LOVD3 Course
Create gene variant database (LSDB)

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Contents

1. Introduction

We provide two articles as example:

1. "Mutations in ABHD12 cause the neurodegenerative disease PHARC: an inborn error of endocannabinoid metabolism" (Fieskerstrand et al., 2010 PubMed: PMC2933347)

2. "Mutations in IMPG2, encoding interphotoreceptor matrix proteoglycan 2, cause autosomal-recessive Retinitis pigmentosa" (Bandah-Rozenfeld et al., 2010 PubMed:PMC2917719)

Choose an article to use as a template for making your new gene variant database. But you may use whatever you like (other articles, personal data, own database, OMIM etc.). In the examples provided in this course manual we will use the first article.

Before you create a gene variant database, you should decide which functional fields (columns) you would want in the database, e.g. look at the mutation tables in the articles.

- Decide which reference sequence to use.
- In case you select the NG, always check that the transcript you want to use is contained in the reference sequence file.
- Look if the disorder has an OMIM entry.
- Go to http://courses.lovd.nl/LOVD3/ and select the directory corresponding to the number assigned to you.

2. Creating a gene variant database (LSDB)

The objective of this chapter is:

1. Add a new gene to the database.

To start this chapter:

- Log in as Manager (with for every one the same username: manager, password: manager1).

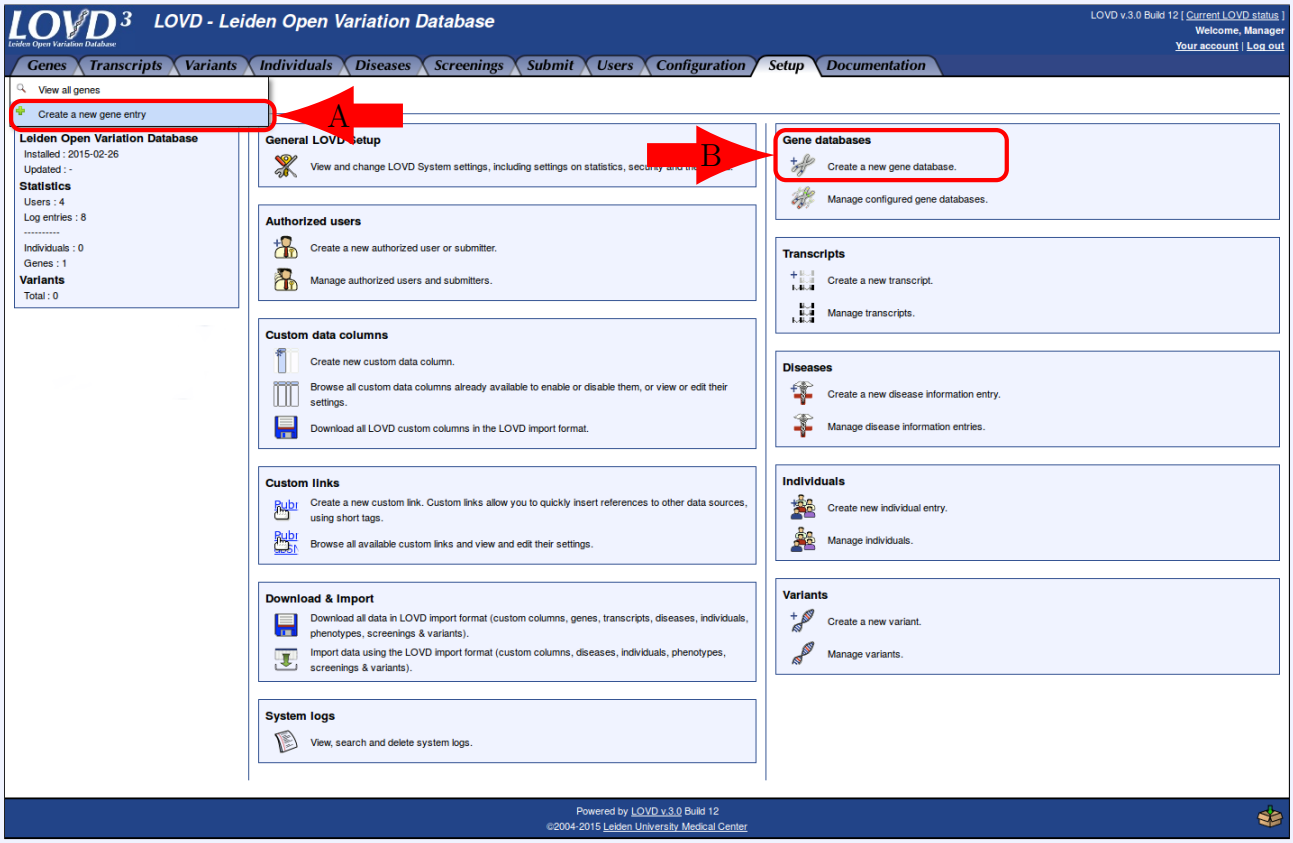

Figure 2.1: Create a new gene. You can do that via the "Create a new gene entry" link from the Genes menu tab drop down menu (A).
You can also do that from the Setup area and click the "Create a new gene database" link listed under "Gene databases" (B).

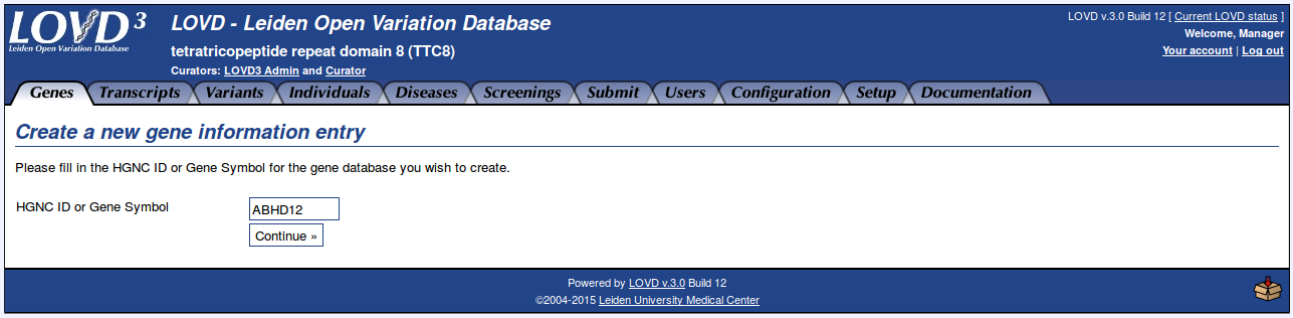

Figure 2.2: Insert the HGNC gene symbol.
You can find the symbol at the HGNC (http://www.genenames.org/) or at the NCBI entrez gene. In our example we will use the gene symbol ABHD12. Hereafter, press "Continue".

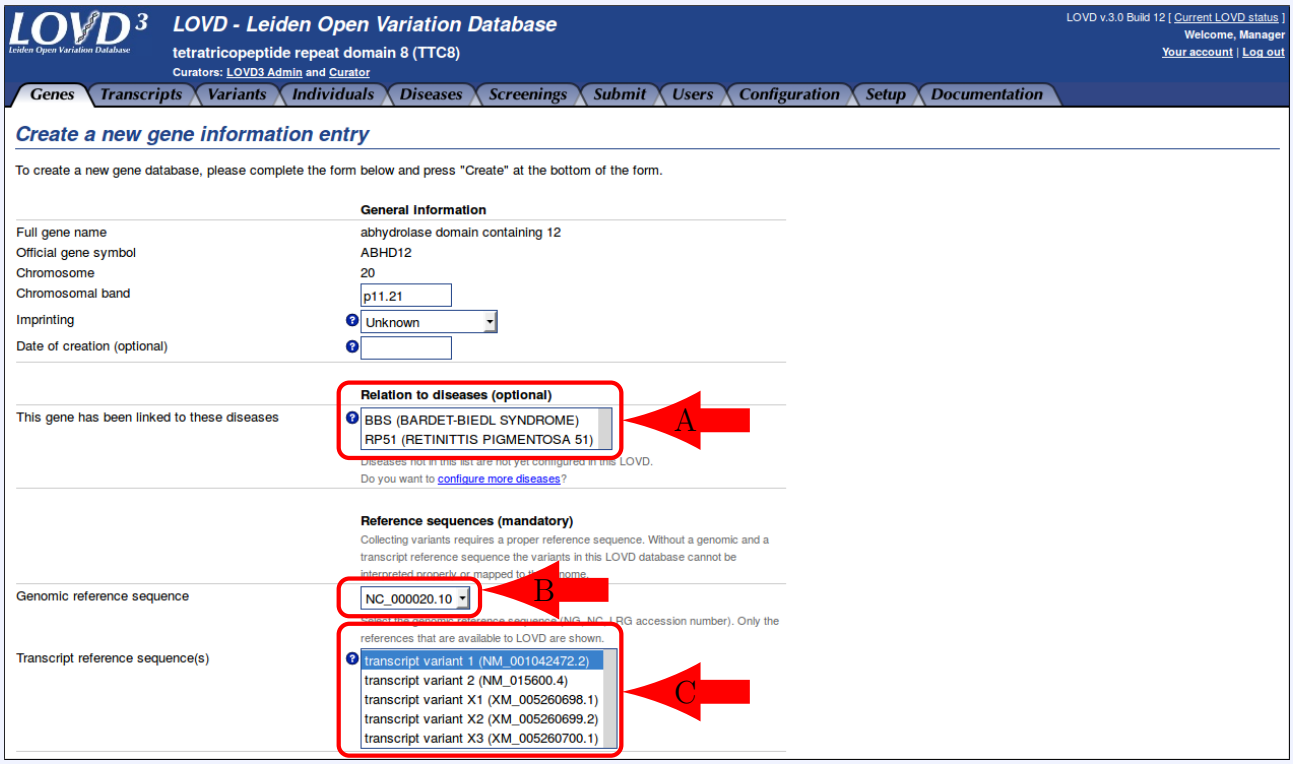

Figure 2.3: You can link a Disease to this gene (A). But the genes listed here are from previous exercises and are not linked to our current gene. We could create diseases from here, see figure 2.3 in course manual "Curate gene variant database" how. For now, do not select any disease, we will create a new disease later.
For "Genomic reference sequence", select "NC_000020.10" (B) and for "Transcript reference sequences", select "NM_001042472.2" (C).

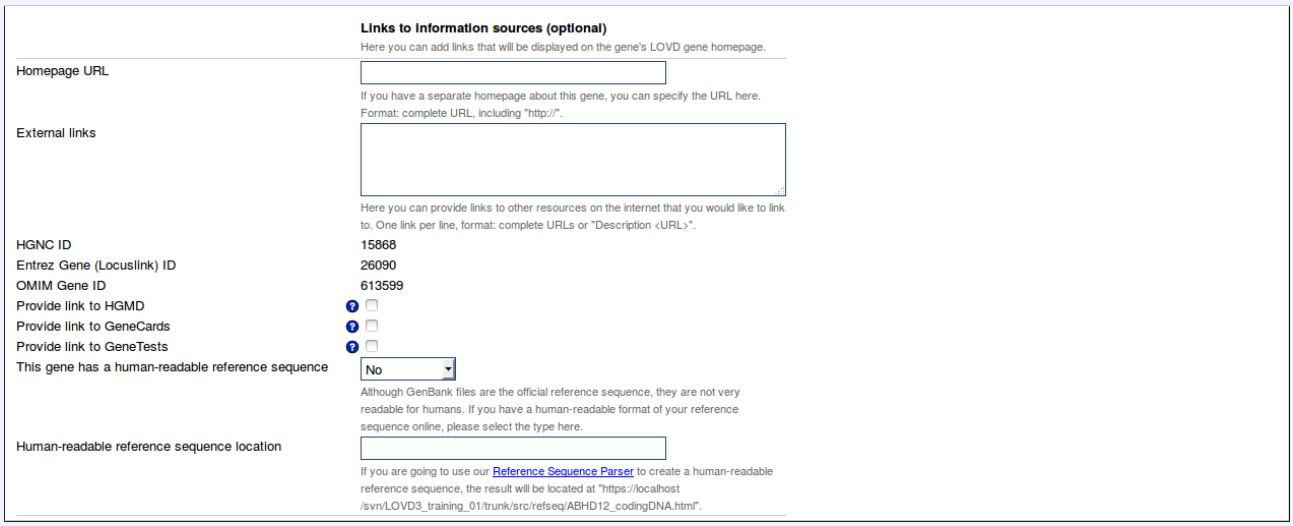

Figure 2.4: You can provide links to other resources on the internet that you would like to link to.

