

ClinGen VCEP SOP for ClinVar Registration and ClinVar Submission

A. Registering in ClinVar

1. Only register after ClinVar has confirmed receipt of the EP application and specifications (i.e. - please wait until a folder for your VCEP is created on the ClinVar/ClinGen FTP site: ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/ClinGen/expert_panels/).
2. You must have a MyNCBI account to register. If you do not have a MyNCBI account, you can create one by going to <https://submit.ncbi.nlm.nih.gov/clinvar/>, clicking the log in button, and following instructions to create an account. Please make sure that your account has your first and last name. For more information on how to create an account, please visit: https://www.ncbi.nlm.nih.gov/clinvar/docs/submission_portal/



3. To begin registration, navigate to <https://submit.ncbi.nlm.nih.gov/clinvar/> and select “New Organization”

Tab 1: Organization Information

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2 SUBMITTER INFORMATION

3 OVERVIEW

Organization Information

Your ClinVar submission will be attributed to the combination of your organization and institution (i.e., Organization name, Institution name). We consider the organization to be a smaller entity within the larger institution. The organization is typically a lab, center, clinic, department, etc. The institution is typically a university, a hospital, or a research center like the NIH. This distinction allows ClinVar to give proper attribution when there are multiple submitting organizations from the same institution. Examples:

- * Emory Genetics Laboratory, Emory University School of Medicine
* Inova Translational Medicine Institute, Inova Health System
* Richard Lifton Laboratory, Yale University
* For a company with a single name, please avoid redundancy and store only the Organization name, e.g., GeneDx or Invitae

* Organization name ?

* Organization abbreviation ?

* Organization type
consortium

Institution name ?
ClinGen

Department name ?

Street address ?

* City ?
Bethesda

State or Province ?
Maryland

* Country or Region ?
United States

Postal code ?
20892

Organization URL ?

Base URL for variant links ?

If your organization is registered with GTR and you know your organization ID, [use this link to fill in this form with the data we already have.](#)

Continue

Organization Name: Use the official name from the “[ClinGen VCEP and GCEP Names](#)” google spreadsheet, ‘ClinVar Submission & Publication (External)’ column (Column W).

Organization abbreviation: Use the abbreviated name from the ‘Website (VCEP Short Name)’ column (Column U) from the “[ClinGen VCEP and GCEP Names](#)” google spreadsheet.

Organization type: Please select 'Consortium'

Institution Name: ClinGen

City: Bethesda

State: Maryland

Country: United States

Postal code: 20892

Organization URL: Please put the URL for your VCEP page on the ClinGen webpage. For example, the PTEN VCEP would list "<https://clinicalgenome.org/affiliation/50012/>"

Base URL for variant links: Leave empty

Tab 2: Submitter Information

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Submitter

* First name ⓘ	* Last name ⓘ
Steven	Harrison
* Phone number ⓘ	* Email address ⓘ
	sharrison6@partners.org
dbSNP handle ⓘ	ClinVar Person ID ⓘ
	2800
<input checked="" type="checkbox"/> Display my name with my organization on ClinVar's website	
Role ⓘ	
<div></div>	

Other submitters

* First name ⓘ	* Last name ⓘ	* Phone number ⓘ	* Email address ⓘ	DbSNP handle ⓘ	ClinVar Person ID ⓘ	Public <input checked="" type="checkbox"/>	Role ⓘ	Delete
							<div></div>	
Add another Submitter								

Continue

The Submitter section is automatically completed for the person registering the organization in ClinVar. Please select your role and whether or not your name and contact information should be displayed on the submitter page on ClinVar. For example:

<https://www.ncbi.nlm.nih.gov/clinvar/submitters/506439/>

Please add any other contact individuals who should be listed on the submitter page. Adding individuals at this step simply adds their name to the ClinVar Submitter page - it does not grant access to submit on behalf of the organization. Once the organization is approved, you can grant submitting privileges to other individuals by following directions here:

https://www.ncbi.nlm.nih.gov/clinvar/docs/submitter_groups/

Tab 3: Overview

Please review the information to make sure everything is correct. Once complete, you may submit. Once submitted, ClinVar will email the submitter confirmation of the organization registration and will email again once registration is complete (typically 1-3 days).

