



LOVD3 Course

Curate gene variant database (LSDB)

Build 3.0-14

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1. Editing a curator account

The objective of this chapter is:

1. Edit some basic information of the curator user account.

To start this chapter:

- Go to <http://courses.lovd.nl/LOVD3/> and select the directory corresponding to the number assigned to you.
- Log in as Manager (with for every one the same username: manager, password: manager1).

The screenshot shows the LOVD 3.0 Setup page. At the top, there is a navigation bar with tabs: Genes, Transcripts, Variants, Individuals, Diseases, Screens, Users, Configuration, and Setup. The 'Users' tab is highlighted with a red circle and arrow (C). The 'Setup' tab is also highlighted with a red circle and arrow (A). Below the navigation bar, there is a sidebar with statistics: Lieden Open Variation Database (Installed: 2015-02-18, Updated: -, Curators: LOVD3 Admin and Curator), Statistics (Users: 4, Log entries: 13, Individuals: 0, Genes: 1), and Variants (Total: 0). The main content area is titled 'LOVD Setup'. It contains several sections: 'General LOVD Setup' (View and change LOVD System settings, including settings on statistics, security and the legend), 'Authorized users' (Create a new authorized user or submitter, Manage authorized users and submitters - this link is highlighted with a red box and arrow (B)), 'Custom data columns' (Create new custom data column, Browse all custom data columns already available to enable or disable them, or view or edit their settings, Download all LOVD custom columns in the LOVD import format), 'Custom links' (Create a new custom link, Custom links allow you to quickly insert references to other data sources, using short tags, Browse all available custom links and view and edit their settings), 'Download & Import' (Download all data in LOVD import format (custom columns, genes, transcripts, diseases, individuals, phenotypes, screenings & variants), Import data using the LOVD import format (custom columns, diseases, individuals, phenotypes, screenings & variants)), 'System logs' (View, search and delete system logs), 'Gene databases' (Create a new gene database, Manage configured gene databases), 'Transcripts' (Create a new transcript, Manage transcripts), 'Diseases' (Create a new disease information entry, Manage disease information entries), 'Individuals' (Create new individual entry, Manage individuals), and 'Variants' (Create a new variant, Manage variants). At the bottom of the page, it says 'Powered by LOVD v.3.0 Build 12' and '©2004-2015 Leiden University Medical Center'.

Figure 1.1: If you want to edit a user account you have to go to “View user accounts” and select a user. You can do that from the Setup area (A), click the “Manage authorized users and submitters.” link listed under “Authorized users”(B).

Alternative: Go directly to “View users accounts” via the Users menu tab (C).

1. Editing a curator account

LOVD - Leiden Open Variation Database
tetrastricopeptide repeat domain 8 (TTC8)
Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Users Configuration Setup Documentation

View user accounts

4 entries on 1 page. Showing entries 1 - 4.

ID	ORCID	Name	Username	Institute	Country	Curated DBs	Status	Last login	Started	Level
00001	LOVD3 Admin	admin	admin	Leiden University Medical Center	Netherlands	1	-	2015-02-18	2015-02-18	Database administrator
00002	Manager	manager	manager	Leiden University Medical Center	Netherlands	0	-	2015-02-18	2015-02-18	Manager
00003	Curator	curator	curator	Leiden University Medical Center	Netherlands	1	-	2015-02-18	2015-02-18	Curator
00004	Submitter	submitter	submitter	Leiden University Medical Center	Netherlands	0	-	2015-02-18	2015-02-18	Submitter

100 per page Legend

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Figure 1.2: Select the user you want to edit, in this example, the curator.

LOVD - Leiden Open Variation Database
tetrastricopeptide repeat domain 8 (TTC8)
Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Users Configuration Setup Documentation

View user account #00003

User ID	00003
ORCID ID	-
Name	Curator
Institute	Leiden University Medical Center
Department	Human Genetics
Telephone	-
Address	Einthovenweg 20 2333 ZC Leiden
City	Leiden
Country	Netherlands
Email address	curator@LOVD.nl
Reference	-
Username	curator
Force change password	No
Session ID	-
Saved work	N/A
Curator for 1 gene	TTC8
Collaborator for 0 genes	-
Data owner for 0 data entries	-
User level	Curator
Allowed IP address list	-
Status	Offline
Locked	No
Last login	-
Created by	LOVD3 Admin
Date created	2015-02-18 10:06:54
Last edited by	N/A
Date last edited	N/A

Options

- Edit user
- Lock user
- Delete user
- Download all this user's data

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Figure 1.3: Click the Options drop down menu and select “Edit user”.

1. Editing a curator account

The screenshot shows the 'Edit user account #00003' page. The top navigation bar includes links for Genes, Transcripts, Variants, Individuals, Diseases, Screenings, Submit, Users, Configuration, Setup, Documentation, Welcome, Manager, Your account, and Log out. Red annotations are present: A points to the 'Name' field containing 'Curator'; B points to the 'Email address(es)' field containing 'curator@LOVD.nl'; C points to the password field which has a large red 'X' over it; D points to the 'Edit user' button at the bottom; and E points to the 'Log out' link in the top right corner.

User details

Name: Curator (A)

Institute: Leiden University Medical Center

Department (optional): Human Genetics

Postal address: Eindhovenweg 20
2333 ZC Leiden

Email address(es), one per line: curator@LOVD.nl (B)

Telephone (optional): ~~1234567890~~ (C)

New password (optional):

New password (confirm, optional):

Must change password at next logon:

Referencing the lab

Country: Netherlands

City: Leiden (D)

Reference (optional):

Security

Level: Submitter (E)

Allowed IP address list:

Your current IP address: 127.0.0.1

Please be extremely careful using this setting. Using this setting too strictly, can deny the user access to LOVD, even if the correct credentials have been provided.

Set to "*" to allow all IP addresses, use "-" to specify a range and use ";" to separate addresses or ranges.

Locked:

Enter your password for authorization: (D)

Buttons

Edit user (D)

Log out (E)

Figure 1.4: Insert your name, in place of “Curator” (A).

Change the e-mail address to your address (so you can receive notifications of new submissions) (B). For the course: **do not change the password** (C).

Confirm changes with the Manager password, submit the form (D) and log out (E).

2. Editing a gene database

The objectives of this chapter are:

1. Inspect form “Edit gene information entry”.
2. Create and add a disease to a gene via the “Edit gene information entry” form.

To start this chapter:

- Log in as Curator (username: curator, password: curator1).

LOVD 3 LOVD - Leiden Open Variation Database
Curators: LOVD3 Admin and Curator

LOVD v.3.0 Build 12 | Current LOVD status | Welcome, Curator | Your account | Log out

Genes A Variants Individuals Diseases Screenings Submit Configuration Documentation

View all genes

1 entry on 1 page. Showing entry 1.
100 per page

Symbol	Gene	Chr	Band	Transcripts	Variants	Unique variants	Last updated	Associated with diseases
TTC8	tetrafunctional repeat domain 8			2	0	0	N/A	-

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Figure 2.1: Via the Genes menu tab (A), you can go to the “View gene” page. If in the header no gene is selected (B), then you are directed to “View all genes”. Click on a gene (C) to go to the “View gene” page to see details of a gene. If a gene is selected in the header and you click the Genes menu tab, you are directed to the “View gene” page, see figure 2.2.

2. Editing a gene database

The screenshot shows the LOVD 3 interface. At the top, the header includes the LOVD logo, the title "LOVD - Leiden Open Variation Database", and a search bar containing "tetrapeptide repeat domain 8 (TTC8)". A red arrow labeled "A" points to the search bar. Below the header is a navigation menu with tabs: "Genes" (highlighted with a red box and arrow "B"), "Variants", "Individuals", "Diseases", "Screenings", "Submit", "Configuration", and "Documentation". The main content area is titled "View gene TTC8". It contains two main sections: "General information" and "Links to other resources". The "General information" section lists various details about the gene, such as Gene symbol (TTC8), Gene name (tetrapeptide repeat domain 8), Chromosome (14), Chromosomal band (q31.3), and Genomic reference (NC_000014.8). The "Links to other resources" section provides links to external databases like HGNC, Entrez Gene, PubMed, OMIM, HGMD, GeneCards, and GeneTests. On the left side of the main content area, there is a sidebar with a "Options" dropdown menu. One item in the dropdown, "Edit gene information", is highlighted with a red box and arrow "C". Below the sidebar is a table showing NCBI ID, NCBI Protein ID, and Variants for two entries: NM_144596.2 (NP_653197.2, 0 variants) and NM_198309.2 (NP_938051.1, 0 variants). At the bottom of the page, there is a disclaimer about the use of the data and copyright information. The footer includes the text "Powered by LOVD v.3.0 Build 12" and "©2004-2015 Leiden University Medical Center".

Figure 2.2: If a gene is selected in the header (A) and you click the Genes menu tab (B), you are directed to the “View gene” page. On the “View gene” page, click the Options drop down menu and select “Edit gene information” (C).

2. Editing a gene database

LOVD³ LOVD - Leiden Open Variation Database
Leiden Open Variation Database
Curators: LOVD Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

Edit gene information entry

To edit this gene database, please complete the form below and press "Edit" at the bottom of the form.