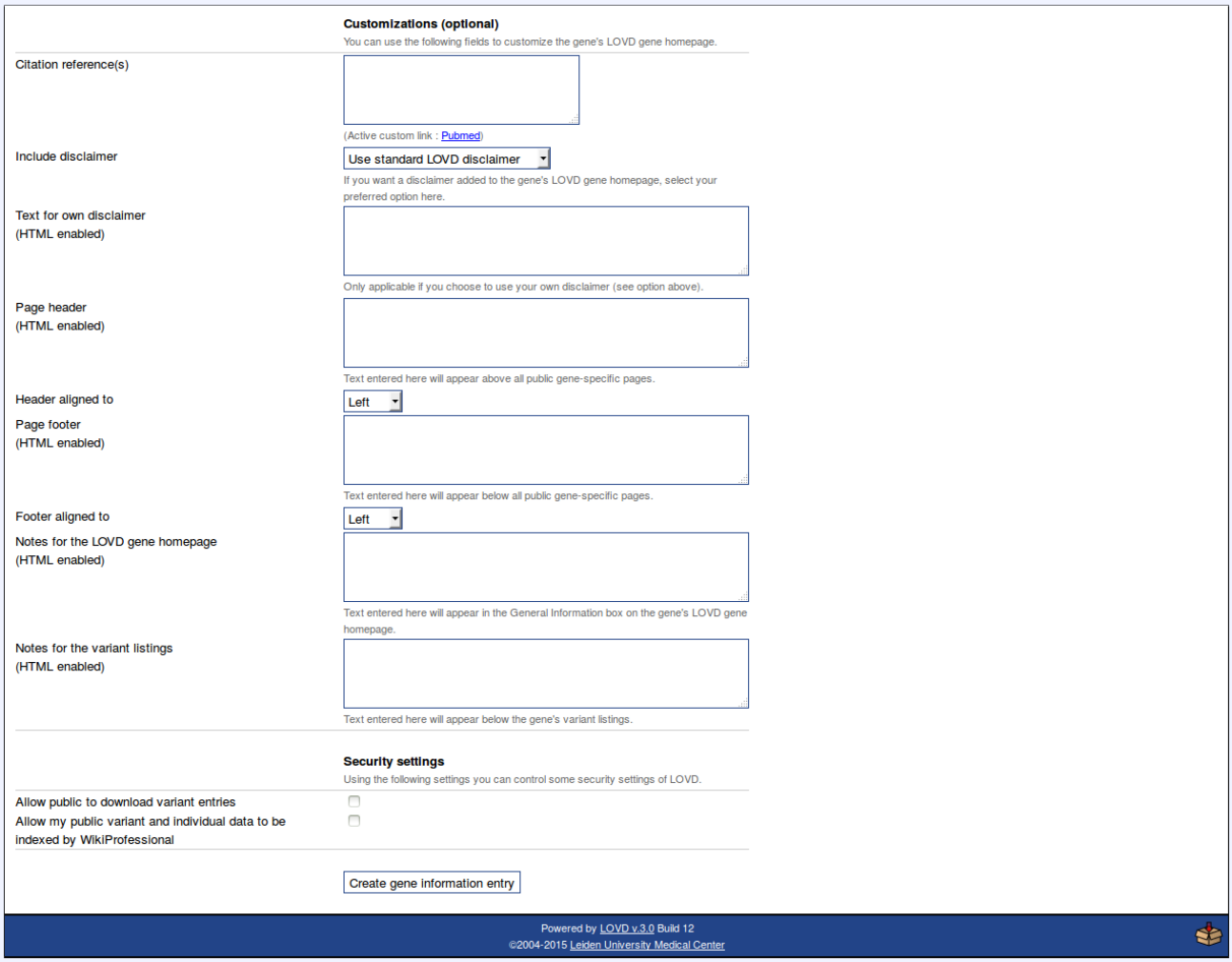

Figure 2.5: You can add a disclaimer, page header or page footer to the gene's LOVD gene homepage. Click "Create gene information entry" when you are ready.
Hereafter you are redirected to "Authorize curators for the gene". We will come back later on this subject in chapter "Creating and managing users". For now, click "Cancel".

3. Creating a disease

The objective of this chapter is:

1. Create a new disease and link this disease to our gene.

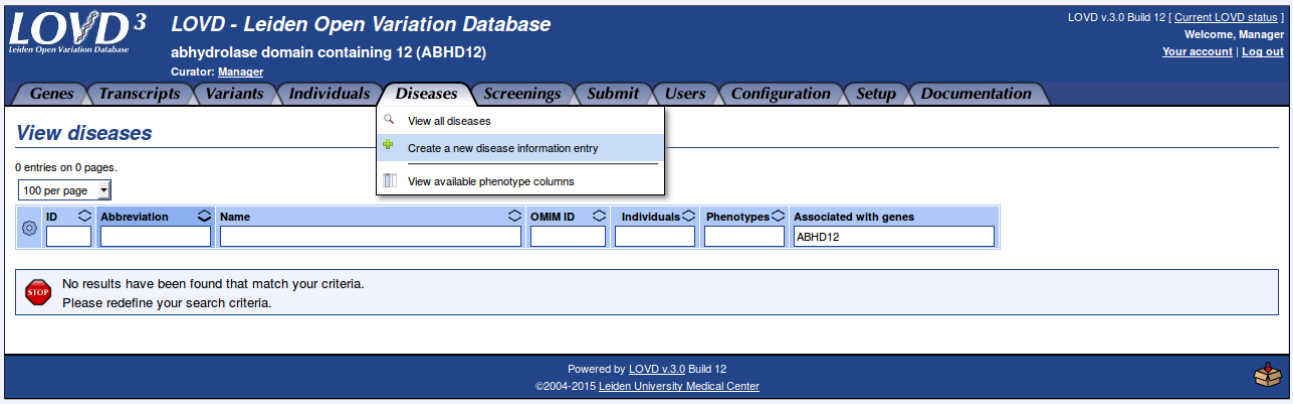

Figure 3.1: To create a new disease, click "Create a new disease information entry" from the Disease menu tab.

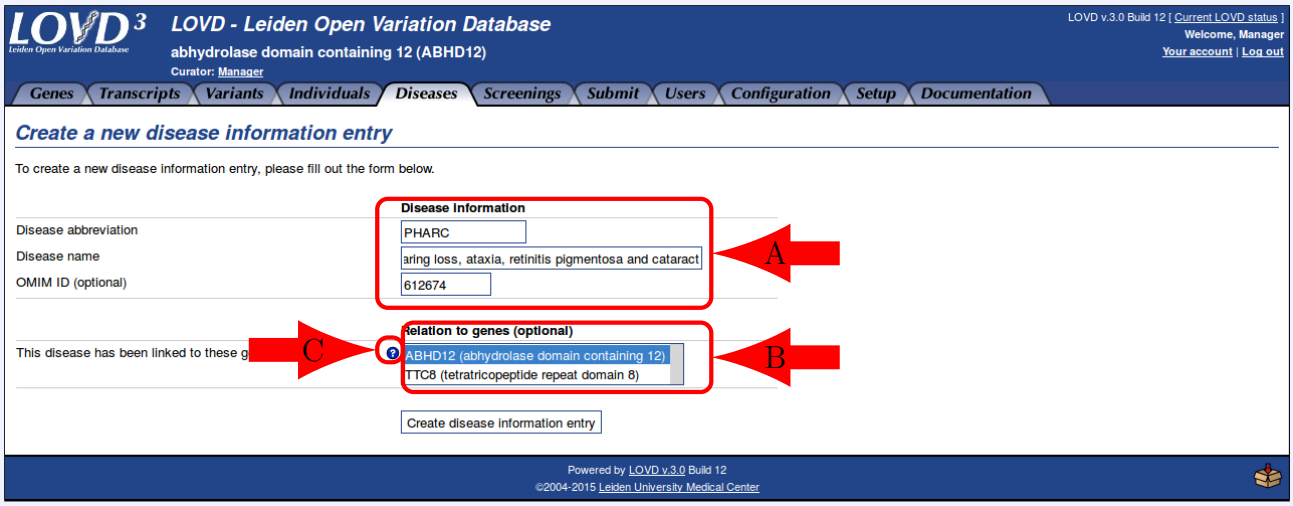

Figure 3.2: Give disease abbreviation, name and OMIM ID (A).
You can make a relation with a gene by selecting one or more genes (B). See help text for how to select multiple genes (C). In our example, we select ABHD12 and click "Create disease information entry".

4. Creating and managing columns

The objective of this chapter is:

1. Create a new custom column.
2. Manage the custom column.

To start this chapter:

- See chapter "Editing columns and legends" in course manual "Curate gene variant database" for some additional exercises on custom columns.

4.1 Create new custom columns

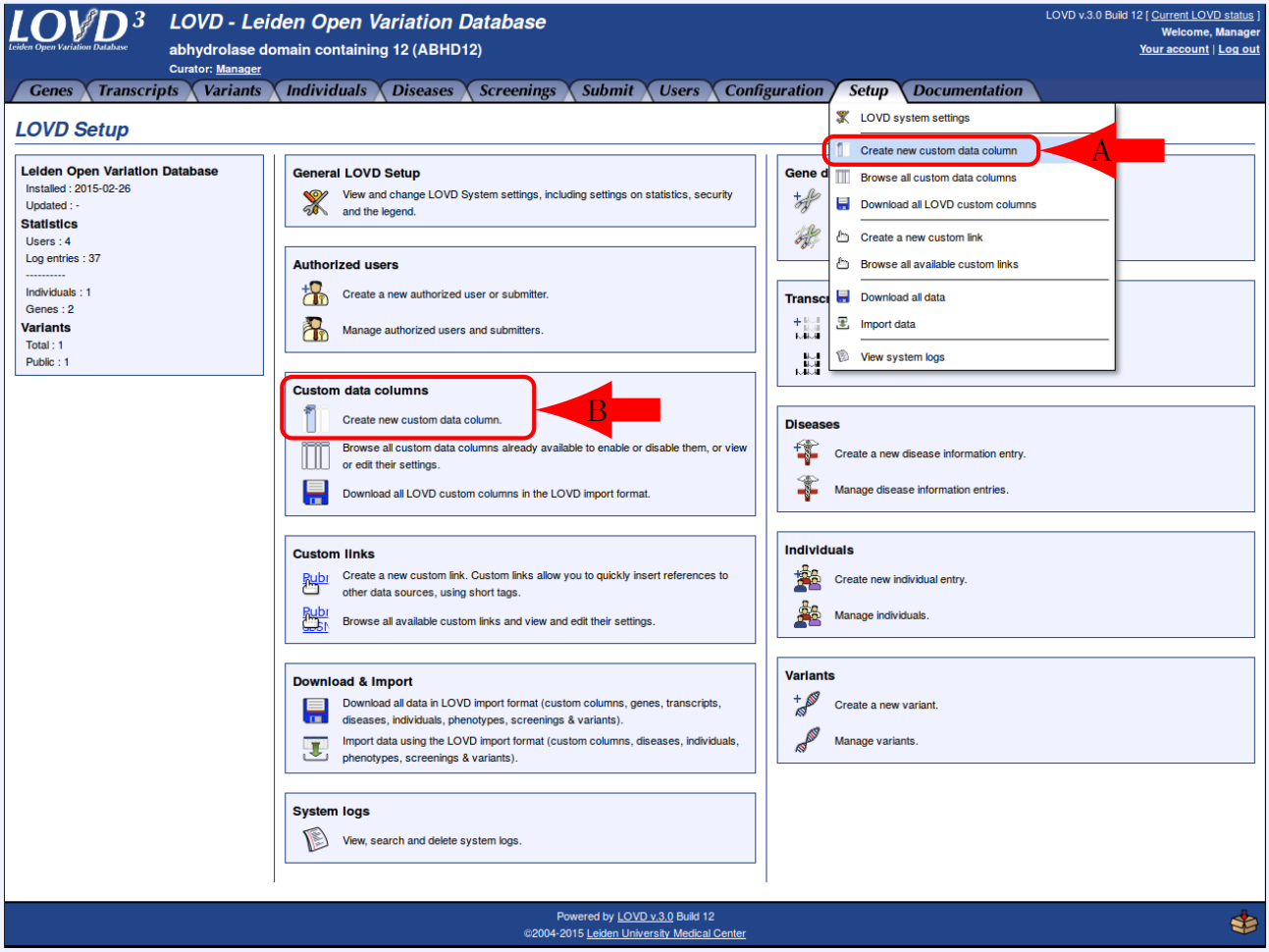

Figure 4.1: You can create a new column via the "Create new custom data column" link from the Setup drop down menu tab (A), or you can click "Create new custom data column" from the Setup area (B).

