PhenoteFX Documentation

Release 0.4.5

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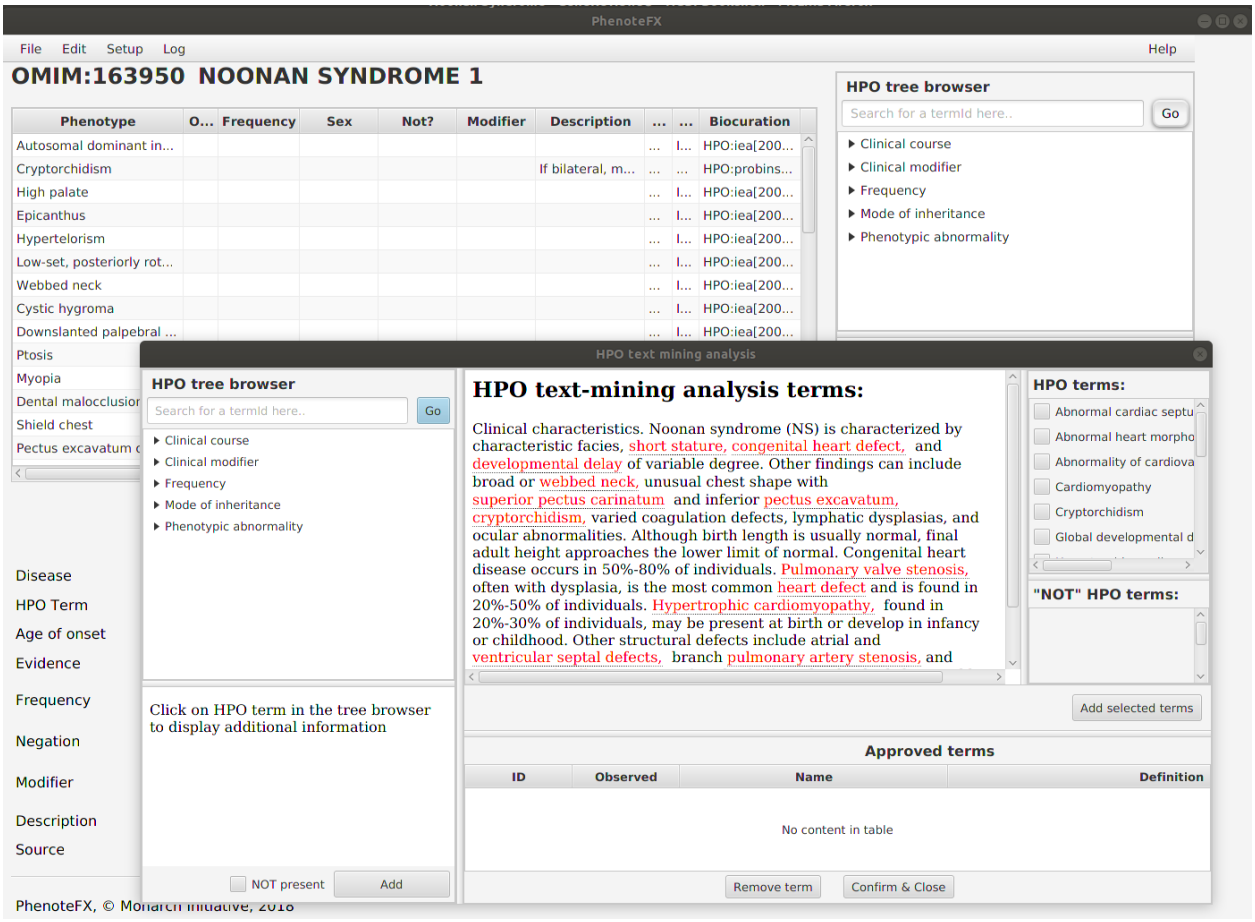
Installation Tutorial

1 Feedback 3

PhenoteFX is a Desktop Java App for working with Human Phenotype Ontology (HPO) Annotation data. It is designed to help curators revise or create phenotype annotation records for Human Phenotype Ontology (HPO) rare disease data. PhenoteFX is currently at version 0.8.5.



CHAPTER 1

Feedback

The best place to leave feedback, ask questions, and report bugs is the PhenoteFX Issue Tracker.

1.1 Installation

1.1.1 Use the prebuilt Jar file

Simply download the latest release of PhenoteFX from the GitHub releases page at https://github.com/monarch-initiative/PhenoteFX/releases.

Note: This is the recommended way of installing for normal users.

1.1.2 Install from Source

You only need to install from source if you want to contribute to the development of PhenoteFX yourself. To use PhenoteFX for annotation, it is recommended to download the prebuilt version.