General Information	
Full gene name	tetratricopeptide repeat domain 8 (TTC8)
Official gene symbol	TTC8
Chromosome	14
Chromosomal band	q31.3
Imprinting	Unknown
Date of creation (optional)	2015-02-26
Relation to diseases (optional)	
This gene has been linked to these diseases	
<input checked="" type="radio"/> No disease entries available <small>Diseases not in this list are not yet configured in this LOVD instance.</small>	
Do you want to configure more diseases?	
Reference sequences (mandatory) <small>Selecting variants requires a proper reference sequence. Without a genomic and a transcript reference sequence the variants in this LOVD database cannot be interpreted properly or mapped to the genome.</small>	
Genomic reference sequence	
NC_000014.8	
<small>Select the genomic reference sequence (NG, NC, LRG accession number). Only the references that are available to LOVD are shown.</small>	
Transcriptomic reference sequence(s)	
<small>To add, remove or edit transcriptomic reference sequences for this gene, please see the gene's detailed view.</small>	
Links to information sources (optional)	
Homepage URL	
<small>If you have a separate homepage about this gene, you can specify the URL here. Format: complete URL, including "http://".</small>	
External links	
<small>Here you can provide links to other resources on the internet that you would like to link to. One link per line, format: complete URLs or "Description <URL>".</small>	
HGNC ID	
Entrez Gene (Locuslink) ID	
OMIM Gene ID	
<small>Provide link to HGMD</small>	
<small>Provide link to GeneCards</small>	
<small>Provide link to GeneTests</small>	
This gene has a human-readable reference sequence	
<small>Although GenBank files are the official reference sequence, they are not very readable for humans. If you have a human-readable format of your reference sequence online, please select the type here.</small>	
Human-readable reference sequence location	
<small>If you used our Reference Sequence Parser to create a human-readable reference sequence, the result is located at "https://localhost:8080/LOVD3_training_01/trunk/src/refseq/TTC8_codingDNA.html".</small>	
Customizations (optional)	
Citation reference(s)	
<small>You can use the following fields to customize the gene's LOVD gene homepage.</small>	
<small>(Active custom link : PubMed)</small>	
<small>Use standard LOVD disclaimer</small>	
<small>If you want a disclaimer added to the gene's LOVD gene homepage, select your preferred option here.</small>	
Text for own disclaimer (HTML enabled)	
<small>Only applicable if you choose to use your own disclaimer (see option above).</small>	
Page header (HTML enabled)	
<small>Text entered here will appear above all public gene-specific pages.</small>	
<small>Header aligned to</small>	
<small>Page footer (HTML enabled)</small>	
<small>Footer aligned to</small>	
<small>Notes for the LOVD gene homepage (HTML enabled)</small>	
<small>Notes for the variant listings (HTML enabled)</small>	
<small>Text entered here will appear below the gene's variant listings.</small>	
Security settings	
<small>Using the following settings you can control some security settings of LOVD.</small>	
<input type="checkbox"/> Allow public to download variant entries <input type="checkbox"/> Allow my public variant and individual data to be indexed by Wikipedia	

[Edit gene information entry](#)

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Figure 2.3: Look around and add or change things if you wish. This gene does not have a relation to disease, yet. As an example we will add a related disease to this gene.

There are two ways to add a disease and link this disease to a gene. The first method is demonstrated below; the second method is explained in chapter “[Creating a disease](#)”. Click “configure more diseases” to create a new disease (A).

2. Editing a gene database

Create a new disease information entry

To create a new disease information entry, please fill out the form below.

Disease abbreviation
Disease name
OMIM ID (optional)

This disease has been linked to these genes

Disease Information
BBS
BARDET-BIEDL SYNDROME
209900

Relation to genes (optional)
TTC8 (tetratricopeptide repeat domain 8)

Create disease information entry

A
B
C

Figure 2.4: To create a new disease, fill in the disease abbreviation, name and OMIM ID fields (A).

Note that the OMIM ID field is an unique field. Therefore you can not use the same OMIM ID with different disease names.

To make a relation between this new disease select a gene, in our example TTC8 (B). When you are curator of more genes, you can select a range of genes.

When you are ready, click “Create disease information entry” (C).

LOVD 3 LOVD - Leiden Open Variation Database tetratricopeptide repeat domain 8 (TTC8) Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Users Configuration Setup Documentation

Edit gene information entry

To edit this gene database, please complete the form below and press "Edit" at the bottom of the form.

General Information

Full gene name: tetratricopeptide repeat domain 8
Official gene symbol: TTC8
Chromosome: 14
Chromosomal band: q31.3
Imprinting: Unknown
Date of creation (optional): 2015-02-16

Relation to diseases (optional)

This gene has been linked to these diseases: BBS (BARDET-BIEDL SYNDROME)

Security settings
Using the following settings you can control some security settings of LOVD.

Allow public to download variant entries:
Allow my public variant and individual data to be indexed by WikiProfessional:

Edit gene information entry

Powered by LOVD v.3.0 Build 12 | Current LOVD status | Welcome, Manager | Your account | Log out

Figure 2.5: Gene TTC8 now has a relation to disease Bardet-Biedl Syndrome (A).

Note that when you have more diseases in your installation, the diseases are ordered by abbreviation. If a diseases entry has no abbreviation, it is listed below the disease with abbreviation.

When you have finished modifying the gene information, click “Edit gene information entry” to save the changes (B).

3. Creating a disease

The objective of this chapter is:

1. Create disease information and add a disease to a gene via the Disease menu tab.

The screenshot shows the LOVD3 interface with the 'Diseases' tab selected. A red arrow points to the 'Create a new disease information entry' option in the dropdown menu. The main content area displays a table of diseases, including BARDET-BIEDL SYNDROME (ID: 00001, Abbreviation: BBS). The bottom of the page includes a footer with copyright information and a logo.

Figure 3.1: Create a disease via “Create a new disease information entry” on the Diseases menu tab drop down menu.

The screenshot shows the 'Create a new disease information entry' form. Red boxes highlight three key areas: (A) the 'Disease Information' section containing fields for abbreviation (RP51), name (RETINITIS PIGMENTOSA 51), and OMIM ID (613464); (B) the 'Relation to genes (optional)' section where 'TTC8 (tetratricopeptide repeat domain 8)' is selected; and (C) the 'Create disease information entry' button at the bottom. Red arrows point from the labels A, B, and C to their respective highlighted areas.

Figure 3.2: To create a disease, fill in the disease abbreviation, name and OMIM ID fields (A).

To make a relation to this new disease, select a gene, in our example TTC8 (B). When you are curator of more genes, you can select a range of genes.

When you are ready, click “Create disease information entry”(C).

3. Creating a disease

The screenshot shows the LOVD 3 - Leiden Open Variation Database interface. At the top, there is a navigation bar with tabs for Genes, Transcripts, Variants, Individuals, Diseases (which is highlighted with a red box and arrow A), Submit, Users, Configuration, Setup, and Documentation. To the right of the tabs, there is a status bar with "LOVD v.3.0 Build 12 [Current LOVD status]", "Welcome, Manager", "Your account | Log out", and a small logo.

The main content area is titled "View diseases" and displays a table of two entries. The table has columns for ID, Abbreviation, Name, OMIM ID, Individuals, Phenotypes, and Associated with genes. The first entry is BBS (Barber-Biedl Syndrome) with ID 00001, Abbreviation BBS, Name BARDET-BIEDL SYNDROME, OMIM ID 209900, Individuals 0, Phenotypes 0, and Associated with genes TTC8. The second entry is RP51 (Retinitis Pigmentosa 51) with ID 00002, Abbreviation RP51, Name RETINITIS PIGMENTOSA 51, OMIM ID 613464, Individuals 0, Phenotypes 0, and Associated with genes TTC8. Both entries have a red box around the "Associated with genes" column, and a red arrow B points to the TTC8 entry in the second row.

ID	Abbreviation	Name	OMIM ID	Individuals	Phenotypes	Associated with genes
00001	BBS	BARDET-BIEDL SYNDROME	209900	0	0	TTC8
00002	RP51	RETINITIS PIGMENTOSA 51	613464	0	0	TTC8

At the bottom of the page, there is a footer with "Powered by LOVD v.3.0 Build 12" and "©2004-2015 Leiden University Medical Center".

Figure 3.3: We now have created two diseases, one in chapter “[Editing a gene database](#)” and one in this chapter. To see them click the Diseases menu tab (A). You can see both diseases are associated with the TTC8 gene(B).

4. Reference sequences and the reference sequence parser

The objective of this chapter is:

1. Create an HTML page of the DNA reference sequence with exon/intron boundaries, upstream and downstream and intronic sequences.

To start this chapter:

- Log in as Curator (with for every one the same username: curator, password: curator1).

4. Reference sequences and the reference sequence parser

LOVD 3 - LOVD - Leiden Open Variation Database
Leiden Open Variation Database
tetrapeptide repeat domain 8 (TTC8)

Curators: LOVD3 Admin and Curator

Genes A Variants Individuals Diseases Screenings Submit Configuration Documentation

View gene TTC8

General information

Gene symbol	TTC8
Gene name	tetrapeptide repeat domain 8
Chromosome	14
Chromosomal band	q31.3
Imprinted	Unknown
Genomic reference	NC_000014.8
Transcript reference	NM_144596.2, NM_198309.2
Associated with diseases	BBS, RP51
Citation reference(s)	-
Allow public to download all variant entries	X
Allow data to be indexed by WikiProfessional	X
Curators (2)	LOVD3 Admin and Curator
Collaborators (0)	-
Total number of public variants reported	0
Unique public DNA variants reported	0
Individuals with public variants	0
Hidden variants	0
Created by	LOVD3 Admin
Date created	2015-02-26
Last edited by	Curator
Date last edited	2015-02-26 10:19:33
Last updated by	N/A
Date last updated	N/A

Links to other resources

HGNC	20087
Entrez Gene	123016
PubMed articles	TTC8
OMIM - Gene	608132
OMIM - Diseases	BBS (BARDET-BIEDL SYNDROME) RP51 (RETINITIS PIGMENTOSA 51)
HGMD	TTC8
GeneCards	TTC8
GeneTests	TTC8

Options

- Edit gene information
- Add transcript(s) to gene
- Sort/hide curator names
- Empty this gene database
- View graphs about this gene database
- View enabled variant columns
- Re-order enabled variant columns
- View all available variant columns
- Download all this gene's data
- Create human-readable refseq file B

Property of the respective curator(s). Any unauthorised use, copying, storage or distribution of this material without written permission is possible only on a legal basis. Copyright © 2015. All Rights Reserved. For further details, refer to Directive 96/9/EC of the European Union.