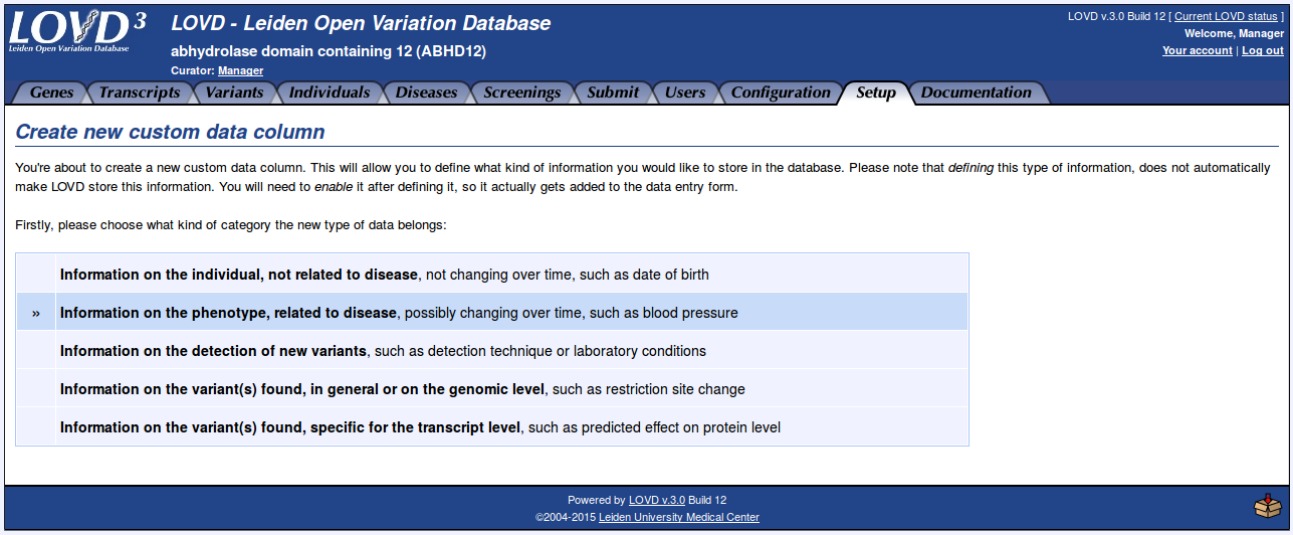

Figure 4.2: Select the category to which the new type of data belongs to. In our example we will add a new column "Neurography and EMG" (see table 1 in Fiskerstrand et al., 2010). This is a phenotype field related to disease, so select "Information on the phenotype, related to disease".

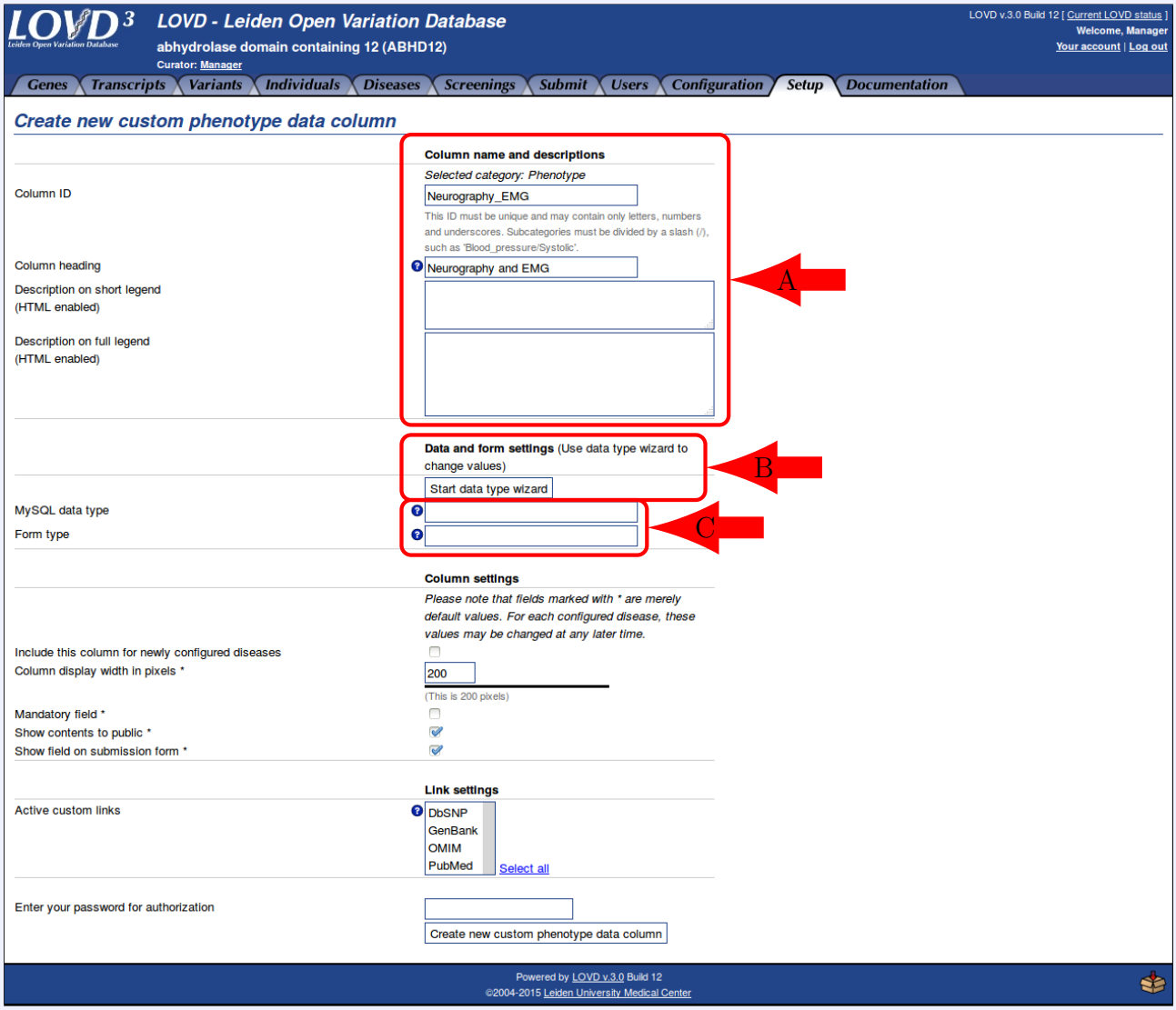

Figure 4.3: Fill the fields under section "Column name and description": Column ID, Column heading, Description on short legend and Description on full legend (A).
Then press "Start data type wizard" under section "Data and form settings" (B). This will open a pop-up screen where you can determine the data type for your new column. The results of the data type wizard are saved in the fields "MySQL data type" and "Form type" (C).
Only if you really know what you're doing, you can edit "MySQL data type" and "Form type" directly (C).

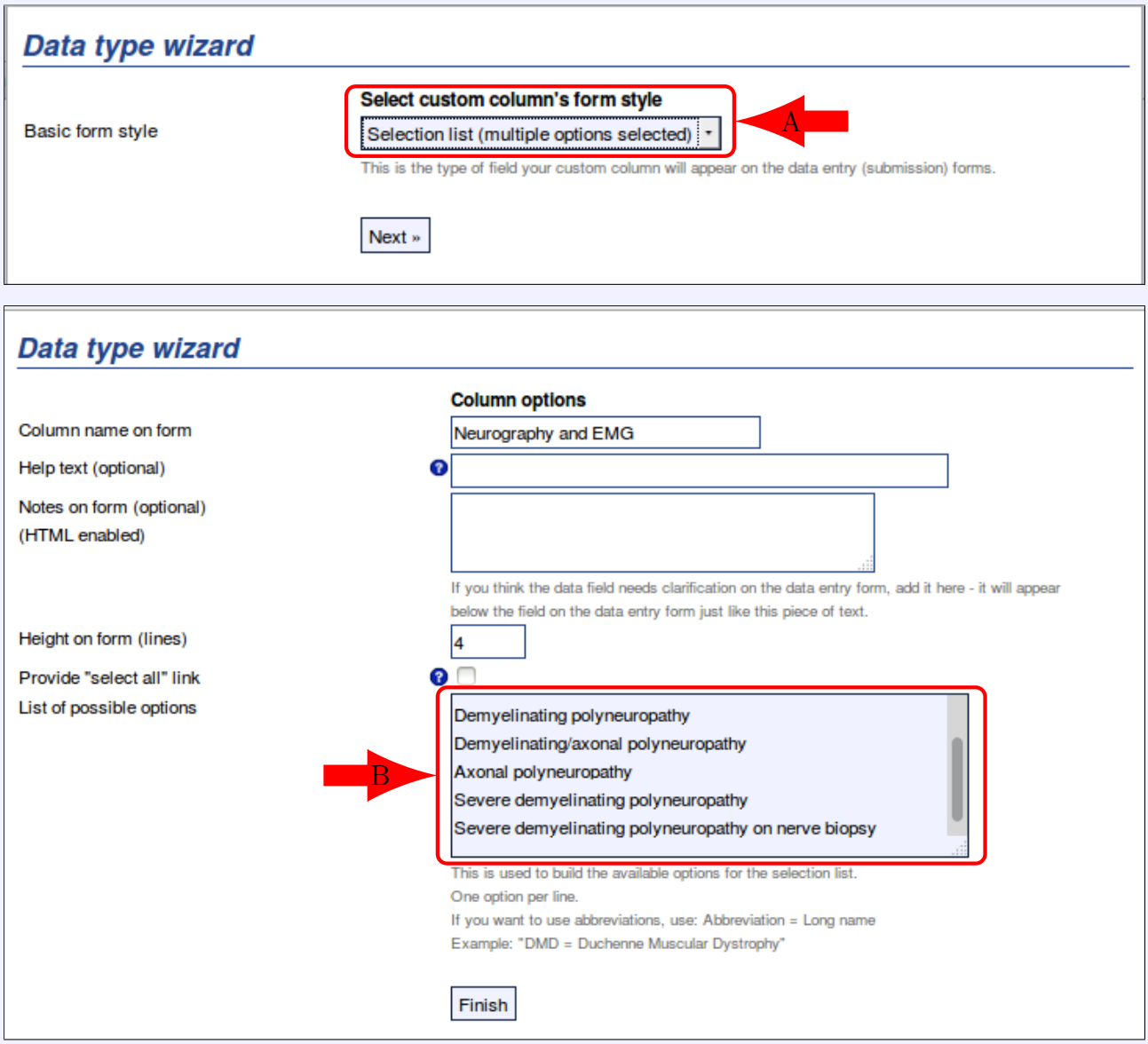

Figure 4.4: Here you may choose between the following type of input fields (A): Text/numeric, Integer, Decimal, Large multi-row textual, Drop down list (1 option selected), Selection list (multiple selection), date and On/Off checkbox. In our example we use "Selection list (multiple options selected)". Press "Next" when you're ready.
In the second data type wizard form you can list the options (B). Use the data from the "Neurography and EMG" (see table 1 in Fiskerstrand et al 2010) column. Put each option on a new line and press "Finish" when you are ready.