B. Submitting to ClinVar

After confirmation of the organization registration in ClinVar, you can navigate back to the ClinVar Submission Portal (<https://submit.ncbi.nlm.nih.gov/clinvar/>). If your organization is registered, it will appear under “Your Organizations” (see below for CDH1 VCEP).

1. To submit an excel spreadsheet, select “Upload new file submission” under the ‘Upload submission files’ section.

Your Organizations

New organization

ClinGen CDH1 Variant Curation Expert Panel

Edit this organization

Request deletion

Organization information

Address Bethesda, Maryland, United States 20892

Status **Processed**

Organization ID 506817

Single submission wizard

Use this wizard to submit a single variant interpretation to ClinVar. [Read more about using the wizard.](#)

If you are submitting a batch of variant interpretations (i.e. more than two), use the file upload option below for faster processing.

ClinVar single submission wizard

No data submitted yet.

Upload submission files

Use this button to upload a batch of variant interpretations as a file to ClinVar. [Read more about submission to ClinVar](#), including information about our submission spreadsheet templates.

Upload new file submission

Submission ID	Submission name	Status	Submitter name
SUB5047497	SUB5047497	Processed	Steven Harrison

Single SCV Update

Use this button to update a single submitted record (SCV).

Only the following fields may be updated: clinical significance, date last evaluated, comment on clinical significance, mode of inheritance, the interpreted condition, and citations for the submission. If you need to update a different field, please use the option for file submission above.

Single SCV Update

No data submitted yet.

2. 'Submission Information' tab

- ClinVar will auto-populate the Submission name field with a SUB ID#
- Under the 'Review status' section, please select "**Reviewed by expert panel**" (the submission portal always defaults to "----" so please remember to change this section otherwise your submission will not go live in ClinVar as an Expert Panel!).
- If your submission form was generated by the Variant Curation Interface, please select "**GRCh38**" under "Assembly name" (the submission portal defaults to no build).

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Submission Information

★ Submission name ?

SUB6304074

Is this data from a named study, like Framingham or CSER?
Most ClinVar submitters select 'no'.

☒ No
☐ Yes

★ Submission format ?

☒ Excel spreadsheet
☐ XML
☐ Tab-separated values
☐ Comma-separated values

Review status ?

☐ -----
☐ Reviewed by single submitter
☒ Reviewed by expert panel
☐ Practice guideline

Release status ?

☒ Public
☐ Hold until published

Do you need ClinVar accession numbers before processing is complete (e.g. for a publication)? ?
Note - we may contact you to clarify or correct data provided for the required fields before we can provide your accession numbers.

☒ No
☐ Yes

Citation ?

★ Assembly name ?

GRCh38

3. "Organization" tab

- a. Select "Single organization" and "No - I am submitting for my own organization"

ClinVar File Submission submission: SUB6304074
SUB6304074

Unfinished at the Organization step

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Organization

Submitting organization(s)

★ Is this submission from a single organization or from multiple organizations?

☒ Single organization

☐ Multiple organizations - all organizations listed below...

★ Are you submitting on behalf of another organization? ?

☒ No - I am submitting for my own organization

☐ Yes - I am submitting on behalf of another organization

Please note: Information about individuals, and the relationship of those individuals to your organization, are managed in the Organization submission form. Please review and submit any modifications there.

