PhenDisco User Manual

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Getting Started

- The Getting Started menu will provide links to the PhenDisco home page, Top Level studies, user manual, PhenDisco domain model, and PhenDisco ontology.

Getting Started

- PhenDisco
- Browse Top Level Studies
- Download Manual
- Download PhenDisco domain model
- Download PhenDisco ontology

- Clicking the PhenDisco blue and green disco ball on any page will also return you to the home page.

How to Search

Entering Search Terms:

Enter search terms into the search bar.

Syntax:
- Use quotation marks when you want to search two-word terms. Example: "heart attack"
- Connect multiple terms by Boolean operators (AND, OR).
-

Type of search:

- The default search type is "concept-based". This expands your search using synonymous or more specific terms.
- For example, when you search "heart attack" using the concept-based option, PhenDisco searches other synonymous terms such as "heart attack", "myocardial infarction", and "MI".
- If you want to retrieve the studies that have your search term in its exact form in the database, unclick the "concept-based" box on the right side of the search bar.

Syntax for Filtered Search:

- If you want to search studies by a specific field, you can conduct a filtered search by typing the specific field and then the value you are searching for.
- For example, if you want to search for studies on "heart attack" and you want those studies to have IRB approval and the maximum age to be 50 yeas old, you would type into the search bar:

"heart attack" AND IRB: required AND AgeMax: [to 50]"
- Here is a list of specific fields and what values can be used:
    - **Title:** Study Title
    - **Desc**: Study Description
    - **Type:** Study Type ex. Case-control (Any of the terms from Study Design on the Advanced Search page are acceptable values)
    - **Cohort:** Number of Participants
    - **Platform:** Platform ex. HiSeq 2000 (Any of the terms from Platform on the Advanced Search page are acceptable values)
    - **IRB:** "No" or "Required"
    - **ConsentType:** "Restricted", "No", or "Unspecified"
    - **AgeMin:** [# to] means from age # and older
    - **AgeMax:** [to #] mean ages under #
    - **PhenDesc:** Phenotype description
    - **PhenID:** Phenotype ID
    - All values are searched in the selected field by partial string matching except the terms in quotation marks.

Advanced Search:

- The advanced search allows you to pre-select specific search parameters for a more specific search
- Advanced search fields:
    - Sample Size
    - Age
    - Sex
    - IRB
    - Consent Type
    - Study Design
    - Study Subject
    - Ethnicity
    - Race
    - Nationality/Ethno-Religious Group
    - Platforms
    - Machine
    - Data Analysis Methods
    - Sequencing Techniques

- The values in each field are organized in a hierarchical structure. A yellow arrow next to a value indicates that it is a higher-level value and there are more specific terms beneath it. By clicking the yellow arrow, the value will expand to show all of the more specific values. If you select a higher-level value, the more specific
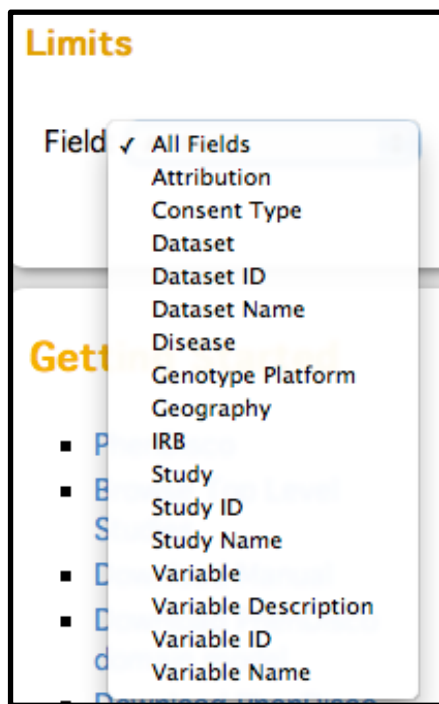
values are automatically selected. You can unselect and select each term specific to your search.

Study Design:

- ☐ › Interventional Studies
- ☐ Multicenter Study
- ☐ ˅ Observational Studies
    - ☑ ˅ Case-Control Study
        - ☑ Nested Case Control Study
- ☐ › Cross Setional Study
- ☐ Genome Wide Association Study
- ☐ › Longitudinal Study
- ☐ Population Based Study
- ☐ Prospective Cohort Study

Limits:

- You can limit your search by clicking on the Limits button below the search bar. This opens a box for Limits options.
- You can limit your search to the following fields by using the drop down menu: Attribution, Consent Type, Dataset, Dataset ID, Dateset Name, Disease, Genotype Platform, Geography, IRB, Study, Study ID, Study Name, Variable, Variable Description, Variable ID, Variable
- Once limits are selected, click the search button.
- For example, if you wanted to search for a specific study, e.g., NHLBI GO-ESP: Early-Onset Myocardial Infarction, type the name of the study into the search box and limit your search to "Study Name".

