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"

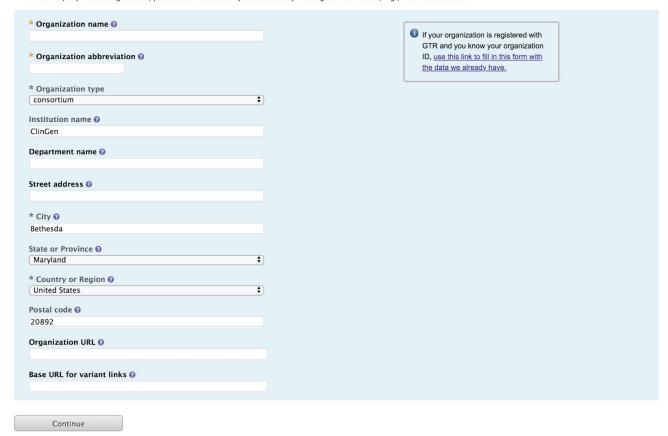
Tab 1: Organization Information



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: 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

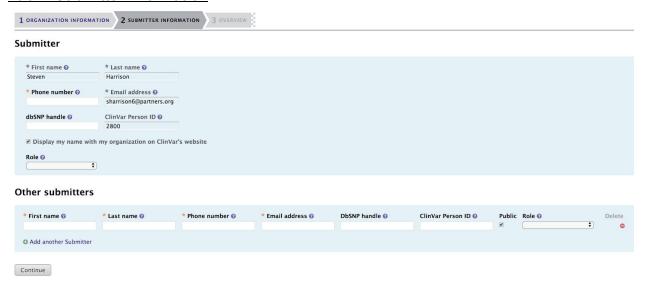
Postal code: 20892

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



The Submitter section is automatically completed for the person registering the organization in ClinVar. Please select your role and 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/

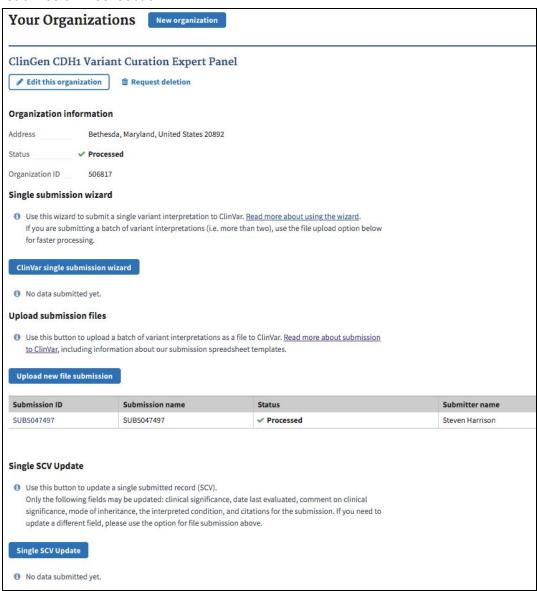
Tab 3: Overview

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

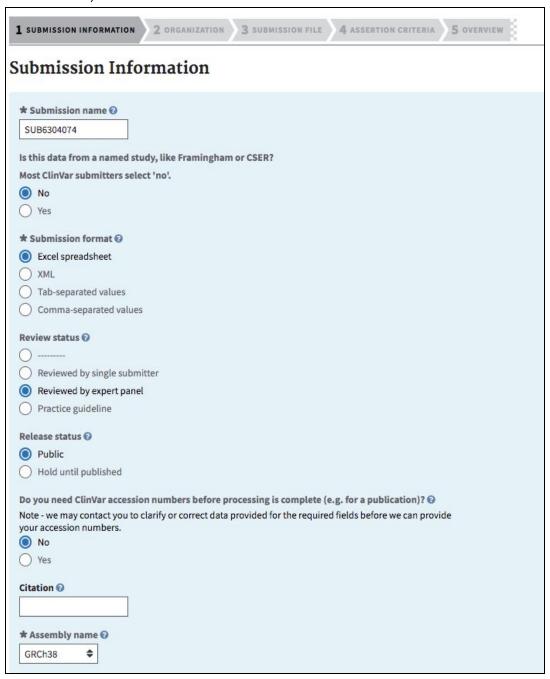
After confirmation of the organization registration in ClinVar, you can navigate back to the ClinVar Submission Portal (https://submit.ncbi.nlm.nih.gov/clinvar/). If your organization is registered, it will appear under "Your Organizations" (see below for CDH1 VCEP).

1. To submit an excel spreadsheet, select "Upload new file submission" under the 'Upload submission files' section.



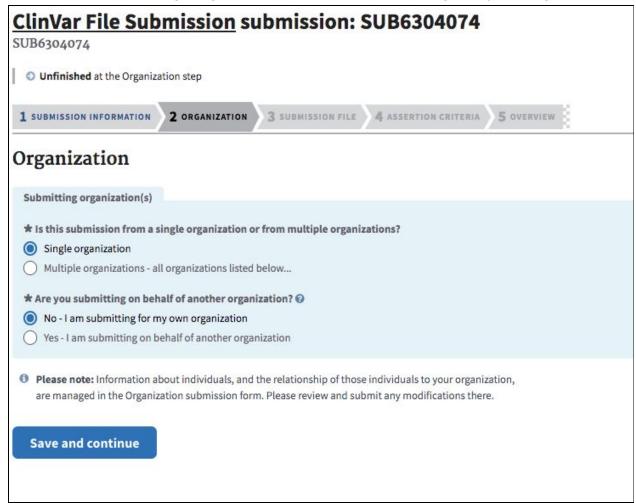
2. 'Submission Information' tab

- a. ClinVar will auto-populate the Submission name field with a SUB ID#
- b. 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!).
- c. If your submission form was generated by the Variant Curation Interface, please select "*GRCh38*" under "Assembly name" (the submission portal defaults to no build).



