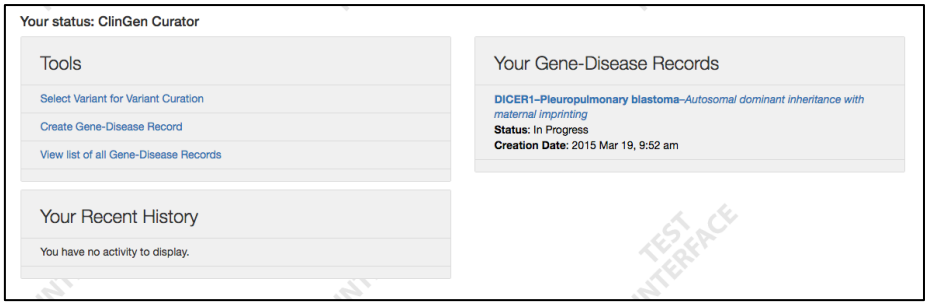
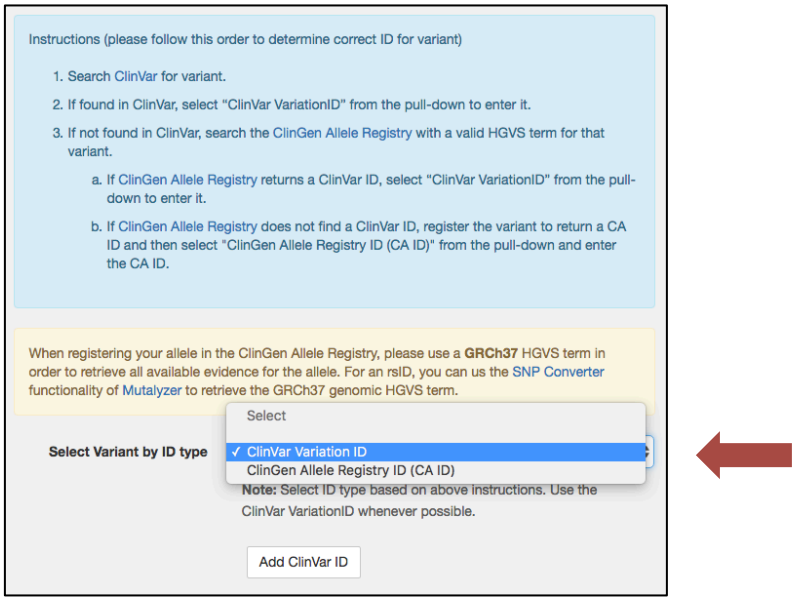
Variant Curation Interface – Quick Start Guide (July 2016)

1. Go to https://variant-curation-alpha1.demo.clinicalgenome.org/ and click the “Login” button. If you are registered but have not yet created a Persona account, you will be prompted to do so.

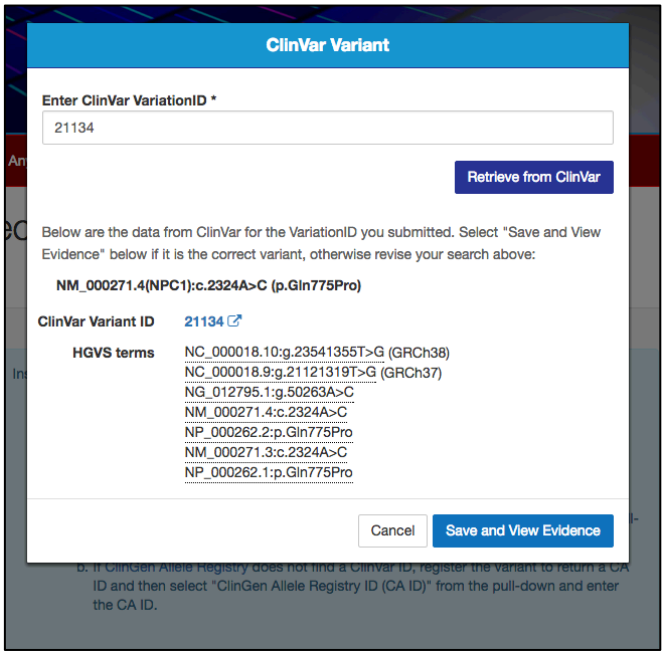
2. Select “Select Variant for Variant Curation” under the “Tools” Section



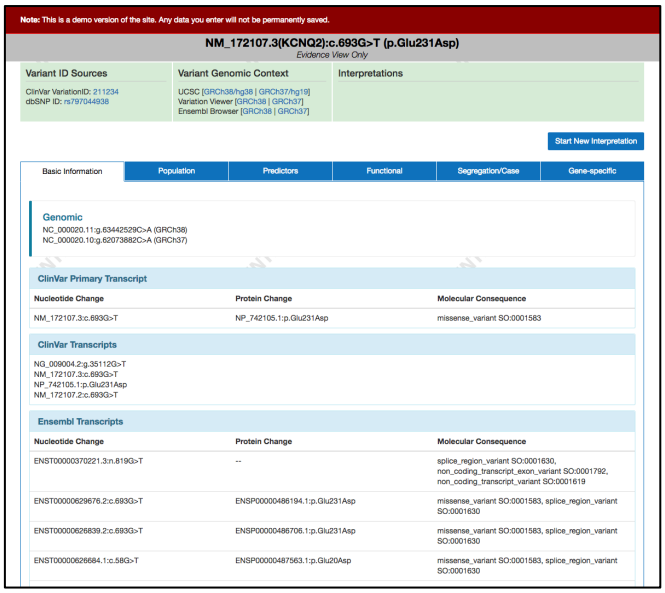
3. Read instructions carefully on how to select a variant, then choose whether you want to enter a ClinVar variant or a novel variant that you have registered with Baylor’s ClinGen Allele Registry. Select “Add ClinVar ID.”