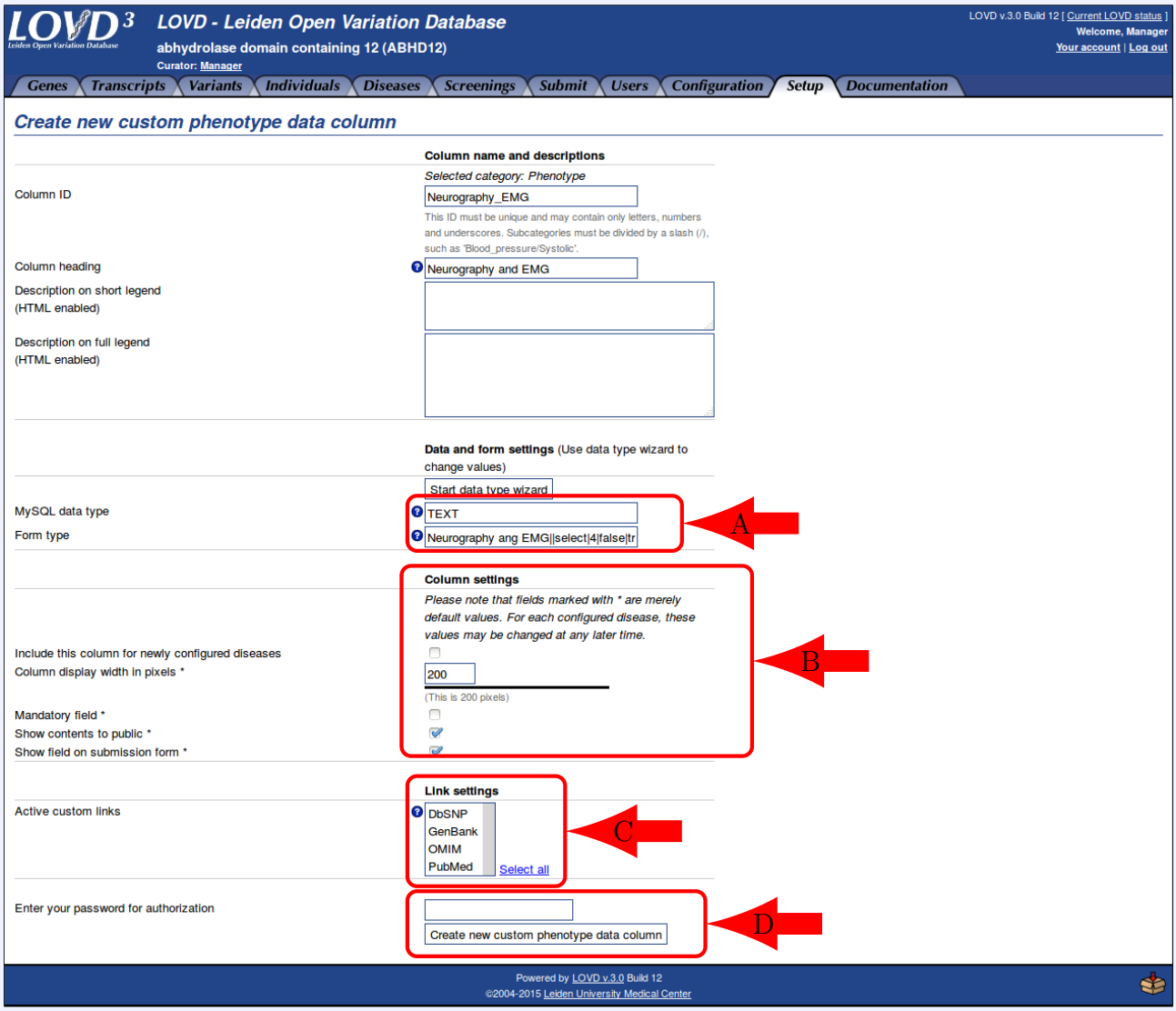

Figure 4.5: The data type wizard will automatically fill the fields “MySQL data type” and “Form type” (A). Choose column settings (B), specific custom links for this column (C), confirm with your password and press “Create new custom phenotype data column” (D) when you are ready.

4.2 Managing custom columns

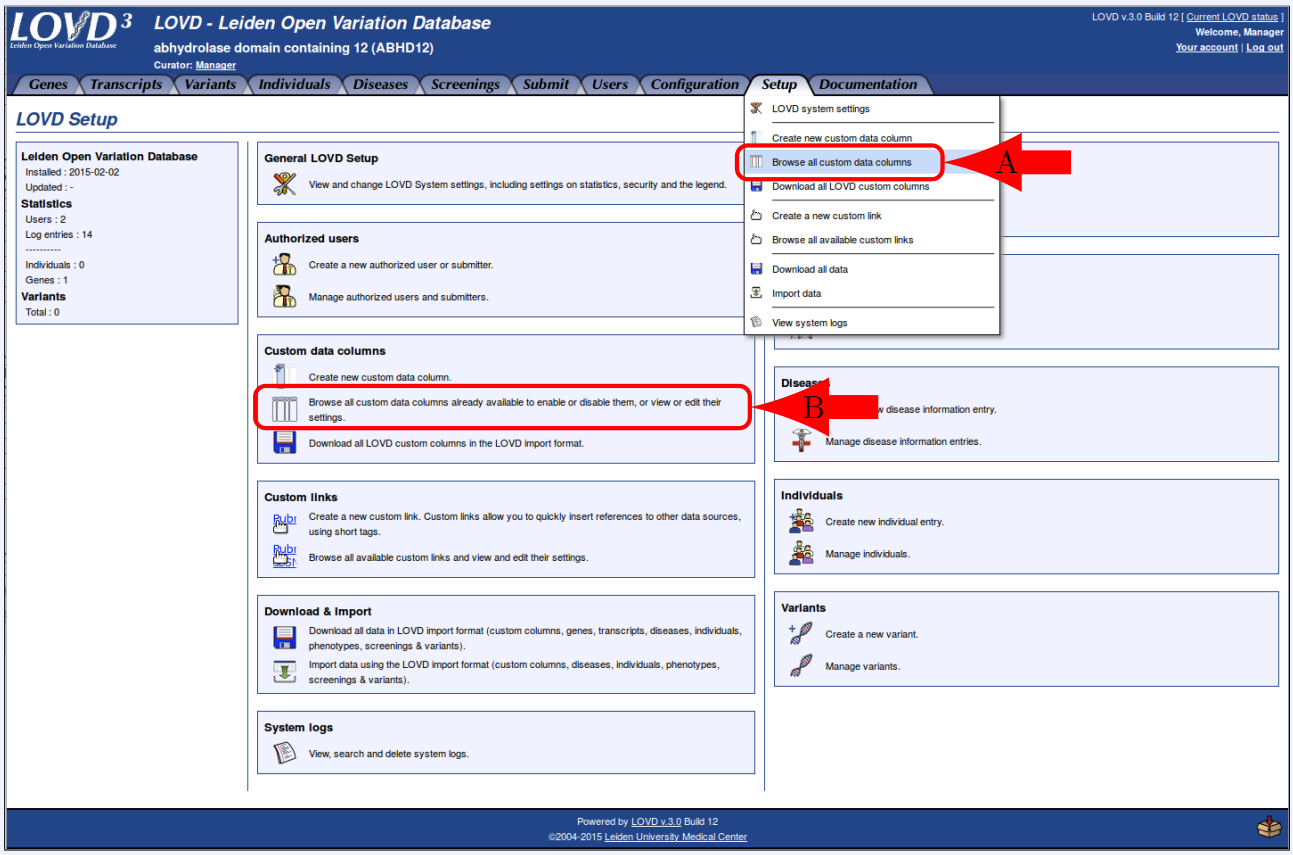

Figure 4.6: You can browse all custom columns via the "Browse all custom data columns" link from the Setup drop down menu tab (A), or you can select "Browse all custom data columns (...)" from the Setup area (B).

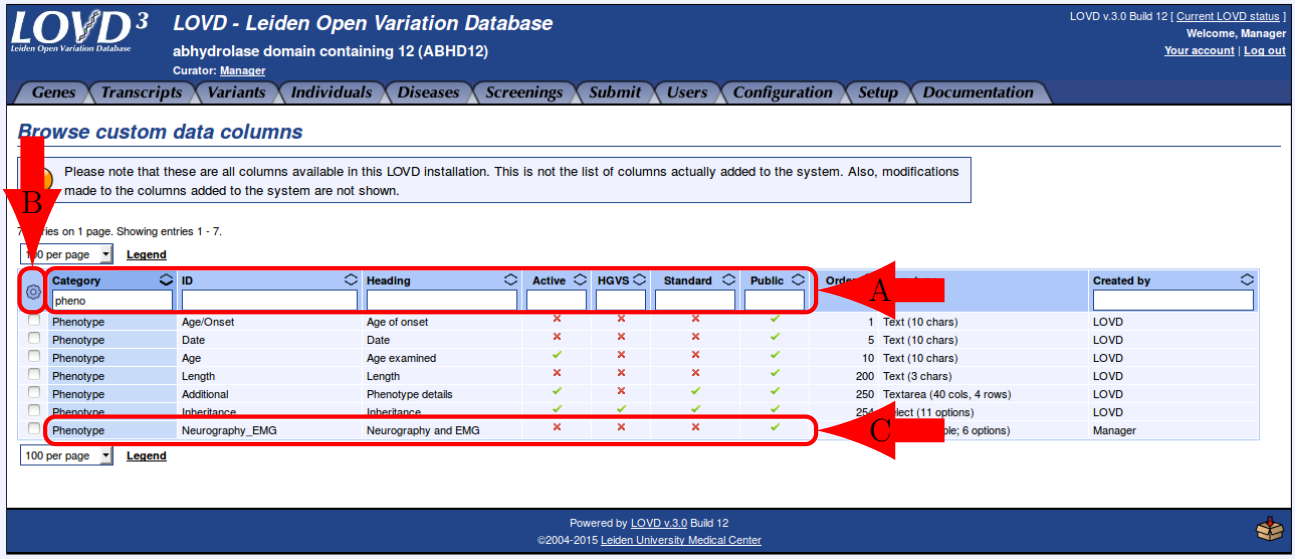

Figure 4.7: If you want to display custom columns only applicable for phenotype, you can use the headers as a filter (A). Or you can use the menu on the left (B) and select "Show only Phenotype columns". Look for your newly created custom column (C). You can see that your new custom column is not active yet.
Click anywhere on the row to go to the details of your custom column.

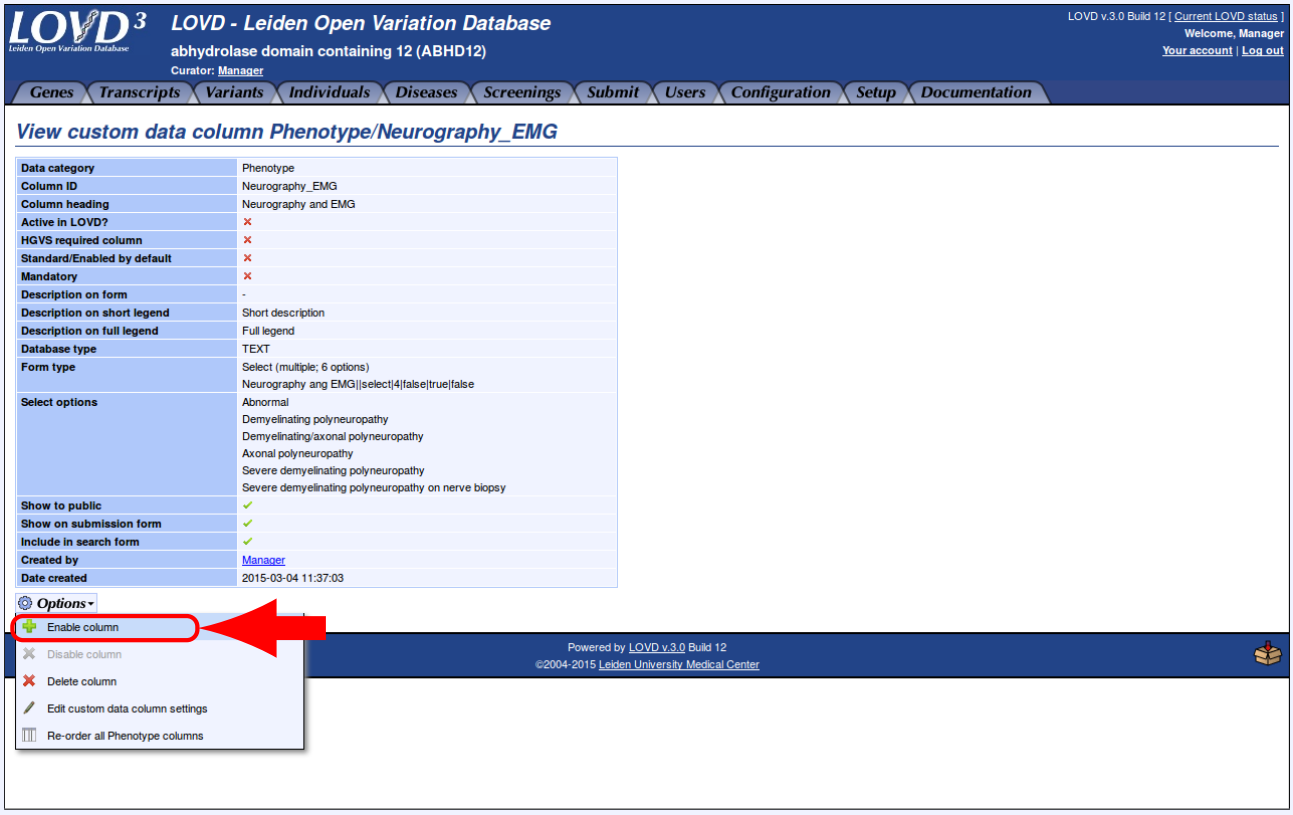

Figure 4.8: Click the Options drop down menu and click "Enable column". If you click "Edit custom data column settings", you will go to the form "Edit custom data column". This form is similar to the form "Create custom data column", see figure 4.3.