3. "Organization" tab

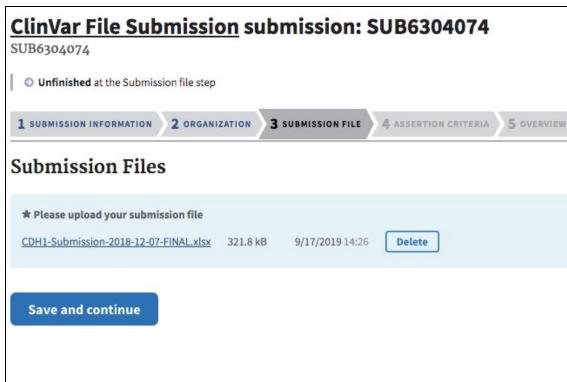
a. Select "Single organization" and "No - I am submitting for my own organization"



4. "Submission File" tab

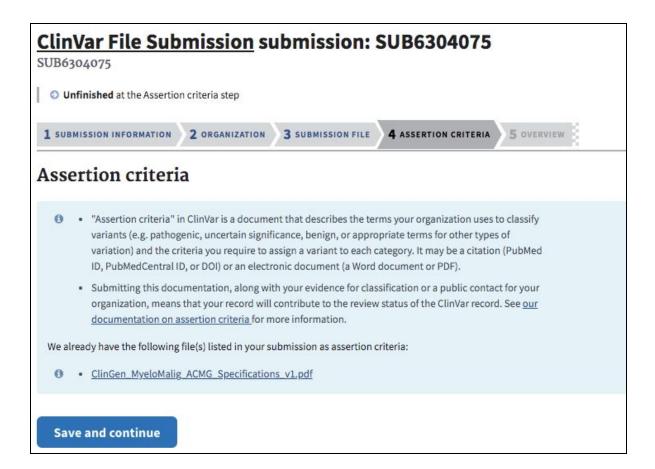
- a. Click "Choose File" and select your completed excel file.
- b. After uploaded, your file name will appear



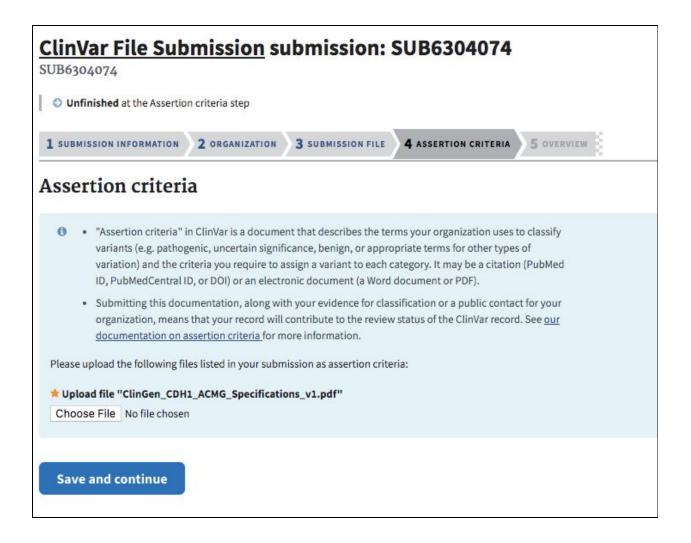


5. 'Assertion Criteria' tab

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'



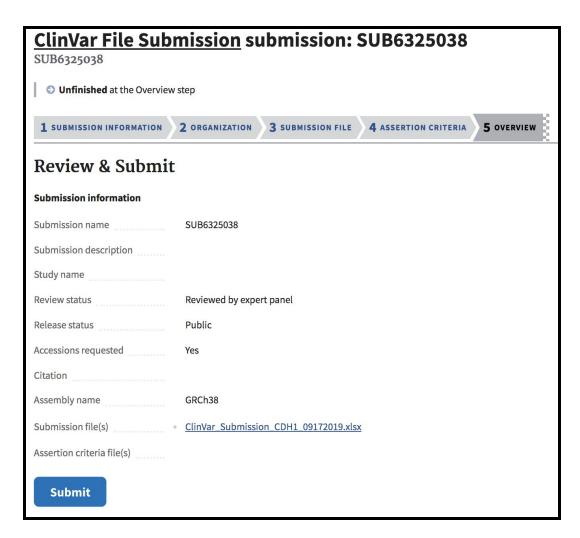
b. If this is your first submission with a new assertion criteria document version (or your first submission ever for this VCEP), then ClinVar will recognize that the cited assertion document is not present in their archive. Please upload the new assertion criteria version by selecting "Choose File". 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).



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 (<u>steven@broadinstitute.org</u>) to confirm that fields were completed correctly.

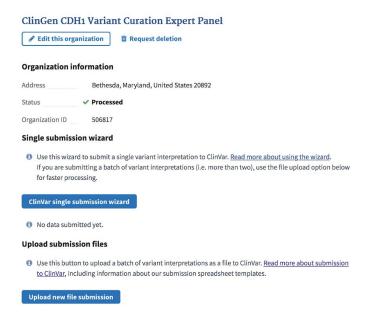
6. Overview Tab

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



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.



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)