Prerequisites

For building PhenoteFX, you will need

1. Java JDK 16 or higher for compiling OntoLib,
2. Maven for building PhenoteFX, and
3. Git for getting the sources.

Git Checkout

The following command downloads the sources of PhenoteFX.

```
# git clone https://github.com/monarch-initiative/PhenoteFX.git
# cd PhenoteFX
```

Building

You can build PhenoteFX using `mvn package`.

```
$ mvn package
```

You shuld now be able to start the app with the following command.

```
$ java -jar target/PhenoteFX.jar
```

Maven Proxy Settings

If you are behind a proxy, you may encounter problems with Maven downloading dependencies. Edit the `settings.xml` file in your `.m2` maven directory to adjust the settings for your proxy server.

```
<settings>
  <proxies>
   <proxy>
      <active>true</active>
      <protocol>http</protocol>
      <host>proxy.example.com</host>
      <port>8080</port>
      <nonProxyHosts>*.example.com</nonProxyHosts>
    </proxy>
  </proxies>
</settings>
END
```

1.2 Setting up PhenoteFX

When you use PhenoteFX for the first time, you need to download some files and tell PhenoteFX where you would like to store the annotation files.

1. **Download Medgen HPO OMIM** This is a file frlom Medgen that contains the names of OMIM entries; we use this file to translate between an item such as OMIM:100001 and Smith Syndrome. This file will be replaced by a MONDO file soon.

2. **Download HPO** This will download the latest release of hp.obo from the HPO GitHub page.

3. **Set biocurator id** Enter whatever you would like be be nano-attributed by, e.g., MGM:rrabbit.

4. **Set default directory for phenoteFX files** Usually this will be the directory to which you have downloaded the GitHub repository for the HPO annotations. Please choose the subdirectory `hpo-annotation-data/rare-diseases/annotated`.

5. **Show settings** This item opens a window to show the current settings.

The remaining menu items in the setup menu are experimental and not needed for rare disease annotation.

1.3 Using a local copy of the HPO for annotations

In many cases, new term requests on the HPO GitHub issue tracker contain both a new term request as well as a list of diseases that should be annotated with the new term. It would be nice to annotate the diseases at the same time as we make new terms. However, PhenoteFX uses the release version of the `hp.obo` file by default; since this file is updated only about once a month, we would have to wait to annotate the diseases.

Instead, PhenoteFX is able to use a local copy of the `hp.obo` file that has been created from the most up-to-date edit file of the HPO (`hp-edit.owl`).

1.3.1 How to create a local hp.obo version

Please see the tutorial at http://hpo-workbench.readthedocs.io/en/latest/latest.html We are working on making this slightly easier.

1.4 Working with PhenoteFX

PhenoteFX is designed to add HPO disease annotations to the HPO computational disease models (See for instance, the entry for Marfan syndrome). Annotations can be added one by one using the autocomplete menus or they can

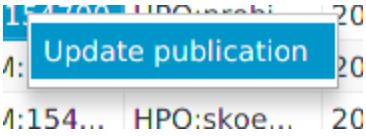
1.4.1 Updating the evidence code for an existing entry

Occasionally, we would like to update an existing annotation. For instance, we would like to add a PubMed citation for an entry that was created by text mining. In this case, we will update the evidence code from `IEA` (inferred from electronic annotation) to `PCS` (published clinical study). To do this, right click on the table cell with the evidence code and choose the updated evidence code. You will see a popup menu with the four available evidence codes. Click the desired code to update the code in the table.

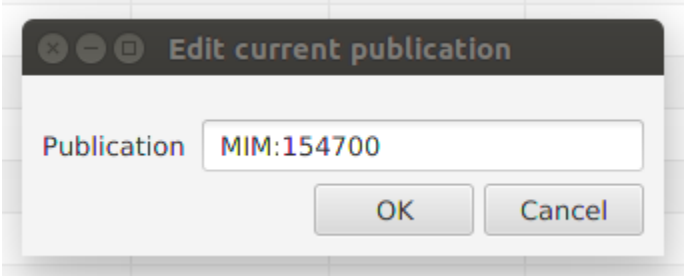
1.4.2 Updating the citation for an existing entry

The citation (reference) for an HPO annotation is kept in the **Pub** column. We will sometimes want to update this field. For instance, if an entry was created by text mining, we may have a citation such as "OMIM:154700", but say we have found a PubMed entry that provides more information. Then, we would like to update the entry to PMID:123, where 123 stands for the PubMed id. For instance, the if the HPO entry for Marfan syndrome shows an annotation for `Autosomal dominant inheritance` that is referenced to `OMIM:154700`, this indicates that the assertion was derived from the Online Mendelian Inheritance in Man (OMIM) page for Marfan syndrome. The article at PubMed id PMID:17502658 provides more detail about this assertion, and so we would replace `OMIM:154700` with `PMID:17502658`.

To do so, first right click on the publication entry for the annotation of interest. You will see a field to click on.



If you click on the field, a new dialog window will appear with the current publication entry. Replace or edit this entry and click on `OK` in order to change the publication. If you click on `Cancel`, the window will close and the entry will not be changed.



1.4.3 Updating the Description and Frequency fields

The description and the frequency fields can be updated in the same way as the publication field. Please note that for frequency, one of three valid formats must be used.

1. An HPO Term ID from the Frequency subontology, e.g., `HP:0040280`
2. An expression such as 4/6 (meaning that 4 out of 6 patients with the disease in question were found to have the phenotypic feature denoted by the HPO term).
3. An expression such as 45% (meaning that 45% of patients with the disease in question were found to have the phenotypic feature denoted by the HPO term).

PhenoteFX currently does not check that the format of the frequency term is correct, but this feature will be added in the future.