Limits

Field
- ✓ All Fields
- Attribution
- Consent Type
- Dataset
- Dataset ID
- Dataset Name
- Disease
- Genotype Platform
- Geography
- IRB
- Study
- Study ID
- Study Name
- Variable
- Variable Description
- Variable ID
- Variable Name

How to View Results

Basic Results:

	Title	Embargo Release	Details	Participants	Type of Study	Platform
☐	phs000179.v2.p2 Genetic Epidemiology of **COPD** (COPDGene) Funded by the National Heart, Lung, and Blood Institute	Version 1: passed embargo Version 2: 2013-06-19	V D A S	10364	Case-Control	HumanOmni1_Quad_v1-0_B
☐	phs000209.v10.p2 Multi-Ethnic Study of Atherosclerosis (MESA) Cohort	Versions 1-7: passed embargo Version 8: 2012-11-01 Version 9: 2013-03-08 Version 10: 2013-05-01	V D A S	8296	Longitudinal Family	

- Above the table you will see how many studies are being displayed out of the total studies found.
- There are arrows to view each page of the results.

Results

- If a study has a daughter study. A pedigree icon will appear in the first column. By placing the mouse over the icon you cans see details on the daughter study. By clicking this icon, you will be linked to the daughter study.

Display Options:
- For results of studies: the default result table includes Title, Embargo release, details, Participants, Type of Study, and Platform.

- Custom result display: you can set result display by clicking display options. Click data The "Apply" button will update the result table with the selected fields. You can move the mouse over each option to view a description. These study display options include:
- **Title**: Information about study name and ID
- **Study Type**: Information about type of study conducted
- **Sample size**: Number of participants by gender, race, ethnicity, age group, sample role
- **Details**: Contains options to view study Variables, Datasets, Analyses, and Study Documents
- **Embargo Release**: The expiration date of publication exclusivity rights given to contributing investigators
- **Platform**: Type of sequencing platform used
- **Geography**: Geographical location of the study
- **Links**: additional information

- **IRB**: Whether Institutional Review Board is required or not
- **Consent Type:** Data use constraint
- **Topic Disease**: Health topic of the study

Display Options

☑ Title	☑ Embargo Release	☐ Links	☐ Geography
☑ Study Type	☑ Platform	☐ Consent Type	☐ IRB
☑ Sample Size	☑ Details	☐ Topic Disease	

Apply   Restore Defaults   ?

- To restore the default view, click "Restore Defaults."
- Move the mouse over the question mark on the right side of the page to get these directions.
- Scroll horizontally at the bottom of the display table to view additional columns.

Search Relevancy:

- Scroll over a study, the row is highlighted in yellow and the Study Name and ID will appear in the popup box.
- The search terms will be highlighted in yellow where it shows up in Study Names and descriptions.
- The studies are presented in order of relevancy

	Title	Embargo Release	Details	Participants	Type of Study	Platform
☐	phs000294.v1.p1 STAMPEED: **Myocardial Infarction** Consortium	Version 1: passed embargo	V D A S	6042	Case-Control	AFFY_6.0
☐	phs00027 NHLBI GO- Onset **Myc** **Infarction** (Broad EOMI)				Sequencing	Genome Analyzer II

phs000294.v1.p1
STAMPEED: **Myocardial Infarction** Genetics Consortium (MIGen)
Study Description **Myocardial infarction** (MI) is a common...

Viewing Studies:

- Click on the Study Name and ID in the Title box for the study to open in a new page.

Exporting Study Results:

- There are three buttons on top of the display table: "Display All", "Check All", and "Export Selections".
- The "Check All" button will select each of the studies present on that page.
- The "Export Selections" will export a list of Study ID's and other metadata (see the display option) of the checked studies in the comma separated value (CSV) format.
- If you only want to export certain studies that you are interested in, after selected studies, you can click "Display" to only display selected studies or "Display All" to display all studies in default results.

Viewing Phenotype Variables:

- By clicking the variables tab under results, you can display all of the relevant variables of your search.

- Scroll over a variable, the row is highlighted in yellow.
- The search terms will be highlighted in yellow where it shows up variable descriptions.
- Scroll over the Variable Name and a popup box will appear with for information on that variable.

- Click on the Variable ID for the variable to open in a new window.

Exporting Variable Results:

- There are three buttons on top of the display table: "Display All", "Check All", and "Export Selections".
- The "Check All" button will select each of the variables present on that page.
- The "Export Selections" will export a list of Variable ID's and other metadata (Phenotype ID, Study ID, DataSet ID, DataSet Name, Phenotype Name, Phenotype Description, Data Type, Data Value, MIN, MAX, Normed Phenotype Description, Theme, Theme PCN, Theme CUI, Theme Semantic Type, Topic, Topic CUI, Topic Semantic Type, SOI, SOI CU, SOI Semantic Type, Category) of the checked studies in the comma separated value (CSV) format.
- If you only want to export certain variables that you are interested in, after selected variables, you can click "Display" to only display selected variables or "Display All" to display all variables in default results.