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Figure 4.1: Go to the Genes menu tab (A). If in the header no gene is selected, you are directed to “View all genes”. You have to select a gene first, see figure 2.1. On the “View gene” page, click the Options drop down menu and select “Create human-readable refseq file” (B). A popup screen will appear.

Step 1 - Import annotated Genbank sequence to extract genomic sequence

Select gene and transcript	TTC8 (NM_144596.2 -> NP_653197.2)
Genomic sequence to use	NG (gene-specific) when available, NC (chromosomal) otherwise
<input type="button" value="Continue"/>	

Figure 4.2: Select gene and transcript and which sequence you want to use. In our example choose “TTC8(NM_144596.2 -> NP_653197.2)” and “NG (gene-specific) when available, NC (chromosomal) otherwise”. Continue when ready.

Step 3 - Create coding DNA reference sequence

(All fields are mandatory unless specified otherwise)

Notes above sequence
(optional, HTML enabled)
 <p>This file was created to facilitate the description of sequence variants
on transcript NM_144596.2 in the TTC8 gene based on a coding DNA
reference sequence following <a href="http://www.HGVS.org/mutnomen...</p>

Include link to GenBank record in notes above
sequence (optional)
 NC_000014.8

If you fill in a GenBank accession.version number, a link to the record at NCBI will be included.

Provide links to intronic sequences

Provide legend

Input sequence

```
gacgcggccaggcttcactccacggccacctctctcgtggagcgctggccctcgctgg  
ccgcacccggcagcc|atagatcgagatggagatggccgtgtccctggagctatttt  
aggcgccaggaaatcccgatctcgcccgatctatgcacgcaggatgtggagaagtccccct  
tatgaccag:gaaccagatctgaattgcgcgtatgcacgcaggatgtggatctaaag  
caagagccgttaacagaaatggatacatatgcgtatgcgtatgcacgcaggatgtggaaatgt  
cagaaaatgtatgcgtatgcgtatgcgtatgcacgcaggatgtggatctaaag  
aaactccctggaaactatcagacaggaggccgtccgcggccgttag:gccaatcacaca  
agctggaaagaccattacaggttccctcaggcccagcgcgcgcgttag:gtggaaaggccaggcac  
tattnaaacanontatcanaaacacccanaaacccctacacacancccccatacaccacntc
```

If the file is found to exist, I will
 overwrite it

Continue

Figure 4.6: Choose between “overwrite”, “rename the old file” or “skip the file”. In our example choose overwrite it. Since it is the first time we create intronic sequences, there are no old files to overwrite.

Step 3 - Create coding DNA reference sequence

Output for this step :

Successfully wrote coding DNA reference sequence ([TTC8 coding DNA sequence](#)) A

Successfully wrote index file ([TTC8 reference sequences](#)) B

Close

Figure 4.7: The coding DNA sequence (A, see figure 4.8) is created. The second link (B) shows all transcripts for which the reference sequence parser has been run. If there is just one, it redirects you to the coding sequence.

4. Reference sequences and the reference sequence parser

Figure 4.8: The TTC8 (NM_144596.2) coding DNA reference sequence.

5. Editing columns and legends

The objectives of this chapter are:

1. Enable a new Variant on Transcript custom data column
2. Edit the legend of the new column
3. Change column order

To start this chapter:

- Please check existing description for existing custom columns and do not deviate too much. In general, non-standard use of custom columns raises a lot of confusion.
- Log in as Curator (username: curator, password: curator1).

The screenshot shows the LOVD v3.0 Build 12 interface for the tetratricopeptide repeat domain 8 (TTC8) gene. The top navigation bar includes links for Genes, Transcripts, Variants, Individuals, Diseases, Screenings, Submit, Configuration, Documentation, and various user account options. The main content area is titled 'TTC8 configuration'. On the left, there's a sidebar with variant statistics (Total: 0, All uncurred: 0, Pending: 0, Non-public: 0, All curated: 0, Marked: 0, Public: 0) and a contact link for system manager. The central area contains sections for 'Curating TTC8 variants' and 'Custom columns for TTC8'. The 'Custom columns for TTC8' section is highlighted with a red box and arrow A, pointing to the link 'Add variant column to TTC8'. Another red box and arrow B points to the link 'View all available pre-configured variant custom columns...' under the same section. The right side of the page contains other configuration options like 'View uncurred TTC8 variants', 'View TTC8 variants', 'View variant columns enabled in TTC8', and 'LOVD scripts'.

Figure 5.1: On the “Browse VariantOnTranscript custom data columns” page you can see an overview of available custom columns. You can go there directly via “Add variant column to TTC8” on the Configuration menu tab (A). Or from the configuration area, click the “View all available pre-configured variant custom columns...” link listed under “Custom columns for TTC8”(B).

5. Editing columns and legends

Browse VariantOnTranscript custom data columns

Please note that these are all VariantOnTranscript columns available in this LOVD Installation. This is not the list of columns actually added to the system.
Also, modifications made to the columns added to a certain gene are not shown.

10 entries on 1 page. Showing entries 1 - 10.

ID	Heading	Active	HGVS	Standard	Public	Order	Form type	Created by
Location	Location	X	X	X	✓	1	Select (6 options)	LOVD
Exon	Exon	✓	X	✓	✓	2	Text (7 chars)	LOVD
DNA	DNA change (cDNA)	✓	✓	✓	✓	3	Text (30 chars)	LOVD
Published_as	Published as	X	X	X	✓	4	Text (30 chars)	LOVD
Position	Position	X	X	X	✓	5	Text (5 chars)	LOVD
RNA	RNA change	✓	✓	✓	✓	6	Text (30 chars)	LOVD
Protein	Protein	✓	✓	✓	✓	7	Text (30 chars)	LOVD
PolyPhen	PolyPhen prediction	X	X	X	✓	8	Select (4 options)	LOVD
GVS/Function	GVS function	X	X	X	✓	9	Select (14 options)	LOVD
Distance_to_splice_site	Splice distance	X	X	X	✓	10	Text (8 chars)	LOVD

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Figure 5.2: You can see that the custom column “Published_as” is not (yet) active. We are going to enable this custom column. Click on “Published_as”.

View custom data column VariantOnTranscript/Publiched_as

Data category	VariantOnTranscript
Column ID	Published_as
Column heading	Published as
Active in LOVD?	X
HGVS required column	X
Standard/Enabled by default	X
Mandatory	X
Description on form	Variant as originally reported (e.g. 521delT); provide only when different from "DNA change".
Description on short legend	Variant as originally reported (e.g. 521delT); listed only when different from "DNA change". Variants seen in animal models, tested in vitro, predicted from RNA analysis, etc. are described between brackets like c.(456C>G).
Description on full legend	Variant as originally reported (e.g. 521delT); listed only when different from "DNA change". Variants seen in animal models, tested in vitro, predicted from RNA analysis, etc. are described between brackets like c.(456C>G).
Database type	VARCHAR(100)
Form type	Text (30 chars)
Published as Variants seen in animal models, tested in vitro, predicted from RNA analysis, etc. are described between brackets like c.(456C>G). text 30	-
Regular expression pattern	-
Show to public	✓
Show on submission form	✓
Include in search form	✓
Created by	LOVD
Date created	2015-02-20 10:22:36

Options

- + Enable column (highlighted with a red box)
- Disable column
- Delete column
- / Edit custom data column settings
- || Re-order all VariantOnTranscript columns

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Figure 5.3: Click the Options drop down menu and select “Enable column”.

5. Editing columns and legends

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tetrastricopeptide repeat domain 8 (TTC8)
Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

Add/enable custom data column VariantOnTranscript/Published_as

Please select the gene(s) for which you want to add the Published_as column.

Add this column to TTC8 (tetrastricopeptide repeat domain 8)

Enter your password for authorization

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Figure 5.4: Select the gene to which you want to add this custom column. In this case we have only TTC8. When you are curator of more genes, you can select a range of genes. Confirm with your password and click “Add/enable custom data column VariantOnTranscript/Published_as”.

LOVD 3 - LOVD - Leiden Open Variation Database
tetrastricopeptide repeat domain 8 (TTC8)
Curators: LOVD3 Admin and Curator

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Browse VariantOnTranscript custom data columns

Please note that these are all VariantOnTranscript columns available in this LOVD installation. This is not the list of columns actually added to the system. Also, modifications made to the columns added to a certain gene are not shown.