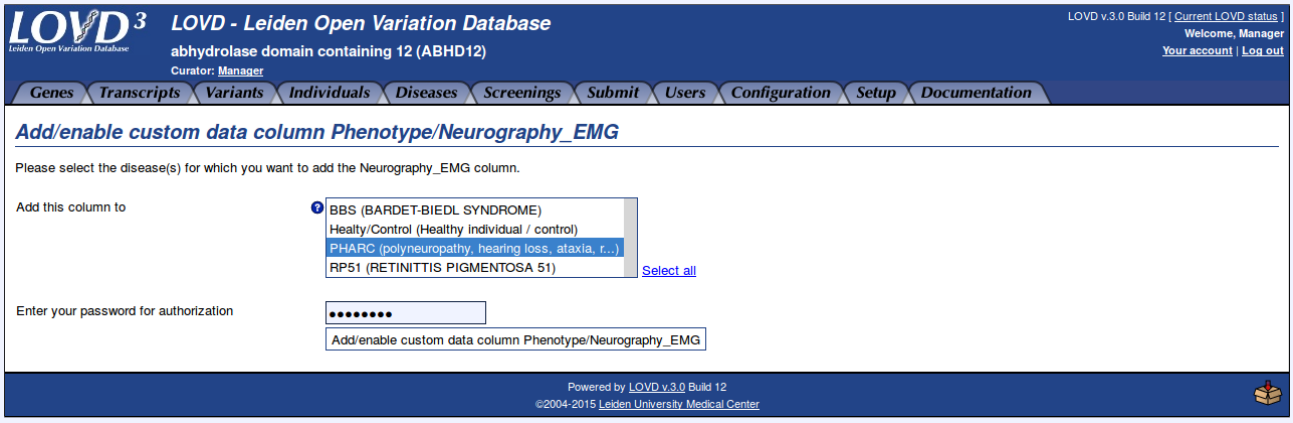

Figure 4.9: Select to which disease you want to add the custom column, in our example we select PHARC. You can select multiple diseases, see help text for how to select multiple diseases. Confirm with your password.

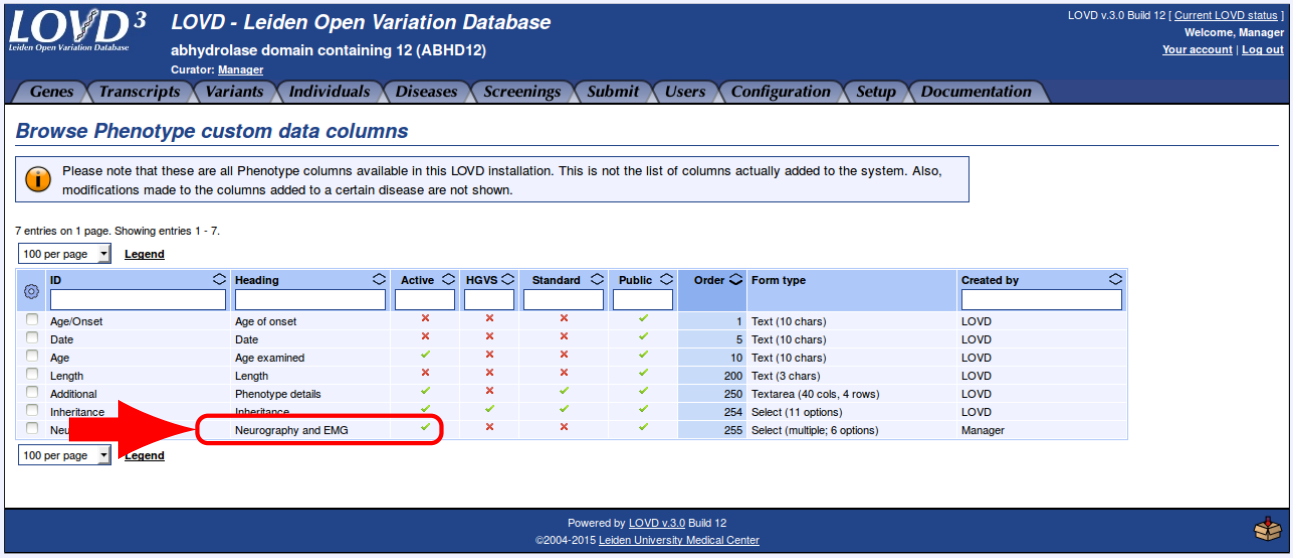

Figure 4.10: In the view "Browse custom data columns" you can see that your custom column is now Active (A).
If you want to change the settings of your custom column go to the "View custom data column" page, see figure 4.8 and click "Edit custom data column settings".

5. Creating a custom link

The objective of this chapter is:

1. Create new custom link.

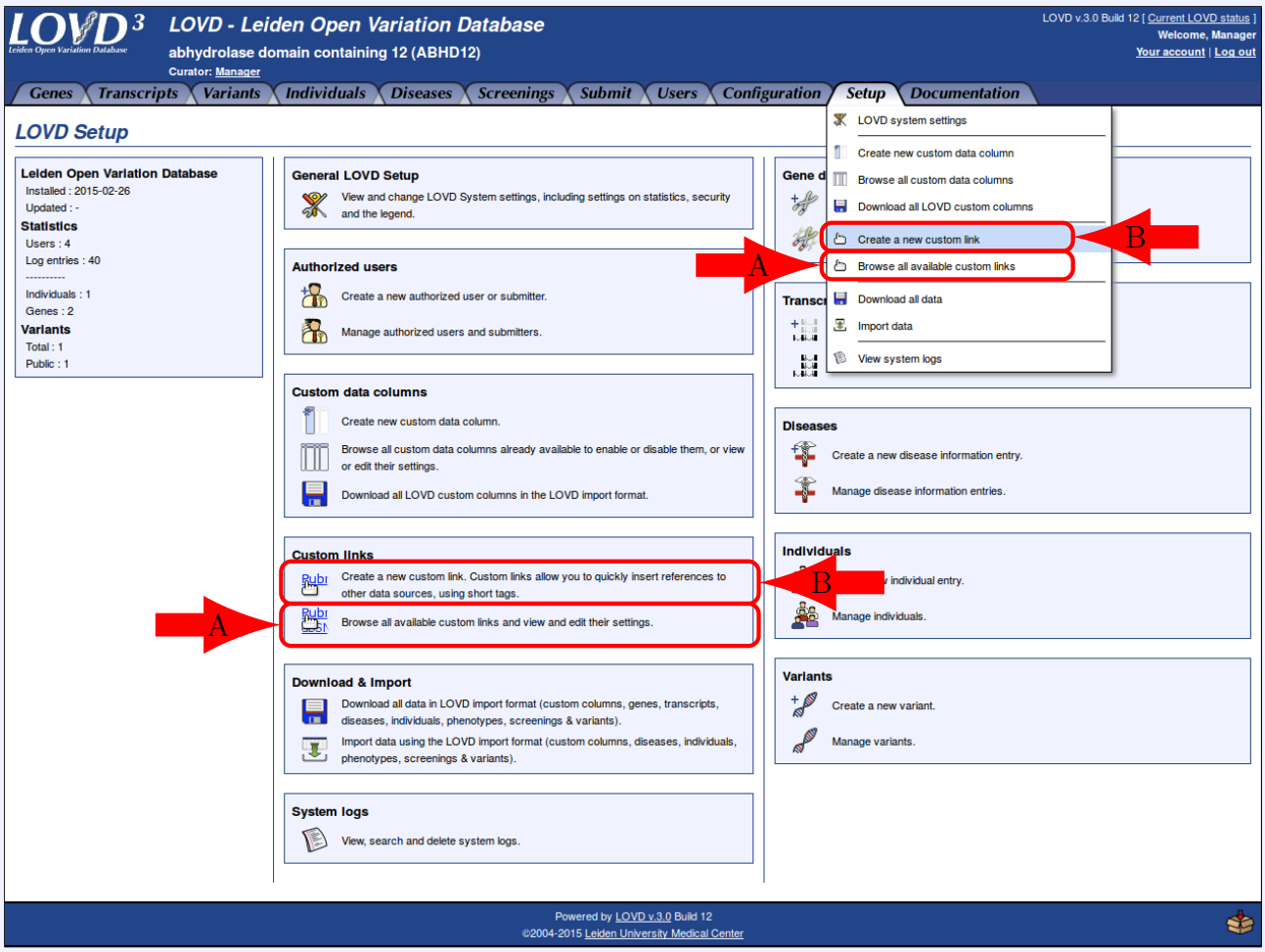

Figure 5.1: From the Setup area you can go to "Browse all available custom links" (A). LOVD has some predefined links: DbSNP, GenBank, OMIM, PubMed and DOI. For the purpose of the course we removed the DOI custom link.
Now we will create a new custom link for DOIs. Click on the "Create a new custom link" link from the Setup drop down menu tab, or select "Create a new custom link" from the Setup area (B).

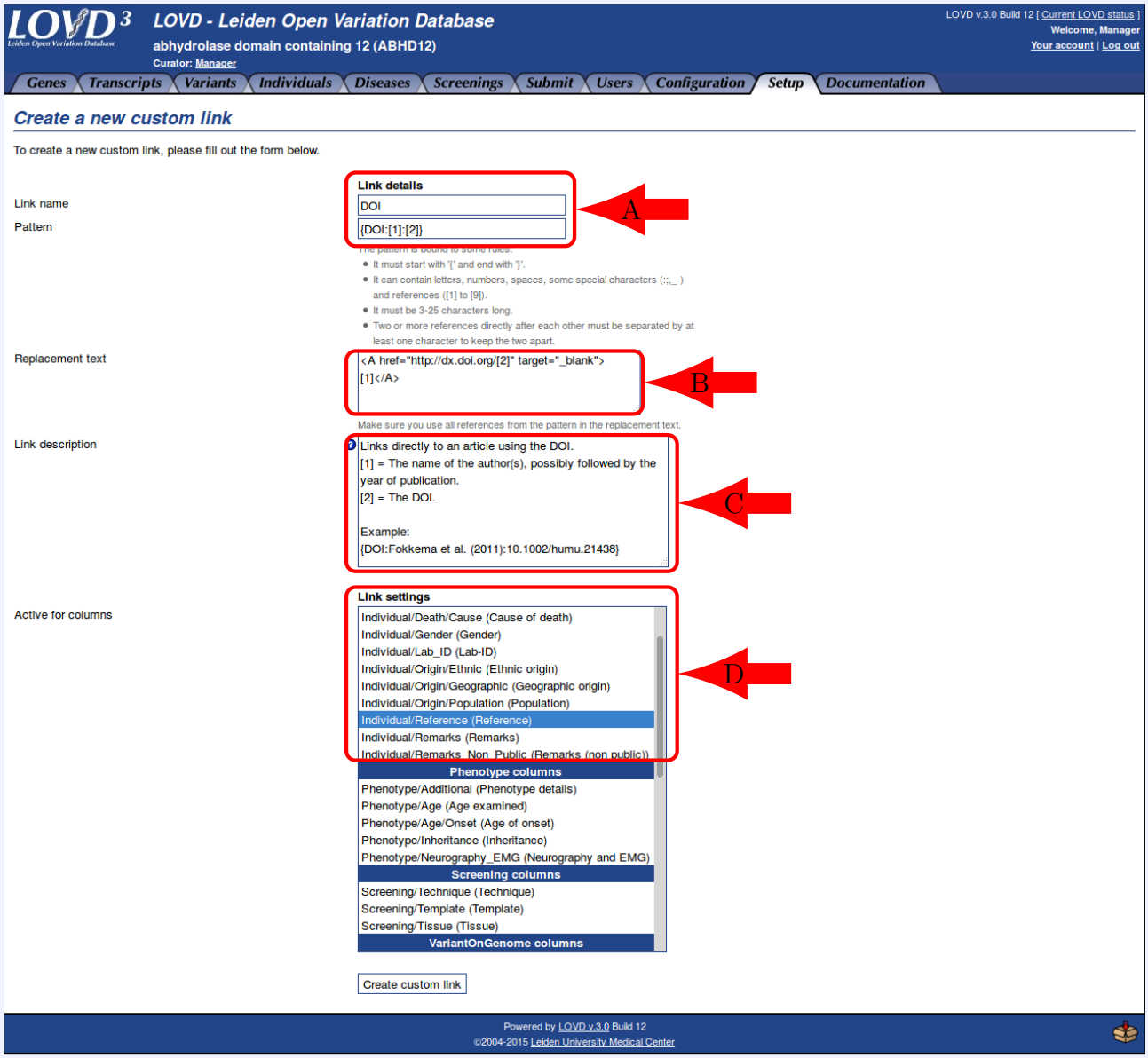

Figure 5.2: Choose a name and pattern that curators need to use for LOVD to recognize the custom link (A). Enter the (HTML enabled) text that should replace the entire pattern (B). You need to use the same number of references that you used in the pattern.
Provide a short description about this link (C). Select the columns for which you want this custom link to be activated (D).
When you are ready, click “Create custom link”.

