonym
    text: "swab collection of %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"  
  vars:  
    - location  
-annotations:  
  - annotationProperty: exact_synonym  
    text: "swab collection of %s"  
    vars:  
      - 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)\*+

‘\*’ Requested new terms

‘+’ Editor

## Pending

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

# 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

## Fanconi Anemia OMIM:227650

Fanconi anemia caused by mutations of the FANCA gene. FANCA gene mutations are the most common cause of Fanconi anemia. This gene provides instructions for making a protein that is in the Fanconi anemia (FA) pathway.

[Export Associations](#)[HPO Associations](#)[Gene Associations](#)[MAxO Associations](#)

### Inheritance [ 1 annotation ]

Term Identifier	Term Name	Onset	Frequency	Source(s)
<a href="#">HP:0000007</a>	Autosomal recessive inheritance	-	-	<a href="#">OMIM</a> <a href="#">🔗</a>

### Growth [ 2 annotations ]

Term Identifier	Term Name	Onset	Frequency	Source(s)
<a href="#">HP:0004322</a>	Short stature	-	-	<a href="#">OMIM</a> <a href="#">🔗</a>
<a href="#">HP:0001518</a>	Small for gestational age	-	-	<a href="#">OMIM</a> <a href="#">🔗</a>

### Ear [ 1 annotation ]



\*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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