10 entries on 1 page. Showing entries 1 - 10.
100 per page

ID	Heading	Active	HGVS	Standard	Public	Order	Form type	Created by
Location	Location	✗	✗	✗	✓	1	Select (6 options)	LOVD
Exon	Exon	✓	✗	✓	✓	2	Text (7 chars)	LOVD
DNA	DNA change (cDNA)	✓	✓	✓	✓	3	Text (30 chars)	LOVD
Published_as	Published as	✓	✓	✓	✓	4	Text (30 chars)	LOVD
Position	Position	✗	✗	✗	✓	5	Text (5 chars)	LOVD
RNA	RNA change	✓	✓	✓	✓	6	Text (30 chars)	LOVD
Protein	Protein	✓	✓	✓	✓	7	Text (30 chars)	LOVD
PolyPhen	PolyPhen prediction	✗	✗	✗	✓	8	Select (4 options)	LOVD
GVS/Function	GVS function	✗	✗	✗	✓	9	Select (14 options)	LOVD
Distance_to_splice_site	Splice distance	✗	✗	✗	✓	10	Text (8 chars)	LOVD

100 per page

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Figure 5.5: In the view “Browse VariantOnTranscript custom data columns” you can see that the published_as custom column has become active.

5. Editing columns and legends

TTC8 configuration

Curating TTC8 variants

- All uncurated: 0
- Pending: 0
- Non public: 0
- All curated: 0
- Marked: 0
- Public: 0

For technical assistance, such as creating new custom columns, please contact the system's manager: Manager.

Custom columns for TTC8

View all available pre-configured variant custom columns to add to the TTC8 gene database.

View the variant custom columns currently enabled for the TTC8 gene.

Configuration

- View uncurred TTC8 variants
- View TTC8 variants
- View variant columns enabled in TTC8** (Red Box A)
- Add variant column to TTC8

Documentation

Edit or sort the list of curators for the TTC8 gene database, and/or hide curators from the list of curators shown on the gene's homepage and in LOVD's header.

Download gene, transcript, variant and individual data

Download all data from the TTC8 gene database.

LOVD Scripts

The LOVD Reference sequence parser creates a nicely formatted HTML page of a coding DNA reference sequence, including exon/intron boundaries and separate files for upstream, intronic and downstream sequences. It accepts different input formats.

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Figure 5.6: To change the custom column descriptions go to “View variant columns enabled in TTC8” via the Configuration menu tab (A). Or from the configuration area, click the “View the variant custom columns currently enabled for the TTC8 gene.” link listed under “Custom columns for TTC8”(B).

View enabled custom data columns for gene TTC8

5 entries on 1 page. Showing entries 1 - 5.

ID	Heading	Width in px	Mandatory	Public	Order	Form type	Created by
Exon	Exon	50	✓	✓	2	Text (7 chars)	LOVD3 Admin
DNA	DNA change (cDNA)	200	✓	✓	3	Text (30 chars)	LOVD3 Admin
Published_as	Published as		✗	✓	4	Text (30 chars)	Curator
RNA	RNA change	200	✓	✓	6	Text (30 chars)	LOVD3 Admin
Protein	Protein	200	✓	✓	7	Text (30 chars)	LOVD3 Admin

Options

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Figure 5.7: On the “View enabled custom data columns for the gene” you can see an overview of the active custom data columns. Click on custom column “Published_as”.

5. Editing columns and legends

LOVD 3 LOVD - Leiden Open Variation Database
tetrastricopeptide repeat domain 8 (TTC8) Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View settings for custom data column Published_as for gene TTC8

Column ID	VariantOnTranscript/Published_as
Displayed width in pixels	200
Mandatory	<input checked="" type="checkbox"/>
Description on form	Variant as originally reported (e.g. 521delT); provide only when different from "DNA change".
Description on short legend	Variant as originally reported (e.g. 521delT); listed only when different from "DNA change". Variants seen in animal models, tested in vitro, predicted from RNA analysis, etc. are described between brackets like c.(456C>G).
Description on full legend	Variant as originally reported (e.g. 521delT); listed only when different from "DNA change". Variants seen in animal models, tested in vitro, predicted from RNA analysis, etc. are described between brackets like c.(456C>G).
Show to public	<input checked="" type="checkbox"/>
Show on submission form	<input checked="" type="checkbox"/>
Created by	Curator
Date created	2015-02-20 12:39:22

Options

- Edit settings for this gene only
- Remove column from this gene

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Figure 5.8: Click the Options drop down menu and select “Edit settings for this gene only”.

LOVD 3 LOVD - Leiden Open Variation Database
tetrastricopeptide repeat domain 8 (TTC8) Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

Edit settings for custom data column Published_as for gene TTC8

Column descriptions

Description on short legend (HTML enabled)	Variant as originally reported (e.g. 521delT); listed only when different from "DNA change". Variants seen in animal models, tested in vitro, predicted from RNA analysis, etc. are described between brackets like c.(456C>G).
Description on full legend (HTML enabled)	Variant as originally reported (e.g. 521delT); listed only when different from "DNA change". Variants seen in animal models, tested in vitro, predicted from RNA analysis, etc. are described between brackets like c.(456C>G).

Form settings

Notes on form (optional) (HTML enabled)	Variant as originally reported (e.g. 521delT); provide only when different from "DNA change".
---	---

If you think the data field needs clarification on the data entry form, add it here - it will appear below the field on the data entry form just like this piece of text.

Column settings

Column display width in pixels	200
(This is 200 pixels)	
Mandatory field	<input type="checkbox"/>
Show contents to public	<input checked="" type="checkbox"/>
Show field on submission form	<input checked="" type="checkbox"/>

Enter your password for authorization

B

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Figure 5.9: Here you can change the “Description on short legend” and the “Description on full legend”. Provide more information when available (A). When done, confirm with your password and click “Edit custom data column” (B). The results of these changes you can see in figure 6.10.

5. Editing columns and legends

LOVD 3 LOVD - Leiden Open Variation Database
tetrastricopeptide repeat domain 8 (TTC8)

Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View enabled custom data columns for gene TTC8

5 entries on 1 page. Showing entries 1 - 5.

100 per page

ID	Heading	Width in px	Mandatory	Public	Order	Form type	Created by
Exon	Exon	50	✓	✓	2	Text (7 chars)	LOVD3 Admin
DNA	DNA change (cDNA)	200	✓	✓	3	Text (30 chars)	LOVD3 Admin
Published_as	Published as	200	✗	✓	4	Text (30 chars)	Curator
RNA	RNA change	200	✓	✓	6	Text (30 chars)	LOVD3 Admin
Protein	Protein	200	✓	✓	7	Text (30 chars)	LOVD3 Admin

100 per page

Options
Change order of columns

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Figure 5.10: To change the order of custom data columns, go to “View enabled custom data columns for gene TTC8”, see figure 5.6 how to get there.
Click the Options drop down menu and select “Change order of columns”.

Change order of enabled custom data columns for gene TTC8

Below is a sorting list of all active columns. By clicking & dragging the arrow next to the column ID you can rearrange the columns. Re-ordering them will affect listings, detailed views and data entry forms in the same way.

Column ID
‡ Exon
‡ DNA
‡ Published_as
‡ RNA
‡ Protein

Save

Figure 5.11: You can click and drag the arrows in front of the column ID and move the columns up and down. Click save when ready. The results of these changes you can see in figure 6.10.

6. Submission

The objectives of this chapter are:

1. Update a submitters registration
2. Submit an individual, phenotype, screening and variant.

To start this chapter:

- Log in as Submitter (username: submitter, password: submitter1).

The screenshot shows the LOVD 3.0 Leiden Open Variation Database user account page. At the top right, there is a blue header bar with the text "LOVD v.3.0 Build 12 [Current LOVD status] Welcome, Submitter Your account Log out". A red arrow labeled "A" points to the "Your account" link. Below the header is a navigation menu with tabs: Genes, Transcripts, Variants, Individuals, Diseases, Screenings, Submit, and Documentation. The main content area is titled "View user account #00004" and displays a table of user information. A red arrow labeled "B" points to the "Update your registration" option in a dropdown menu under the "Options" section. The table data includes:

User ID	00004
ORCID ID	-
Name	Submitter
Institute	Leiden University Medical Center
Department	Human Genetics
Telephone	-
Address	Einthovenweg 20 2333 ZC Leiden
City	Leiden
Country	Netherlands
Email address	I.F.A.C.Fokkema@LUMC.nl
Reference	-
Curator for 0 genes	-
Collaborator for 0 genes	-
Data owner for 0 data entries	-
User level	Submitter
Created by	LOVD3 Admin
Date created	2015-02-20 10:22:50

At the bottom left, there is a "Options" dropdown menu with "Update your registration" highlighted. At the bottom right, there is a "Powered by LOVD v.3.0 Build 12" and "©2004-2015 Leiden University Medical Center" footer.

Figure 6.1: To change the users registration information, click on the “Your account” link (A). Click the Options drop down menu and select “Update your registration”.

6. Submission

LOVD 3 LOVD - Leiden Open Variation Database

Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Submitter
Your account | Log out

Genes Transcripts Variants Individuals Diseases Screenings Submit Documentation

Edit user account #00004

User details

Name: Submitter
Institute: Leiden University Medical Center
Department (optional): Human Genetics
Postal address: Eindhovenweg 20
2333 ZC Leiden

Email address(es), one per line: submitter@LOVD.nl

Telephone (optional):
New password (optional):
New password (confirm, optional):

Referencing the lab

Country: Netherlands
City: Leiden
Reference (optional):

Security

Allowed IP address list: *
Your current IP address: 127.0.0.1
Please be extremely careful using this setting. Using this setting too strictly, can deny the user access to LOVD, even if the correct credentials have been provided.
Set to "" to allow all IP addresses, use ":" to specify a range and use ";" to separate addresses or ranges.