6. Creating and managing users

The objective of this chapter is:

1. Create new user.
2. Manage user.
3. Make user curator.

6.1 Create users

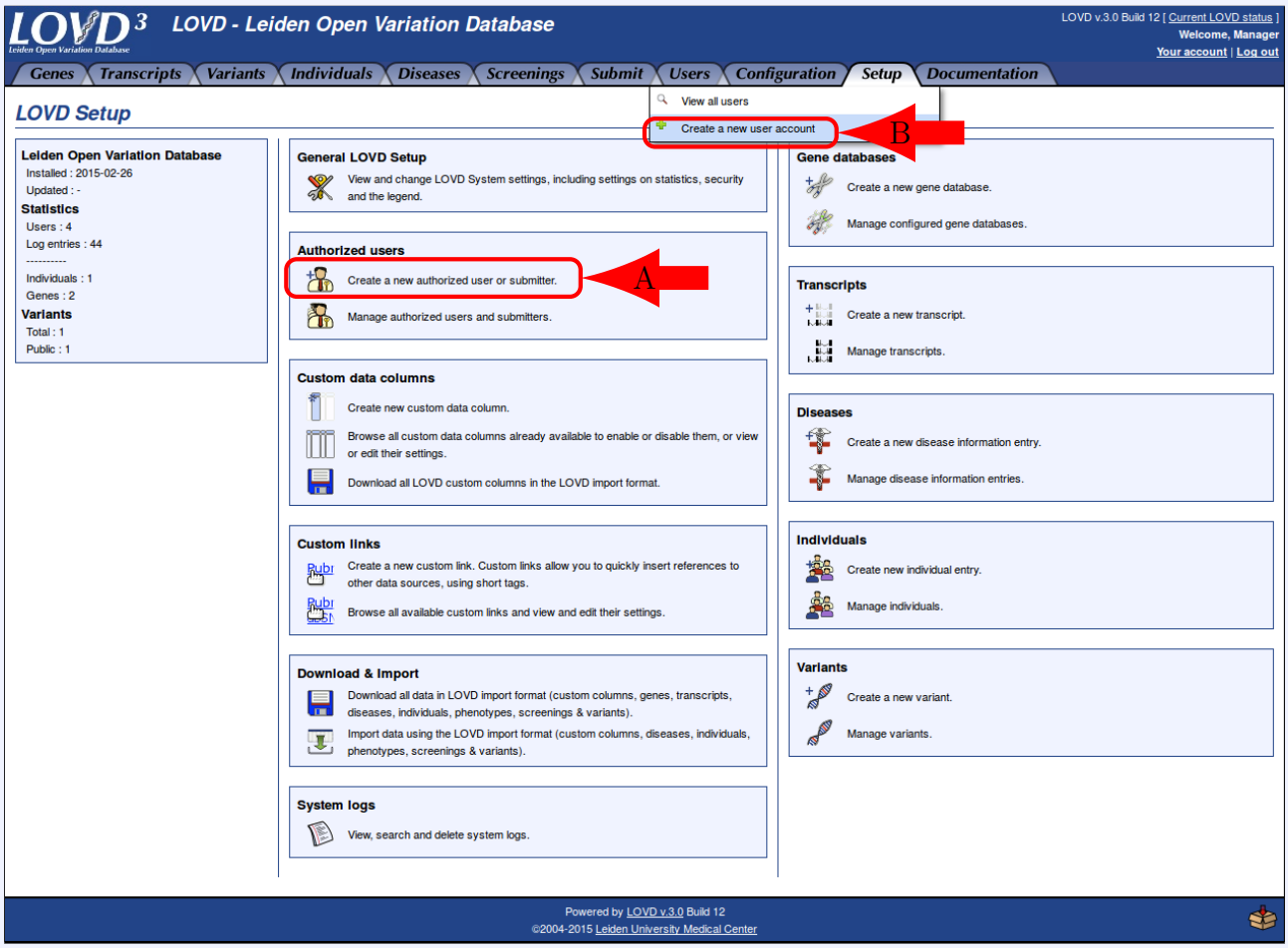

Figure 6.1: You can create a new user via "Create a new authorized user or submitter" from the Setup area (A). Alternative: Use the "Create new a user account" link from the User drop down menu tab (B).

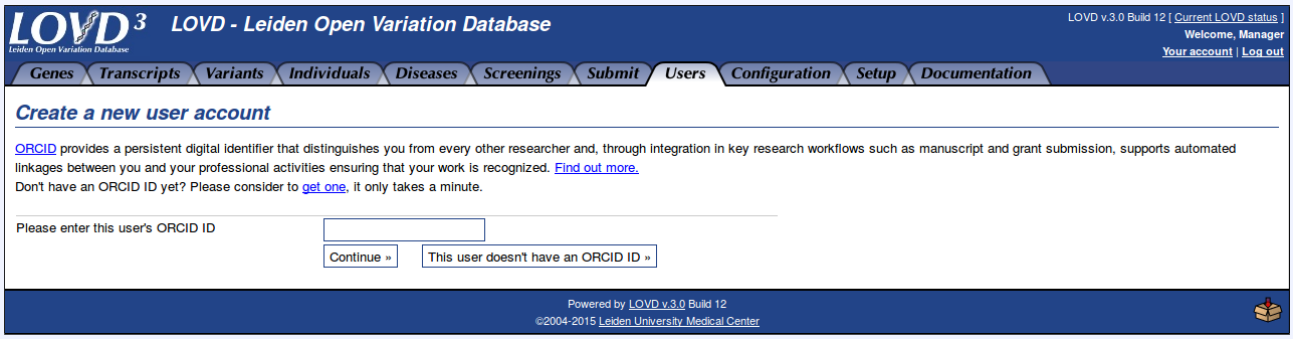

Figure 6.2: You can use individual's ORCID ID to create a new user, but this is not mandatory. Click "This user doesn't have an ORCID ID" if you do not want to use an ORCID ID.

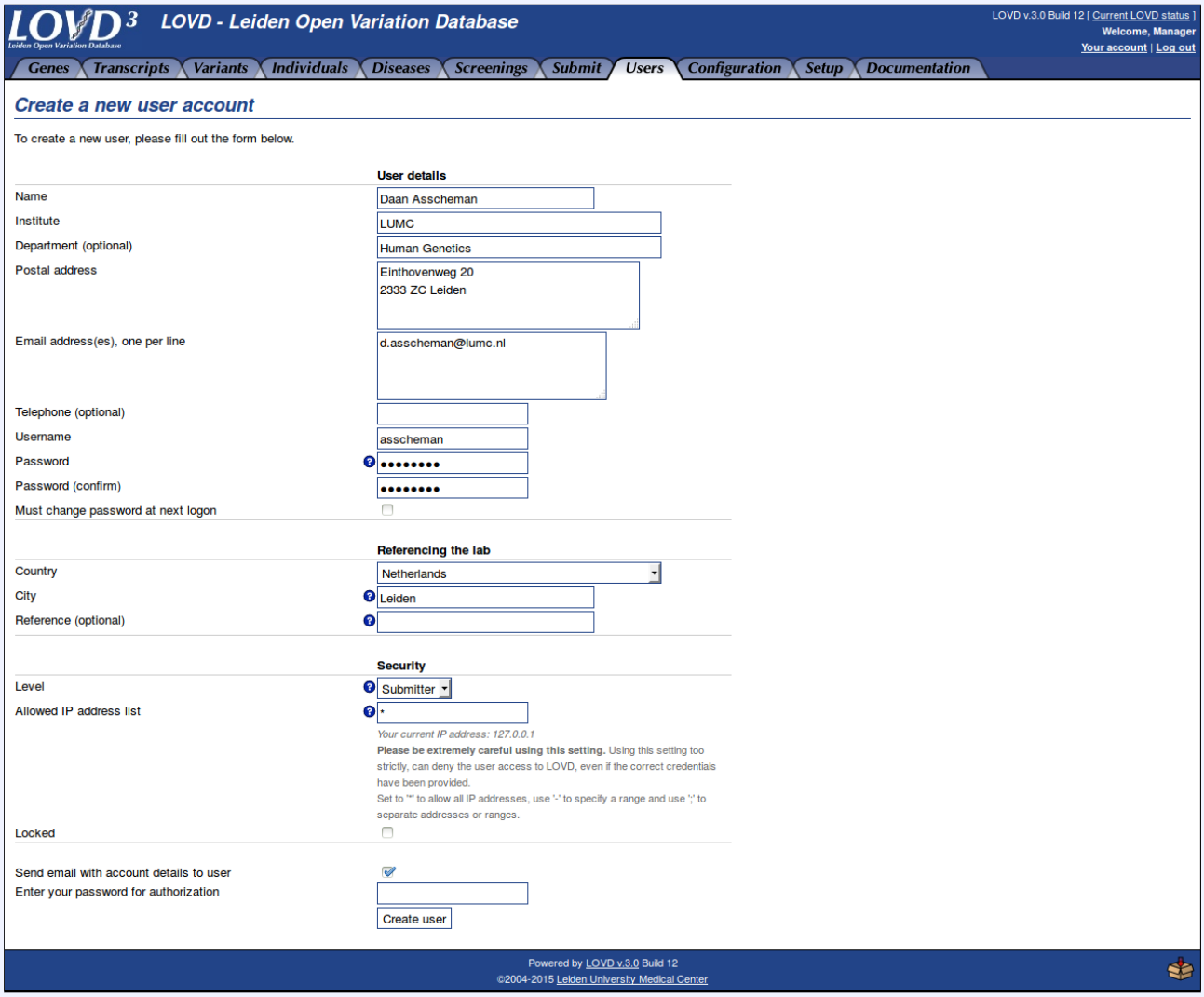

Figure 6.3: Enter the credentials of the new user and confirm with your password.

6.2 Manage users

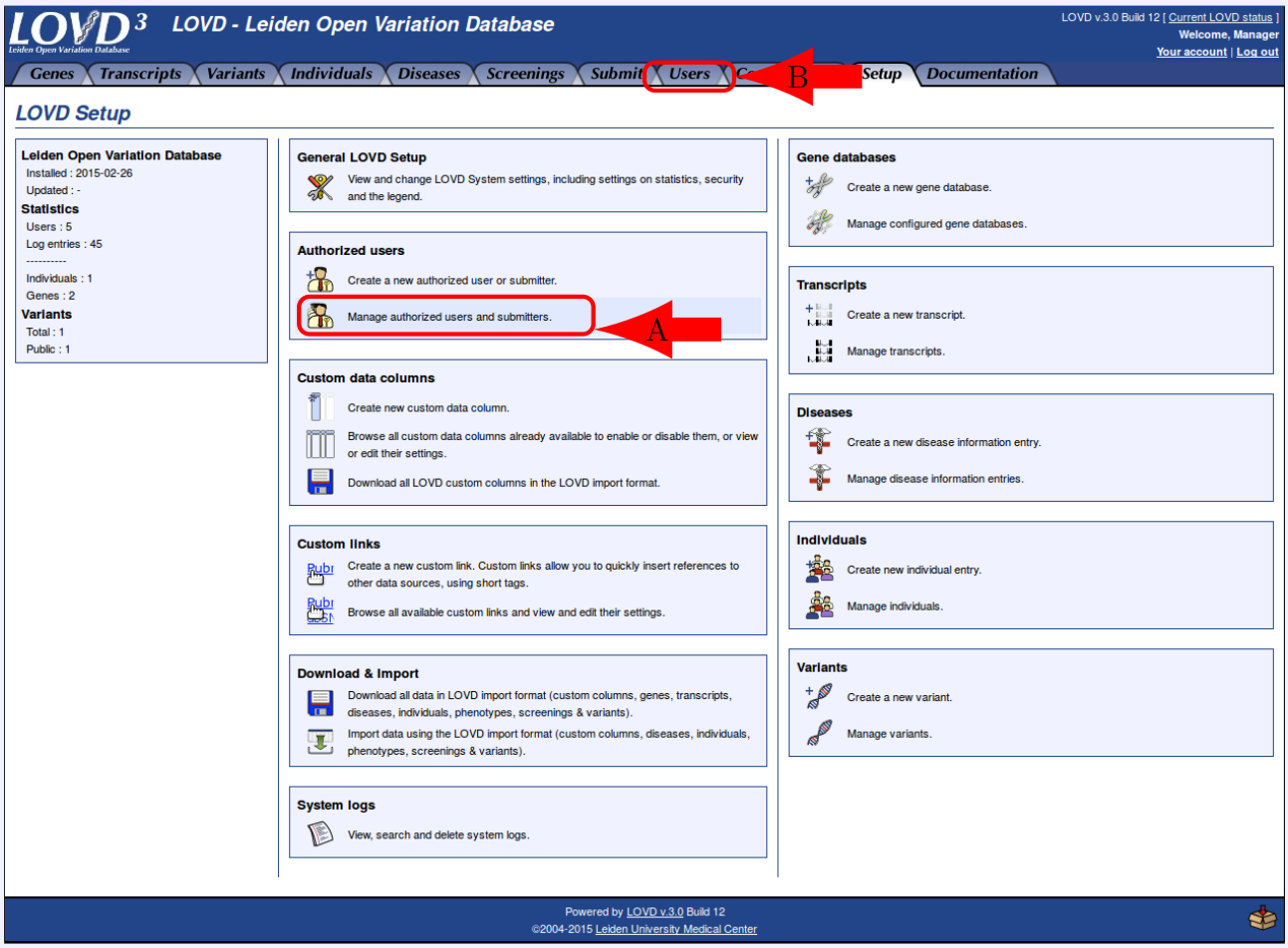

Figure 6.4: You can edit a user via "Manage authorized users and submitters" from the Setup area (A). Or click the Users menu tab (B).