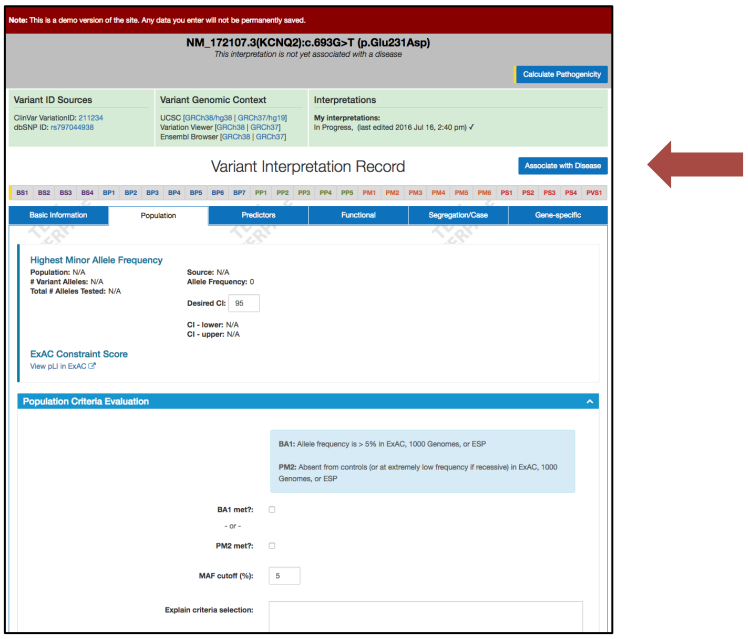
4. Type in the variant ID (ClinVar VariationID or CA ID, depending on selection), click “Retrieve from ClinVar” (or “Retrieve from ClinGen Allele Registry, if you have entered a CA ID). Once you are convinced the ID you have entered represents the correct variant, select “Save and View Evidence.”



5. You will now be in the “Evidence View” for your selected variant. You can see all evidence associated with the variant, clicking between the tabs. In this version, the Basic Information, Population, and Predictors tabs all contain associated evidence for the variant. After viewing the evidence, if you would like to begin an Interpretation, click on the “Start New Interpretation” button in the upper right hand corner.

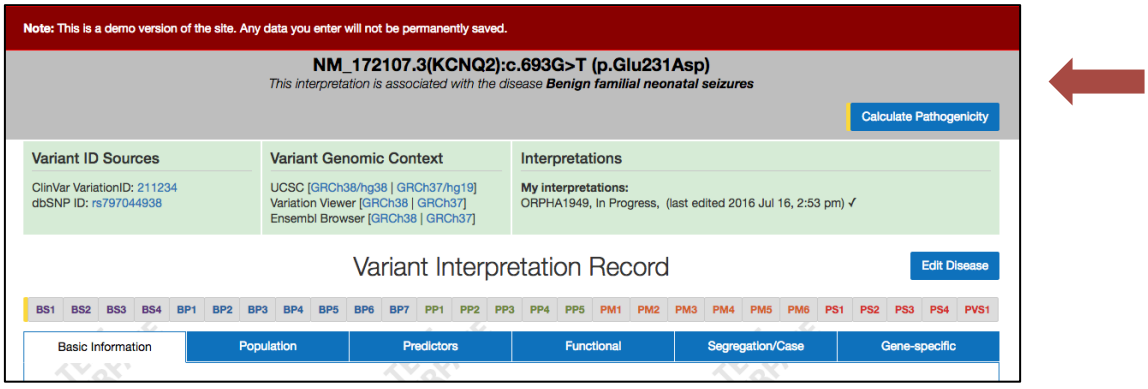


6. Once you are in Interpretation mode, you will see the option to evaluate relevant criteria appear on the tab. You can evaluate criteria, enter text that describes the reason for your selection, and save both by clicking the “Save” button. Note that some criteria are disease-dependent, and you will not be able to evaluate these criteria until you have associated your interpretation with a disease (see step 7, below).



Note that the “Calculate Pathogenicity” and criteria bar are not functional in this version (indicated by the yellow bar to their left).

7. To associate an Interpretation with a disease and be able to evaluate disease-dependent criteria, save any evaluations and click the “Associate with Disease” button (see above), found toward the upper right. Enter the desired Orphanet ID and click OK. Now all disease-independent criteria evaluation boxes will become active and you will see the disease term under the variant name in the gray title area. Your interpretation, along with the Orphanet ID will also display in the green bar under “My Interpretations.”



Note: Click on the home icon at the top of any page to return to the “Dashboard” – if you select the same variant and return to its View Evidence page, you will now see a “Continue Interpretation” button and be able to continue evaluating the same variant. The next version will include improved navigation.