Enter your password for authorization: *****

[Edit user](#)

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Figure 6.2: Change the e-mail address to your e-mail address in the same way as you have done in figure 1.4

6. Submission

LOVD 3 LOVD - Leiden Open Variation Database

Genes Transcripts Variants Individuals Diseases Screenings Submit **A**

Create a new individual information entry

To create a new individual information entry, please fill out the form below.

Lab ID: **B** 16308660-Fams

Reference (optional):

Remarks (optional):

Panel size: 1

ID of panel this entry belongs to (optional):

This individual has been diagnosed with these diseases:

C Relation to diseases:

- Healthy/Control (Healthy individual / control)
- BBS (BARDET-BIEDL SYNDROME)
- RP51 (RETINITIS PIGMENTOSA 51)

Diseases not in this list are not yet configured in this LOVD. If any disease you would like to select is not in here, please mention this in the remarks, preferably including the omim number. This way, a manager can configure this disease in this LOVD.

Create individual information entry

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Figure 6.3: When you are logged in as a submitter, a submission starts with creating new individual information.
 Click on the Submit menu tab (A). Enter the Lab ID (B) and select a related disease (C). Provide more information when available.
 When you are done, click “Create individual information entry”.

LOVD 3 LOVD - Leiden Open Variation Database

Genes Transcripts Variants Individuals Diseases Screenings Submit Documentation **B**

Submission of individual #00000001

What would you like to do?

A I want to add phenotype information to this individual

I want to add a variant screening to this individual

I want to finish this submission

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Figure 6.4: To add phenotype information to the individual you just created, click “I want to add phenotype information to this individual” (A).
 Note that during a submission you can not return to the individual information entry form in case you made an error or forgot something. If you made an error or forgot something, you have to proceed with your submission and when ready you can edit your individual via the “Unfinished submissions” link (B). This also applies for phenotypes, screenings and variants.

6. Submission

LOVD³ LOVD - Leiden Open Variation Database

Leiden Open Variation Database

LOVD v.3.0 Build 12 [[Current LOVD status](#)]
[Welcome, Submitter](#)
[Your account](#) | [Unfinished submissions](#) | [Log out](#)

[Genes](#) [Transcripts](#) [Variants](#) [Individuals](#) [Diseases](#) [Screenings](#) [Submit](#) [Documentation](#)

Create a new phenotype information entry for individual #00000001

Phenotype Information related to BARDET-BIEDL SYNDROME

Additional phenotype details (optional)
Additional information on phenotype of this individual.

Inheritance (optional)
Unknown
Indicates the inheritance of the phenotype in the family; unknown, familial (autosomal/X-linked, dominant/recessive), paternal (Y-linked), maternal (mitochondrial), isolated (sporadic) or complex

[Create phenotype information entry](#) [Cancel](#)

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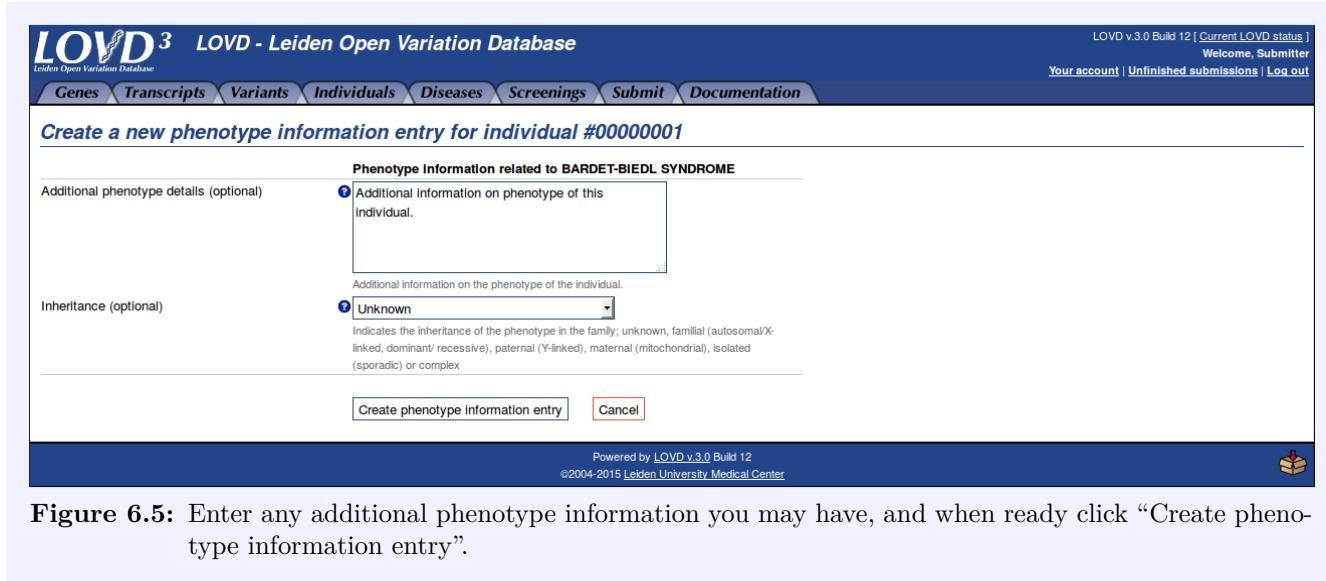


Figure 6.5: Enter any additional phenotype information you may have, and when ready click “Create phenotype information entry”.

LOVD³ LOVD - Leiden Open Variation Database

Leiden Open Variation Database

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Submission of individual #00000001

What would you like to do?

I want to add phenotype information to this individual
» I want to add a variant screening to this individual
I want to finish this submission

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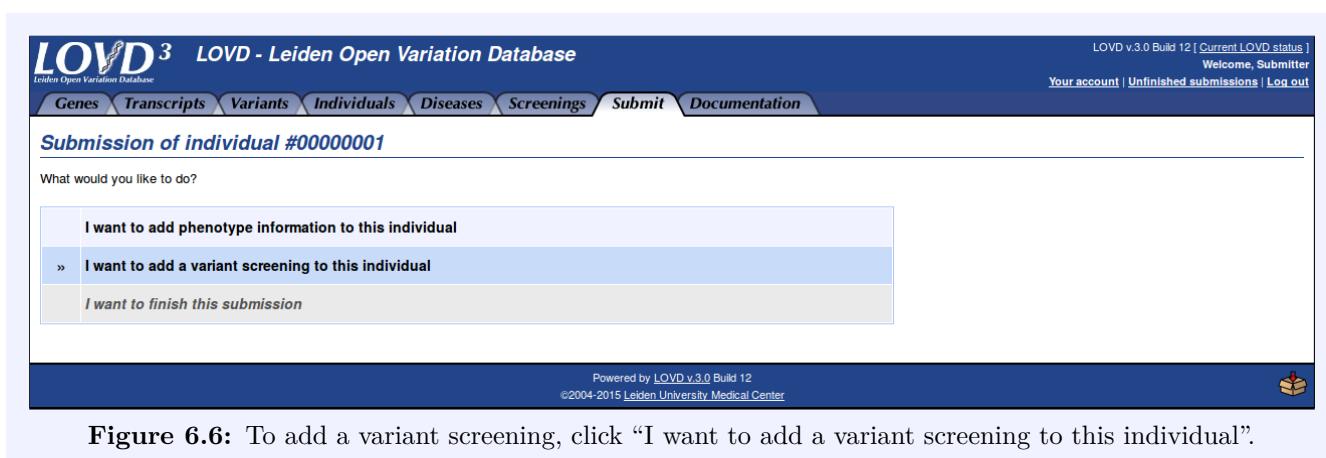


Figure 6.6: To add a variant screening, click “I want to add a variant screening to this individual”.

6. Submission

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Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
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Create a new screening information entry for individual #00000001

To create a new screening information entry, please fill out the form below.

Screening Information

Detection template DNA A

RNA (cDNA)
 Protein

Technique(s) used Reverse Transcription and PCR B

SEQuencing
 Single Base Extension
 Single-Strand DNA Conformation polymorphism Analysis (SSCP)
 SSCP, fluorescent (SSCP)

Genes screened TTC8 (tetrafunctional repeat domain 8) C

Please select no more than 15 genes. For genome-wide analysis, no genes should be selected.

Have variants been found?

[Create screening information entry](#) [Cancel](#)

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Figure 6.7: Select a detection template (A), technique(s) used (B) and gene(s) screened (C). Multiple lines can be selected in all three fields.
When you made your selection, click “Create screening information entry”.

LOVD 3 LOVD - Leiden Open Variation Database

Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
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[Genes](#) [Transcripts](#) [Variants](#) [Individuals](#) [Diseases](#) [Screenings](#) [Submit](#) [Documentation](#)

Submission of screening #0000000001

What would you like to do?

» [I want to add a variant to this screening](#)

[Back to the individual](#)

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Figure 6.8: Now we’re going to add a variant to the screening we just entered. Click “I want to add a variant to this screening”.

6. Submission

LOVD 3 LOVD - Leiden Open Variation Database

Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Submitter
Your account | Unfinished submissions | Log out

Genes Transcripts Variants Individuals Diseases Screenings Submit Documentation

Create a new variant entry

Do you want to confirm already submitted variants with this screening?

Yes, I want to confirm variants found using this screening »»

What kind of variant would you like to submit?

A variant that is located within a gene »» A
 A variant that was only described on genomic level »»

Please find the gene for which you wish to submit this variant below, using the search fields if needed. Click on the gene to proceed to the variant entry form.