Save and continue

4. "Submission File" tab

- a. Click "Choose File" and select your completed excel file.
- b. After uploaded, your file name will appear

ClinVar File Submission submission: SUB6304074
SUB6304074

Unfinished at the Submission file step

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Submission Files

★ Please upload your submission file

Choose File No file chosen

Save and continue

ClinVar File Submission submission: SUB6304074
SUB6304074

Unfinished at the Submission file step

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Submission Files

★ Please upload your submission file

CDH1-Submission-2018-12-07-FINAL.xlsx	321.8 kB	9/17/2019 14:26	Delete
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Save and continue

5. 'Assertion Criteria' tab

- a. **If you are submitting with an assertion criteria document used on a previous submission**, ClinVar will recognize that the assertion criteria document name cited in your submission references a file already archived by ClinVar. In this case, ClinVar will alert the user that ClinVar already has the file, and you can select 'Save and continue'

ClinVar File Submission submission: SUB6304075

SUB6304075

🔄 **Unfinished** at the Assertion criteria step

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Assertion criteria

❗

- "Assertion criteria" in ClinVar is a document that describes the terms your organization uses to classify variants (e.g. pathogenic, uncertain significance, benign, or appropriate terms for other types of variation) and the criteria you require to assign a variant to each category. It may be a citation (PubMed ID, PubMedCentral ID, or DOI) or an electronic document (a Word document or PDF).
- Submitting this documentation, along with your evidence for classification or a public contact for your organization, means that your record will contribute to the review status of the ClinVar record. See [our documentation on assertion criteria](#) for more information.

We already have the following file(s) listed in your submission as assertion criteria:

❗

- [ClinGen_MyeloMalig_ACMG_Specifications_v1.pdf](#)

Save and continue

- b. If this is your first submission with a new assertion criteria document version (or your first submission ever for this VCEP), then ClinVar will recognize that the cited assertion document is not present in their archive. Please upload the new assertion criteria version by selecting "Choose File". Please note, the file name of the assertion criteria document needs to be an exact match with the file name cited in the submission form. (*Only use the assertion criteria document provided by ClinGen that includes the necessary headers and footers*).

ClinVar File Submission submission: SUB6304074
SUB6304074

Unfinished at the Assertion criteria step

1 SUBMISSION INFORMATION

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Assertion criteria

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- "Assertion criteria" in ClinVar is a document that describes the terms your organization uses to classify variants (e.g. pathogenic, uncertain significance, benign, or appropriate terms for other types of variation) and the criteria you require to assign a variant to each category. It may be a citation (PubMed ID, PubMedCentral ID, or DOI) or an electronic document (a Word document or PDF).
- Submitting this documentation, along with your evidence for classification or a public contact for your organization, means that your record will contribute to the review status of the ClinVar record. See [our documentation on assertion criteria](#) for more information.

Please upload the following files listed in your submission as assertion criteria:

★ Upload file "ClinGen_CDH1_ACMG_Specifications_v1.pdf"

Choose File

No file chosen


Save and continue

If neither option for Assertion Criteria tab seems correct, or if ClinVar does not recognize a previously cited assertion criteria document, please email Steven Harrison (steven@broadinstitute.org) to confirm that fields were completed correctly.

6. Overview Tab

- a. Please review all information (especially **Review Status** and **Assembly name**).
If correct, click Submit

ClinVar File Submission submission: SUB6325038
SUB6325038

 **Unfinished** at the Overview step

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Review & Submit

Submission information

Submission name	SUB6325038
Submission description	
Study name	
Review status	Reviewed by expert panel
Release status	Public
Accessions requested	Yes
Citation	
Assembly name	GRCh38
Submission file(s)	* ClinVar Submission CDH1_09172019.xlsx
Assertion criteria file(s)	

Submit

7. Post-submission

- a. After submitting, the submission will appear under ‘Upload submission files’ section.
- b. Once your submission goes live, the Status will change to Processed and a downloadable summary report will be available.

ClinGen CDH1 Variant Curation Expert Panel

[Edit this organization](#) [Request deletion](#)

Organization information

Address Bethesda, Maryland, United States 20892
Status **Processed**
Organization ID 506817

Single submission wizard

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Upload submission files

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Upload new file submission

Submission ID	Submission name	Status	Submitter name	Reports
SUB6308185	SUB6308185	Queued	Steven Harrison	No summary yet
SUB5047497	SUB5047497	Processed	Steven Harrison	SUB5047497_(100)_submitter_report_B.txt (100% complete)