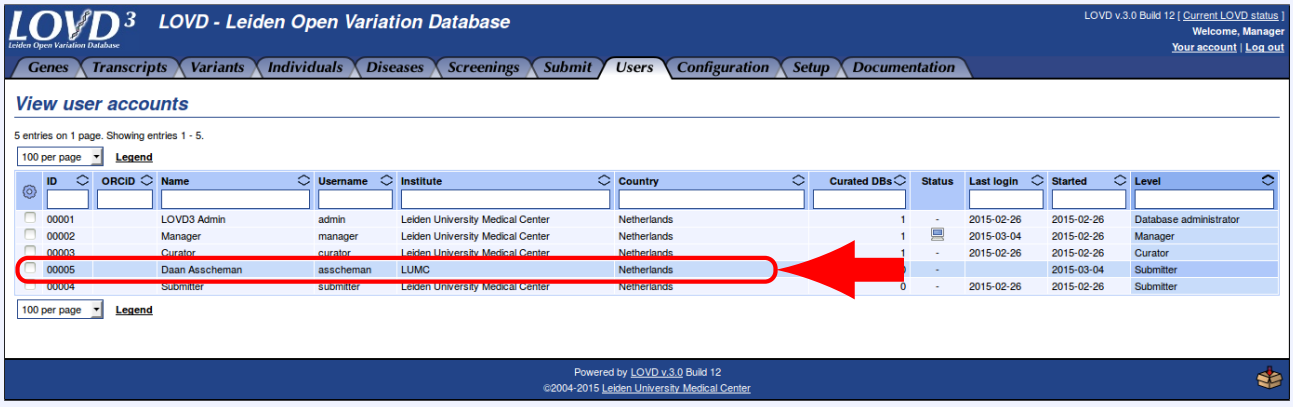

Figure 6.5: Click on the user you want to edit.

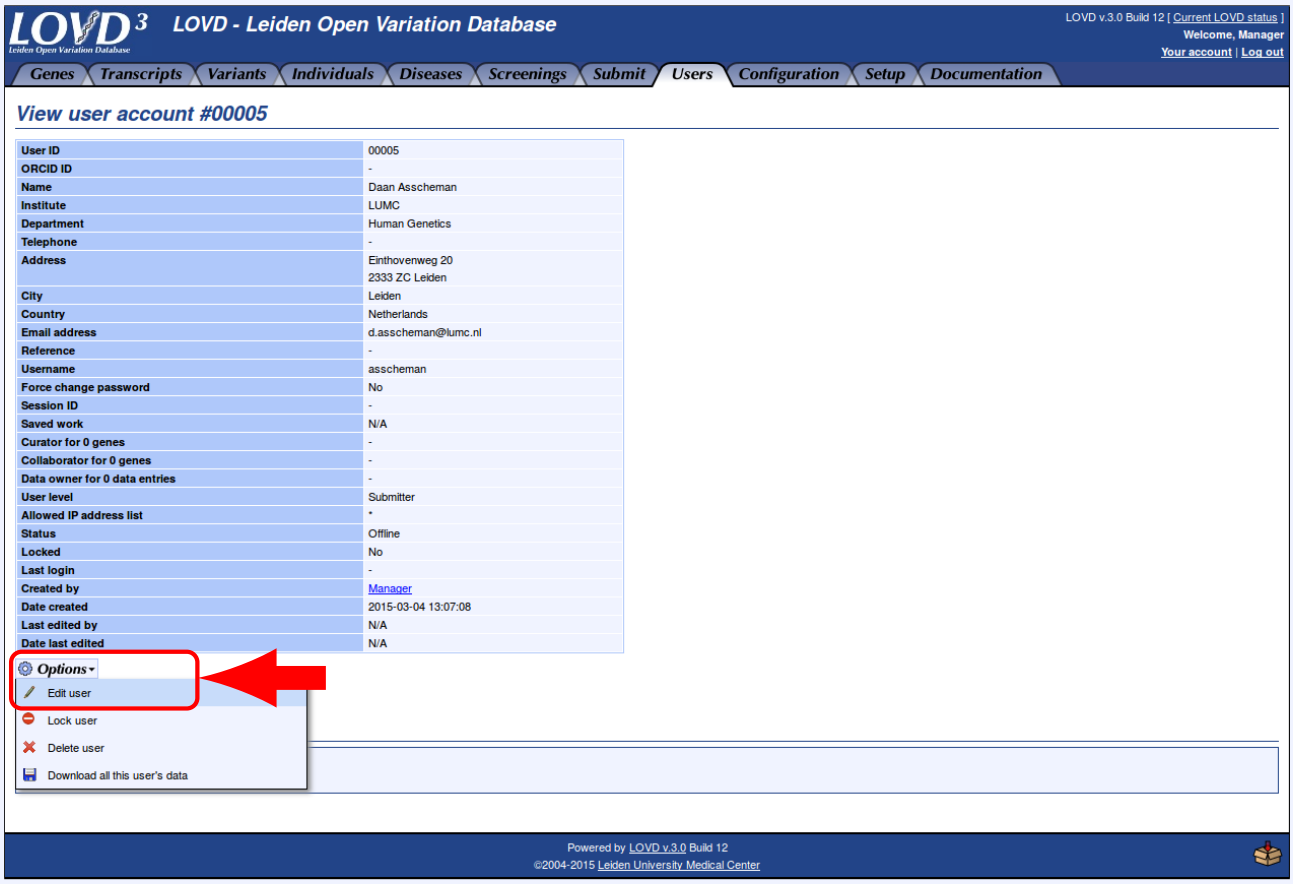

Figure 6.6: Click the Options drop down menu and select “Edit user”. You will go to the form “Edit user”, this form is similar to the form “Create user account”, see figure 6.3.

6.3 Make user curator

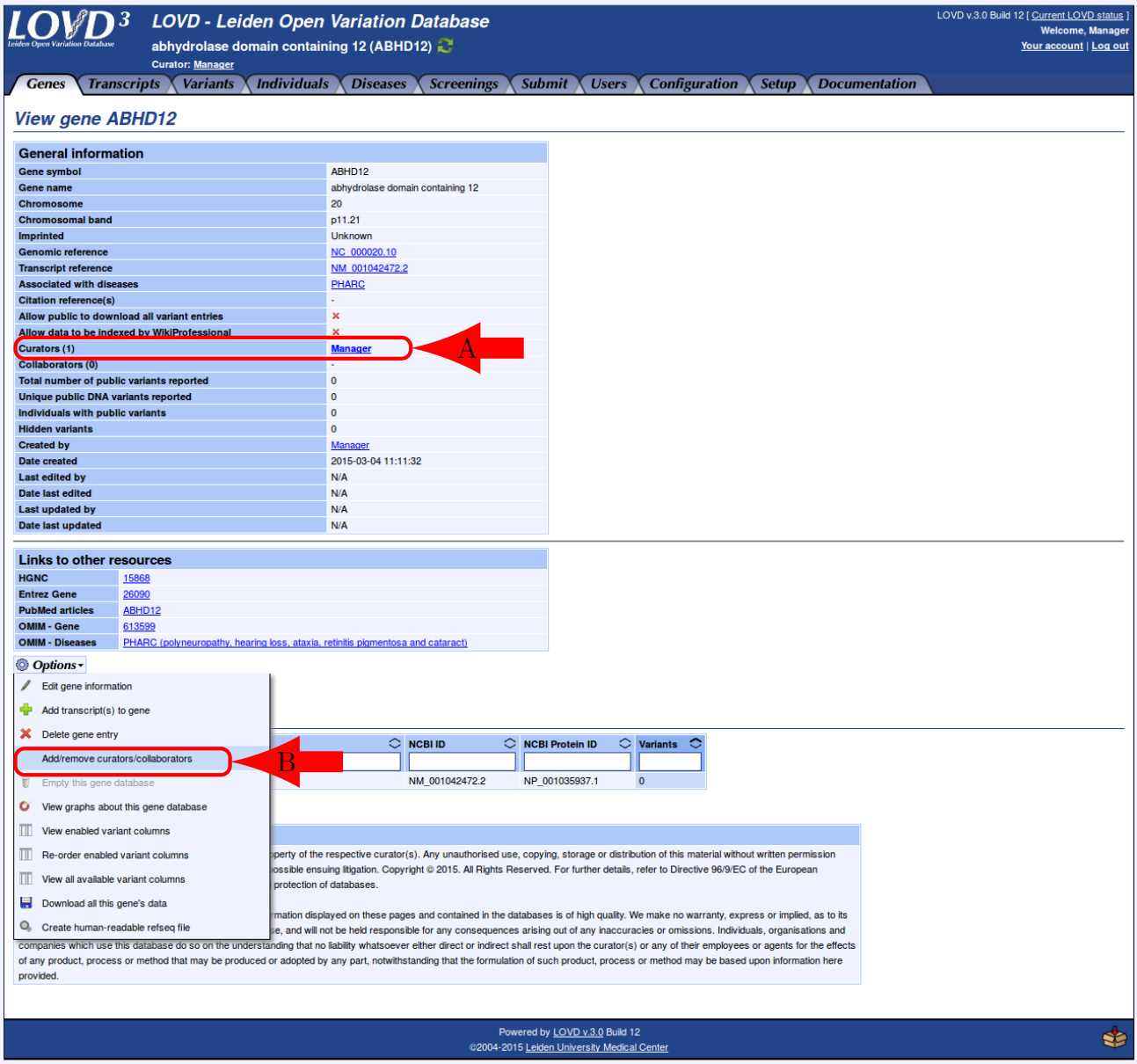

Figure 6.7: On the "View gene" page you can see who curator is for a gene (A), in our example this is only the manager.
We will make our new user curator for the ABHD12 gene. Click on "Add/remove curators/collaborators"(B).

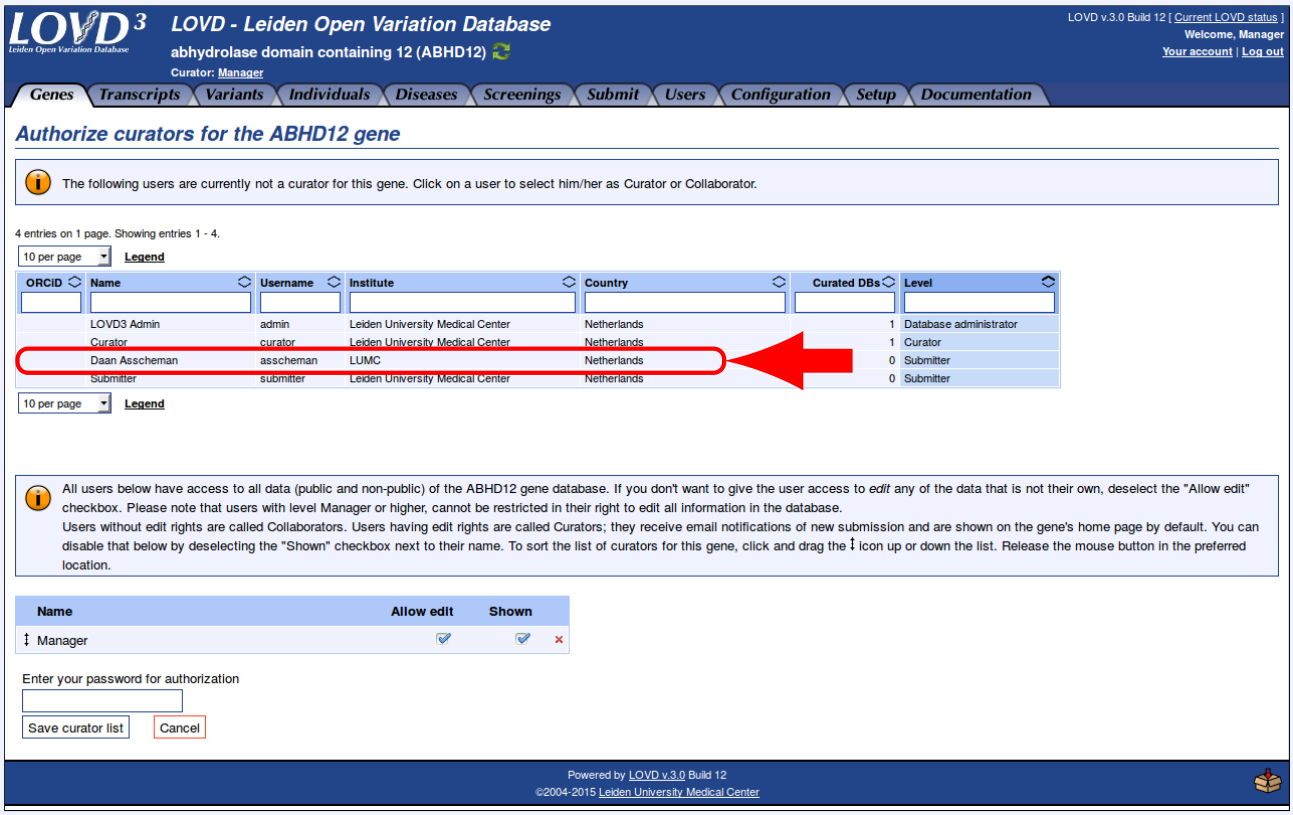

Figure 6.8: Click the user you want to add as a curator.