1 entry on 1 page. Showing entry 1.
10 per page ▾

Symbol	Gene	Chr	Band	Unique variants
=TTC8*				
TTC8	tetratricopeptide repeat domain 8	3		0

10 per page ▾

Cancel

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Figure 6.9: Click “A variant that is located within a gene” (A). Thereafter, you can select a gene to which you want to add a variant. Click on TTC8 (B).

6. Submission

LOVD 3 Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Submitter
[Your account](#) | [Unfinished submissions](#) | [Log out](#)

Create a new variant entry for gene TTC8

To create a new variant entry, please fill out the form below.

[Ignore this transcript](#)

Transcript variant on NM_144596.2 (TTC8)

Exon [DNA change \(HGVS format\)](#) [RNA change \(HGVS format\)](#) [Protein change \(HGVS format\)](#)

Published as (optional)

Affects function (reported)

[Ignore this transcript](#)

Transcript variant on NM_198309.2 (TTC8)

Exon [DNA change \(HGVS format\)](#) [RNA change \(HGVS format\)](#) [Protein change \(HGVS format\)](#)

Published as (optional)

Affects function (reported)

Genomic variant information

Allele If you wish to report an homozygous variant, please select "Both (homozygous)" here.

Chromosome Relative to hg19 / GRCh37.

Reference (optional)

Frequency (optional)

Affects function (reported)

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Figure 6.10: In chapter “Editing columns and legends” we have changed the order and the legend of custom column “Published_as”. The position of the “Published_as” field depends on what you have changed there.
Move your mouse over the blue help icon in front of the “Published_as” field, to see the changes you made in the description or notes fields.
Enter “c.284A>G” in the DNA change field of transcript NM_144596.2 and click “Map to genome”.

6. Submission

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Leiden Open Variation Database

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Welcome, Submitter
Your account | Unfinished submissions | Log out

Genes Transcripts Variants Individuals Diseases Screenings Submit Documentation

Create a new variant entry for gene TTC8

To create a new variant entry, please fill out the form below.

Ignore this transcript

Exon

DNA change ([HGVS format](#))

RNA change ([HGVS format](#))

Protein change ([HGVS format](#))

Published as (optional)

Affects function (reported)

Transcript variant on NM_144596.2 (TTC8)

Exon

DNA change ([HGVS format](#))

RNA change ([HGVS format](#))

Protein change ([HGVS format](#))

Published as (optional)

Affects function (reported)

Transcript variant on NM_198309.2 (TTC8)

Exon

DNA change ([HGVS format](#))

RNA change ([HGVS format](#))

Protein change ([HGVS format](#))

Published as (optional)

Affects function (reported)

Genomic variant Information

Allele

Chromosome

Genomic DNA change ([HGVS format](#))

Reference (optional)

Frequency (optional)

Affects function (reported)

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Figure 6.11: The fields at arrows A, B and C are filled by LOVD.

6. Submission

LOVD 3 LOVD - Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Submitter
[Your account](#) | [Unfinished submissions](#) | [Log out](#)

[Genes](#) [Transcripts](#) [Variants](#) [Individuals](#) [Diseases](#) [Screenings](#) [Submit](#) [Documentation](#)

Create a new variant entry for gene TTC8

To create a new variant entry, please fill out the form below.

Ignore this transcript

Exon (c.284A>G ✓
r.(?) ✓
p.(Lys95Arg) ✓
254A>G

DNA change (HGVS format)
RNA change (HGVS format)
Protein change (HGVS format)
Published as (optional)

Affects function (reported) Effect unknown

Ignore this transcript

Exon c.254A>G ✓
r.(?) ✓
p.(Lys85Arg)
Variant as originally reported (e.g. 521delT); provide only when different from "DNA change".

DNA change (HGVS format)
RNA change (HGVS format)
Protein change (HGVS format)
Published as (optional)

Affects function (reported) Effect unknown

Genomic variant information

Allele Unknown If you wish to report an homozygous variant, please select "Both (homozygous)" here.
14 g.89307227A>G ✓ Relative to hg19 / GRCh37.

Reference (optional) PMID:Stoetzel 2006:16308660 (Active custom links : PubMed, DbsNP, GenBank, OMIM) A

Frequency (optional) 0.012
Affects function (reported) Effect unknown

Create variant entry **Cancel**

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Figure 6.12: Fill in the other fields as indicated in the figure. Below the reference field, the active custom links are displayed (A).

When you hover your mouse cursor over the active custom links, you will see a small window with some custom link information and examples.

When you click on one of the active custom links, the custom link format is entered in the field.

LOVD 3 LOVD - Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Submitter
[Your account](#) | [Unfinished submissions](#) | [Log out](#)

[Genes](#) [Transcripts](#) [Variants](#) [Individuals](#) [Diseases](#) [Screenings](#) [Submit](#) [Documentation](#)

Submission of screening #0000000001

What would you like to do?

I want to add a variant to this screening
Back to the individual
I want to finish this submission

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Figure 6.13: You can finish your submission by clicking "I want to finish this submission". Check your e-mail, you should have received a message that there is a new submission in your gene database.

7. Curating a submission

The objective of this chapter is:

1. Curate submitted data.

To start this chapter:

- Log in as Curator (username: curator, password: curator1).

LOVD 3 - Leiden Open Variation Database
tetrapeptide repeat domain 8 (TTC8)
Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

TTC8 configuration

Variants (Total: 1)
All uncurated: 1
Pending: 1
Non public: 0
All curated: 0
Marked: 0
Public: 0

For technical assistance, such as creating new custom columns, please contact the system's manager: Manager

Curating TTC8 variants
View all uncurated variant entries in the TTC8 gene database (newly submitted, non public and marked entries).
View all data submissions in the TTC8 gene database.

Custom columns for TTC8
View all available pre-configured variant custom columns to add to the TTC8 gene database.
View the variant custom columns currently enabled for the TTC8 gene.

Gene settings
Edit or sort the list of curators for the TTC8 gene database, and/or hide curators from the list of curators shown on the gene's homepage and in LOVD's header.
Delete all variants and associated data from the TTC8 gene database.
Data associated with other genes as well will be kept.

Download gene, transcript, variant and Individual data
Download all data from the TTC8 gene database.

LOVD scripts
The LOVD Reference sequence parser creates a nicely formatted HTML page of a coding DNA reference sequence, including exon/intron boundaries and separate files for upstream, intronic and downstream sequences. It accepts different input formats.

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Figure 7.1: Click on the Configuration menu tab. On the left part of your screen you can see an overview of the number of variants in your gene database (A). One variant in uncurated. To curate the new variant, click the “View all uncurated variant entries in the TTC8 gene database” link listed under “Curating TTC8 variants”(B).

Note: the link in the submission email that was send to the curator will take you directly to the following page.

LOVD 3 - Leiden Open Variation Database
tetrapeptide repeat domain 8 (TTC8)
Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View transcript variants in TTC8

The variants shown are described using the NM_144596.2 transcript reference sequence.

Effect	Exon	DNA change (cDNA)	RNA change	Protein	Published as	DNA change (genomic)	Reference	DB-ID	Frequency	Owner	Var. status
?	04	c.284A>G	t.(?)	p.(Lys95Arg)	254A>G	g.89007227A>G	[Stoezl]	-	0.012	Submitter	Pending

1 entry on 1 page. Showing entry 1.
100 per page Legend
100 per page Legend

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Figure 7.2: You will see a variant which is grey and with status pending, click on the variant.

7. Curating a submission

LOVD 3 LOVD - Leiden Open Variation Database
tetra-tripeptide repeat domain 8 (TTC8)
Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View genomic variant #0000000001

Individual ID	00000001 (Pending) A
Chromosome	14
Allele	Unknown
Affects function (reported)	Effect unknown
Affects function (concluded)	Not classified
DNA change (genomic)	g.89307227A>G
Reference	[Steetzel]
DB-ID	-
Frequency	0.012
Automatic mapping	Off
Average frequency (large NGS studies)	Retrieve
Owner	Submitter
Variant data status	Pending B
Created by	Submitter
Date created	2015-02-26 10:46:57
Last edited by	N/A
Date last edited	N/A

Options

- Edit variant entry
- Publish (curate) variant entry C
- Manage transcripts for this variant
- Delete variant entry
- Search public LOVDs
- Visualize in UCSC genome browser
- Visualize in Ensembl genome browser

TTC8
NM_144596.2
Effect unknown
Not classified
04
c.284A>G
r.(?)
p.(Lys95Arg)
254A>G

Variant on transcripts

Gene	Transcript	Affects function	Exon	DNA change (cDNA)	RNA change	Protein	Published as
TTC8	NM_144596.2	?.	04	c.284A>G	r.(?)	p.(Lys95Arg)	254A>G
TTC8	NM_198309.2	?.	04	c.254A>G	r.(?)	p.(Lys85Arg)	-

Screenings

Screening ID	Template	Technique	Genes screened	Variants found	Owner
0000000001	DNA	SEQ	TTC8	1	Submitter

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Figure 7.3: On the “View genomic variant” page you can see that the individual (A) and the variant (B) both have status pending. If the information is correct, you can curate the variant (C) by clicking on the Options drop down menu and selecting “Publish (curate) variant entry”. This variant is now published and visible to others.

7. Curating a submission

LOVD 3 Leiden Open Variation Database

tetratricopeptide repeat domain 8 (TTC8)

Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View genomic variant #000000001

Individual ID	00000001 (Pending) B
Chromosome	14
Allele	Unknown
Affects function (reported)	Effect unknown
Affects function (concluded)	Not classified
DNA change (genomic)	g.89307227A>G
Reference	Sheetzell
DB-ID	TTC8_000001
Frequency	0.012
Automatic mapping	Off
Average frequency (large NGS studies)	Retrieve
Owner	Submitter
Variant data status	Public A
Created by	
Date created	2015-02-26 10:46:57
Last edited by	Curator
Date last edited	2015-02-26 10:51:06

[Options](#)

Variant on transcripts

Gene	Transcript	Affects function	Exon	DNA change (cDNA)	RNA change	Protein	Published as
TTC8	NM_144596.2	?.	04	c.284A>G	r.(?)	p.(Lys95Arg)	254A>G
TTC8	NM_198309.2	?.	04	c.254A>G	r.(?)	p.(Lys85Arg)	-

Screenings

Screening ID	Template	Technique	Genes screened	Variants found	Owner
000000001	DNA	SEQ	TTC8	1	Submitter

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LOVD 3 LOVD - Leiden Open Variation Database
 Leiden Open Variation Database
 Curators: LOVD Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View individual #00000001

Lab-ID	16308660-Fams
Reference	-
Remarks	-
Remarks (non public)	-
Panel size	1
Diseases	RSS
Owner name	Submitter
Individual data status	Pending
Created by	Submitter
Date created	2015-02-26 10:34:41
Last edited by	N/A
Date last edited	N/A

Options:

- Edit individual entry
- Publish (curate) individual entry **A**
- Add phenotype information to individual
- Add screening to Individual
- Delete individual entry

Publ (curate) individual entry

dyte for: Associate

Inheritance Owner Status

Submitter Pending

Screenings

Screening ID	Template	Technique	Genes screened	Variants found	Owner
0000000001	DNA	SEQ	TTC8	1	Submitter

Variants

1 entry on 1 page. Showing entry 1.

100 per page

Chr	Allele	DNA change (genomic)	Reference	DB-ID	Frequency	Owner	Var. status	Effect	Exon	DNA change (cDNA)	Published as	RNA change	Protein
14	Unknown	g.89307227A-G	[Submitter]	TTC8_000001	0.012	Submitter	Public	??	04	NM_144596.2:c.284A>G, NM_198309.2:c.254A>G	254A>G,	t(?)	p.(Lys95Arg), p.(Lys85Arg)

100 per page

Powered by LOVD v.3.0 Build 12 | Current LOVD status
[Welcome](#) [Curator](#)
[Your account](#) [Log out](#)

Figure 7.5: If the information is correct, you can curate the individual (A) by clicking on the Options drop down menu and selecting “Publish (curate) individual entry”.

7. Curating a submission

LOVD 3 LOVD - Leiden Open Variation Database

tetratricopeptide repeat domain 8 (TTC8)

Curator: LOVD Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View individual #00000001

Lab-ID: 1630860-Fams
Reference:
Remarks:
Remarks (non public):
Panel size: 1
Diseases: BBS
Owner name: Submitter
Individual data status: Public
Created by: Submitter
Date created: 2015-02-26 10:34:41
Last edited by: Curator
Date last edited: 2015-02-26 10:51:55

Options:

Phenotypes

BARDET-BIEDL SYNDROME (BBS) Add phenotype for this disease

Phenotype ID: 00000001 Phenotype details: Inheritance: Owner: Status: Pending

Additional information on phenotype of this individual: Submitter: Pending

A red arrow labeled 'A' points to the 'Status: Pending' field.

Screenings

Screening ID	Template	Technique	Genes screened	Variants found	Owner
00000001	DNA	SEO	TTC8	1	Submitter

Variants

1 entry on 1 page. Showing entry 1.
100 per page Legend

Chr	Allele	DNA change (genomic)	Reference	DB-ID	Frequency	Owner	Var. status	Effect	Exon	DNA change (cDNA)	Published as	RNA change	Protein
14	Unknown	g.89307227A>G	[Submitter]	TTC8_000001	0.012	Submitter	Public	P	04	NM_144598.2:c.284A>G, NM_198359.2:c.254A>G	254A>G, c.254A>G	P.Lys85A(g), P.Lys85A(g)	

100 per page Legend

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Figure 7.6: The phenotype submission is still gray. Click on the phenotype entry to curate.

LOVD 3 LOVD - Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Curator
Your account | Log out

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View phenotype #0000000001

Individual ID: 00000001 (Public)
Associated disease: BBS
Phenotype details: Additional information on phenotype of this individual.
Inheritance: -
Owner name: Submitter
Phenotype data status: Pending
Created by: Submitter
Date created: 2015-02-26 10:35:21
Last edited by: N/A
Date last edited: N/A

Options:

- Edit phenotype information
- Publish (curate) phenotype entry** (highlighted)
- Delete phenotype entry

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Figure 7.7: If the information is correct, you can curate the phenotype by clicking on the Options drop down menu and select “Publish (curate) phenotype entry”.

8. Export and import

The objectives of this chapter are:

1. Export your gene data
2. Delete your gene data
3. Import your gene data

To start this chapter:

- Log in as Curator (username: curator password: curator1).

The screenshot shows the LOVD 3.0 Configuration page for the TTC8 gene. The top navigation bar includes links for Genes, Transcripts, Variants, Individuals, Diseases, Screenings, Submit, Configuration, and Documentation. The Configuration tab is active. On the left, there's a sidebar with variant statistics: Total: 1, All uncurated: 0 (Pending: 0, Non public: 0), All curated: 1 (Marked: 0, Public: 1). Below this is a note for system managers about custom columns. The main content area has three sections: 'Curating TTC8 variants' (with links to uncurred variants and submissions), 'Custom columns for TTC8' (with links to available and currently enabled custom columns), and 'Gene settings' (with links to edit the database, manage curators, and delete variants). At the bottom right of this section is a red box containing the 'Download gene, transcript, variant and individual data' link, which is also highlighted with a red arrow. To the right of this box is the 'LOVD scripts' section, which describes the Reference sequence parser. The footer contains the LOVD version (v.3.0 Build 12), copyright information (©2004-2015 Leiden University Medical Center), and a small logo.

Figure 8.1: On two places you can download your gene related data. You can do that from the Configuration area, by clicking the “Download all data from the TTC8 gene database” link listed under “Download gene, transcript, variant and individual data”. Or you can do that from the “View gene” page, see figure 8.2.

8. Export and import

LOVD 3 LOVD - Leiden Open Variation Database

tetratricopeptide repeat domain 8 (TTC8)

Curators: LOVD3 Admin and Curator

Genes Transcripts Variants Individuals Diseases Screenings Submit Configuration Documentation

View gene TTC8

General information

Gene symbol	TTC8
Gene name	tetratricopeptide repeat domain 8
Chromosome	14
Chromosomal band	q31.3
Imprinted	Unknown
Genomic reference	NC_000014.8
Transcript reference	NM_144596.2 , NM_198309.2
Exon/intron information	NM_144596.2
Associated with diseases	BBS , RP51
Citation reference(s)	-
Allow public to download all variant entries	x
Allow data to be indexed by WikiProfessional	x
Refseq URL	Genomic reference sequence
Curators (2)	LOVD3 Admin and Curator
Collaborators (0)	-
Total number of public variants reported	1
Unique public DNA variants reported	1
Individuals with public variants	1
Hidden variants	0
Created by	LOVD3 Admin
Date created	2015-02-26
Last edited by	Curator
Date last edited	2015-02-26 10:19:33
Last updated by	Curator
Date last updated	2015-02-26 10:51:06
Version	TTC8:150226

Graphical displays and utilities

Graphs	Graphs displaying summary information of all variants in the database »
UCSC Genome Browser	Show variants in the UCSC Genome Browser (full view , compact view)
Ensembl Genome Browser	Show variants in the Ensembl Genome Browser (full view , compact view)
NCBI Sequence Viewer	Show distribution histogram of variants in the NCBI Sequence Viewer

Links to other resources

HGNC	20087
Entrez Gene	123016
PubMed articles	TTC8
OMIM - Gene	608132
OMIM - Diseases	BBS (BARDET-BIEDL SYNDROME) RP51 (RETINITIS PIGMENTOSA 51)
HGMD	TTC8
GeneCards	TTC8
GeneTests	TTC8

Options ▾

- Edit gene information
- Add transcript(s) to gene
- Sort/Hide curator names
- Empty this gene database **B**
- View graphs about this gene database
- View enabled variant columns
- Re-order enabled variant columns
- View all available variant columns
- Download all this gene's data **A**
- Create human-readable refseq file

NCBI ID	NCBI Protein ID	Variants
NM_144596.2	NP_653197.2	1
NM_198309.2	NP_938051.1	1

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Figure 8.2: Also from the “View gene” page you can download the gene related data. Click the Options drop down menu and select “Download all this gene’s data” (A). The download begins immediately. When you have downloaded and saved your gene data, we are going to empty this gene database. Click the Options drop down menu and select “Empty this gene database” (B).

8. Export and import

Empty TTC8 gene database

Emptying TTC8 gene database

Deleting 2 variants and all associated data (screenings, individuals, phenotypes).
All data (variants, screenings, Individuals and phenotypes) only linked to TTC8 and not linked to any other gene will be deleted!

Enter your password for authorization

Empty gene database

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Figure 8.3: You get an extra warning that you are about to delete data and which data. Note that genes, transcripts and diseases are not deleted. Confirm with your password.

LOVD Setup

Leiden Open Variation Database

- Installed : 2015-02-26
- Updated : -
- Statistics**
 - Users : 4
 - Log entries : 31
 -
 - Individuals : 0
 - Genes : 1
 - Variants**
 - Total : 0

General LOVD Setup

View and change LOVD System settings, including settings on statistics, security and the legend.