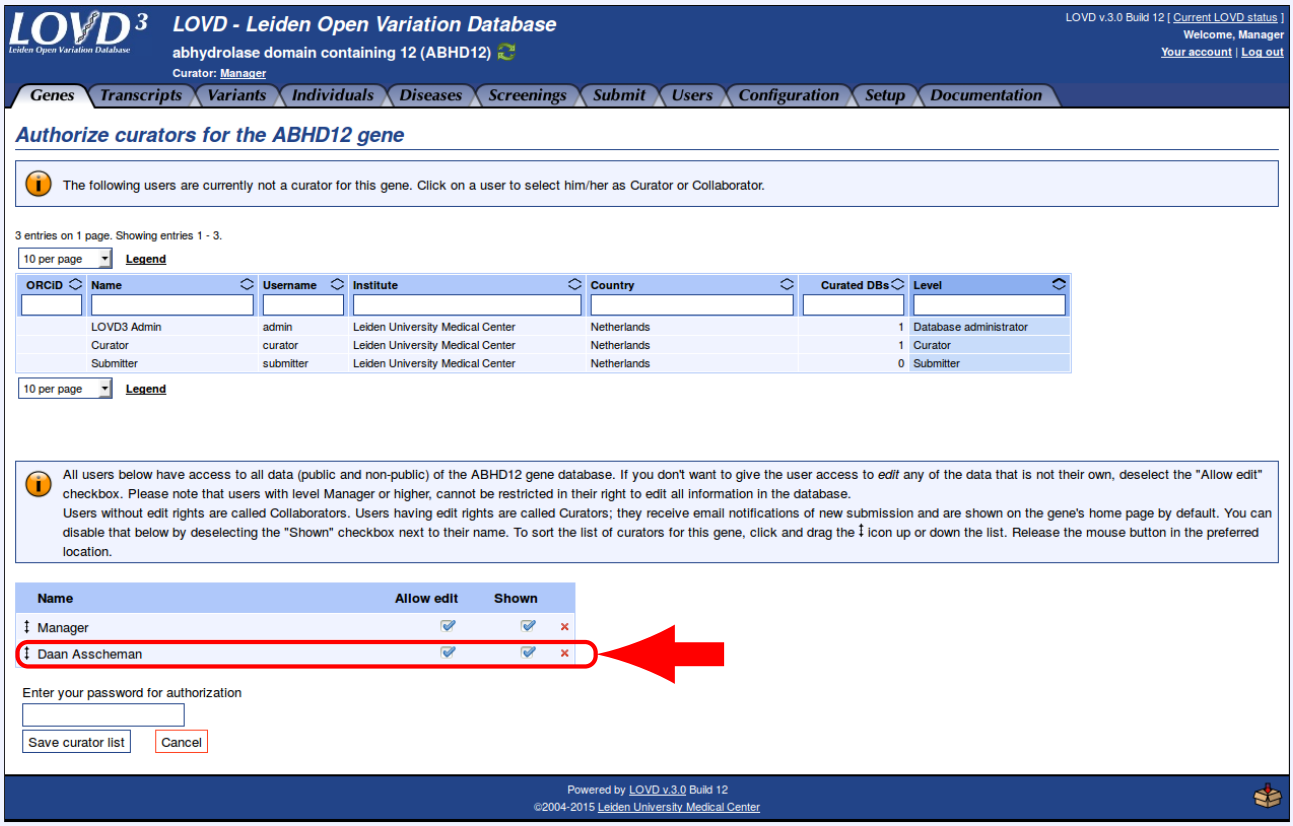

Figure 6.9: The user will appear in the list of curators. You can make the user a collaborator by unchecking the "Allow edit" field, the user can still see all public and unpublic data in this gene database, but he can't edit it, like curators can.
The "Shown" checkbox indicates whether or not the user's name and email address is shown on the gene homepage and on the top of every page while this gene is selected.
To remove an user as a curator or collaborator, click the red cross at the far right side of the table. Confirm with your password.

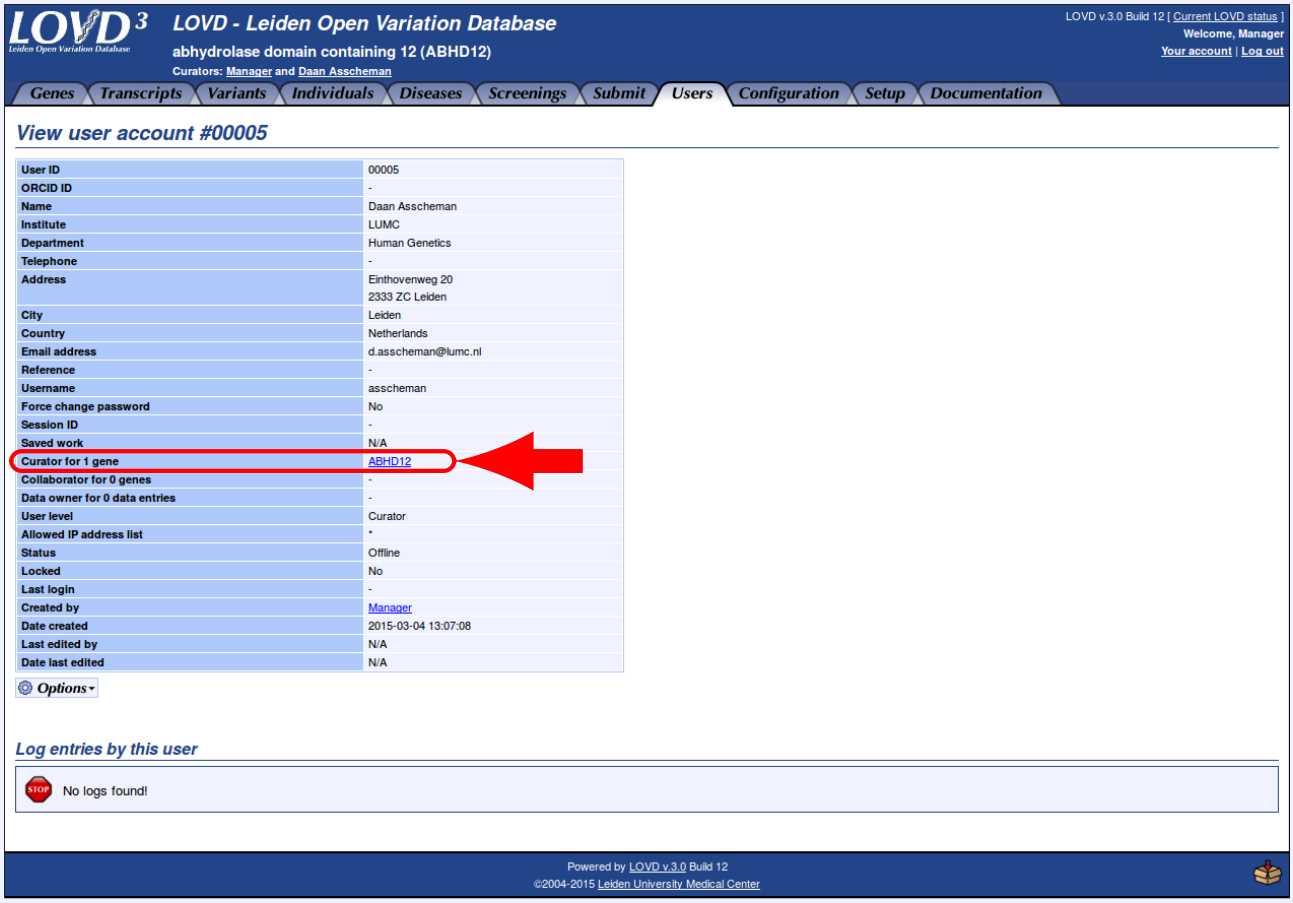

Figure 6.10: In the "View user account" you will see that the gene is added to "Curator for".

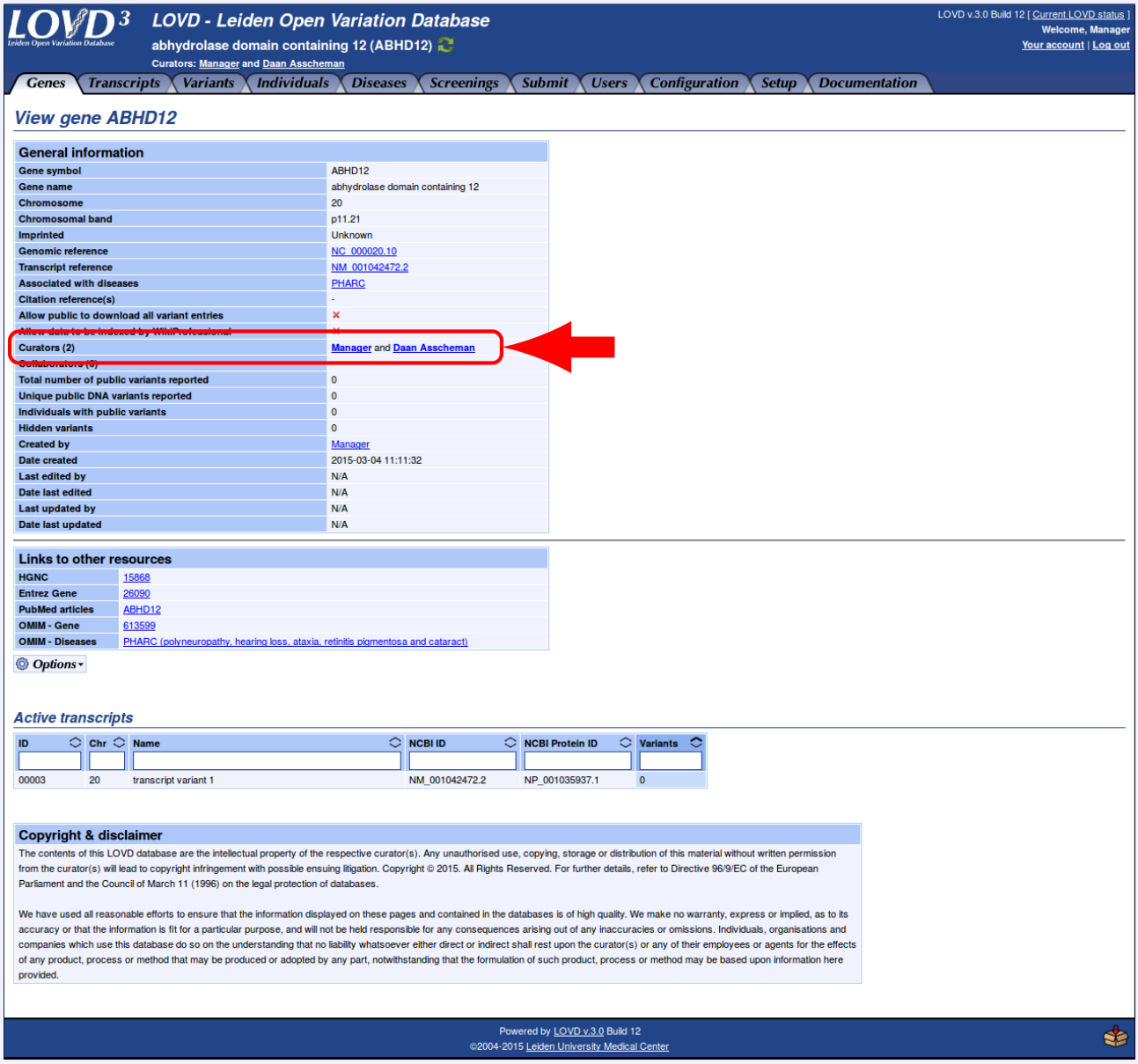

Figure 6.11: In the "View gene" you can see that the user is added to Curators.