Authorized users

Create a new authorized user or submitter.
Manage authorized users and submitters.

Custom data columns

Create new custom data column.
Browse all custom data columns already available to enable or disable them, or view or edit their settings.
Download all LOVD custom columns in the LOVD import format.

Custom links

Create a new custom link. Custom links allow you to quickly insert references to other data sources, using short tags.
Browse all available custom links and view and edit their settings.

Download & Import

Download all data in LOVD import format (custom columns, genes, transcripts, diseases, individuals, phenotypes, screenings & variants).

Import data using the LOVD import format (custom columns, diseases, individuals, phenotypes, screenings & variants).

System logs

View, search and delete system logs.

Gene databases

Create a new gene database.
Manage configured gene databases.

Transcripts

Create a new transcript.
Manage transcripts.

Diseases

Create a new disease information entry.
Manage disease information entries.

Individuals

Create new individual entry.
Manage individuals.

Variants

Create a new variant.
Manage variants.

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Figure 8.4: Only managers can import data into LOVD3 for the moment. Therefore log in as Manager (username: manager, password: manager1). You can import the data from the Setup area. Click the “Import data using the LOVD import format” link listed under “Download & Import”.

8. Export and import

LOVD 3 LOVD - Leiden Open Variation Database

Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Manager
Your account | Log out

Genes Transcripts Variants Individuals Diseases Screenings Submit Users Configuration Setup Documentation

Import data in LOVD format

Using this form you can import files in LOVD's tab-delimited format. Currently supported imports are custom column, individual, phenotype, screening and variant data.
Genomic positions in your data are assumed to be relative to Human Genome build hg19.

If you're looking for importing data files containing variant data only, like VCF files and SeattleSeq annotated files, please [start a new submission](#).

In some cases importing big files or importing files into big databases can cause LOVD to run out of available memory. In case this server hides these errors, LOVD would return a blank screen. If this happens, split your import file into smaller chunks or ask your system administrator to allow PHP to use more memory (currently allowed: 128MB).

Select the file to import

File selection (LOVD tab-delimited format only!)

Browse... LOVD_full_download_TTC8_2015-02-26_11.06.04.txt

The maximum file size accepted is 2 MB, due to restrictions on this server. If you wish to have it increased, contact the server's system administrator.

Import options

Import mode: Add only, treat all data as new

Please select which import mode LOVD should use; Update existing data & add new data (not yet implemented) or Add only, treat all data as new. For more information on the modes, move your mouse over the ? icon.

Character encoding of imported file: Autodetect

Please only change this setting in case you encounter problems with displaying special characters in imported data. Technical information about character encoding can be found [on Wikipedia](#).

Simulate (don't actually import the data):

Import file

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Figure 8.5: Select the file you downloaded at the beginning of this chapter (A). Check the Simulate check box to do a test import. Click “Import file”.

8. Export and import

LOVD 3 LOVD - Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Manager
Your account | Log out

Genes Transcripts Variants Individuals Diseases Screenings Submit Users Configuration Setup Documentation

Import data in LOVD format

Using this form you can import files in LOVD's tab-delimited format. Currently supported imports are custom column, individual, phenotype, screening and variant data. Genomic positions in your data are assumed to be relative to Human Genome build hg19.

Information: If you're looking for importing data files containing variant data only, like VCF files and SeattleSeq annotated files, please start a new submission.

Warning: In some cases importing big files or importing files into big databases can cause LOVD to run out of available memory. In case this server hides these errors, LOVD would return a blank screen. If this happens, split your import file into smaller chunks or ask your system administrator to allow PHP to use more memory (currently allowed: 128MB).

Simulation successful: no errors found.
[Hide 1 warning](#)

Warning: There is already a disease with disease name BARDET-BIEDL SYNDROME. This disease is not imported!

Select the file to import

File selection (LOVD tab-delimited format only)

Browse... LOVD_full_download_TTC8_2015-03-06_10.41.27.txt

The maximum file size accepted is 2 MB, due to restrictions on this server. If you wish to have it increased, contact the server's system administrator.

Import options

Import mode: Add only, treat all data as new

Please select which import mode LOVD should use: Update existing data & add new data (not yet implemented) or Add only, treat all data as new. For more information on the modes, move your mouse over the ? icon.

Character encoding of imported file: Autodetect

Please only change this setting in case you encounter problems with displaying special characters in imported data. Technical information about character encoding can be found on Wikipedia.

Simulate (don't actually import the data):

A

B

C

Import file

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Figure 8.6: The import simulation has been successful (A). But a warning is given, this is because the diseases were not deleted. LOVD recognized the disease in the import file. To do a real import, select the file again (B), uncheck the Simulate check box (C) and click “Import file”.

LOVD 3 LOVD - Leiden Open Variation Database

LOVD v.3.0 Build 12 [Current LOVD status]
Welcome, Manager
Your account | Log out

Genes Transcripts Variants Individuals Diseases Screenings Submit Users Configuration Setup Documentation

Import data in LOVD format

Parsing file... 100%

Warning: There is already a disease with disease name BARDET-BIEDL SYNDROME. This disease is not imported!

Applying changes... 100%

Done importing!

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Figure 8.7: Import is done

8. Export and import

The screenshot shows the LOVD 3 - Leiden Open Variation Database interface. The top navigation bar includes links for Genes, Transcripts, Variants, Individuals, Diseases, Screenings, Submit, Users, Configuration, Setup, and Documentation. The current page is 'View genomic variant #0000000002'. The main content area displays the variant details, including individual ID (00000002), chromosome (14), allele (Unknown), and various clinical and technical annotations. A red arrow points to the individual ID field. Below this is a table of transcripts for the gene TTC8, showing exon, DNA change, RNA change, protein, and published status. Another red arrow points to the screening table, where the screening ID '0000000002' is highlighted. The bottom of the page includes a footer with copyright information and a logo.

View genomic variant #0000000002

Individual ID	00000002 (Public)
Chromosome	14
Allele	Unknown
Affects function (reported)	Effect unknown
Affects function (concluded)	Not classified
DNA change (genomic)	g.89307227A>G
Reference	[Stoezel]
DB-ID	TTC8_0000001
Frequency	0.012
Automatic mapping	Off
Average frequency (large NGS studies)	Retrieve
Owner	Submitter
Variant data status	Public
Created by	Submitter
Date created	2015-02-26 10:46:57
Last edited by	Curator
Date last edited	2015-02-26 10:51:06

Options

Variant on transcripts

Gene	Transcript	Affects function	Exon	DNA change (cDNA)	RNA change	Protein	Published as
TTC8	NM_144596.2	?/.	04	c.284A>G	r.(?)	p.(Lys95Arg)	254A>G
TTC8	NM_198309.2	?/.	04	c.254A>G	r.(?)	p.(Lys85Arg)	-

Screenings

Screening ID	Template	Technique	Genes screened	Variants found	Owner
0000000002	0000000002	TTC8	1	Submitter

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Figure 8.8: Your database now looks the same as before you deleted the gene's data. Only the ID's are different: individual ID, screening ID and variant ID.

9. Example E-mails

9.1 Welcome new submitter

Dear ... ,

First of all, welcome as new submitter to the gene variant database at the Leiden Muscular Dystrophy pages (www.DMD.nl); great that you have started to actually submit gene variants!

In future new entries will become uploaded automatically and without notification, unless we have specific questions. Please note that after login you can update your records when more information comes in. When you notice mistakes or omissions in the other entries of the database, please do not forget to notify us immediately.

Regarding your submissions we have some questions -just to be sure that we curate them correctly;

...

Since this is the first variant(s) you report we assume that you may have found more in the past. If so, please note that the database tries to store ALL variants identified and not only variants that have not been reported before. This includes ALL variant found in a specific patient (pathogenic AND non-pathogenic). When you have identified more gene variants, please consider submitting these as well. Note that for submission of larger sets of variants we can help you: you send us the variants in electronic format, preferably a spreadsheet format.

Yours sincerely,

Curator: ...

Department: ...

Institute: ...

Database URL: ...

9.2 New publication

Dear ... ,

In PubMed we came across your article entitled “...” (...). When the final version of the paper is out, we would very much appreciate to receive a copy of the paper, preferably in electronic format (.PDF).

We have added the sequence variants described to the DMD gene variant database at (<http://www.LOVD.nl/DMD>). Please have a look to see whether we have done this correctly. We concluded that some patients were described earlier by ... et al; is this correct? Some details from the paper were not clear (see below), please check and clarify;

... one variant was described as ... but this seems not correct ...

... it was unclear which variants where found in which combinations (recessive disease)

... you list some variants as percentages but we would appreciate to list the number of variants alleles / alleles tested (like 12/98)

... phenotype data are largely lacking, we would appreciate to receive any details you may have

In the mean time you probably have identified more variants in the gene and/or other genes involved in the neuromuscular disorders covered by our databases. We would very much appreciate to receive an overview of these variants to be able to add them to the database. Note that the database tries to store ALL variants identified and not only variants that have not been reported before. This includes ALL variants found in a specific patient (pathogenic AND non-pathogenic). When you have identified more gene variants, please consider to submit these as well. Note that for submission of larger sets of variants we can offer our help when you send us the variants in electronic format, preferably a spreadsheet format.

We noticed that you mention our database in your paper. Please consider to register as submitter and submit all variants you identify directly to the database. Note that only a complete and fully up-to-date database is most helpful for those using it, in particular those using it to perform accurate DNA diagnostics for patients and their relatives.

Yours sincerely,

Curator: ...

Department: ...

Institute: ...

Database URL: ...