Medical Genetics and Human Variation 2019 ClinVar Search Survey

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2019 ClinVar Survey, General

1. Which professional category best describes you? Please select only one.

- ○ Bioinformatics Professional
- ○ Clinical Testing Laboratory Staff
- ○ Computer Scientist / Software Developer
- ○ Educator
- ○ Genetic Counselor
- ○ Geneticist
- ○ Other (please specify)
- ○ Librarian / Information Specialist
- ○ Life Science Researcher
- ○ Other Healthcare Professional
- ○ Physician
- ○ Student
- ○ Patient / Family of Patient

2. Pick one category that best describes your organization

- ○ College or University
- ○ Commercial or Industry
- ○ Hospital / Clinical / Medical Practice
- ○ Non-Profit Organization
- ○ Government
- ○ Other (please specify)

3. How likely is it that you would recommend ClinVar to a friend or colleague?

Not at all likely — Extremely likely

0	1	2	3	4	5	6	7	8	9	10

4. Were you able to complete your work to your satisfaction today?

- ○ Yes
- ○ No
- ○ Not completed yet, but I expect to be able to complete
- ○ Unsure

*** 5. Which one of these best describes your primary goal for visiting us today?**

- ○ Working on and / or learning about submitting data
- ○ Understanding how variants are classified
- ○ Finding out who submits to ClinVar
- ○ Finding a specific variant
- ○ Finding all variants in a gene
- ○ Finding all variants submitted by a specific group
- ○ Finding all variants specific to a disease or phenotype
- ○ Getting updated information about my variant of interest
- ○ Downloading data about variants
- ○ Other (please specify)

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ClinVar Survey, Submissions

*** 6. Have you submitted to ClinVar before?**

- ○ Yes
- ○ No

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7. Which one of these best describes your primary goal for visiting us today?

- ○ Understand how to begin a submission
- ○ Prepare data to make a submission
- ○ Begin new submission
- ○ Complete a started submission
- ○ Determine the status of a previous submission
- ○ Assist a colleague with their submission
- ○ Other (please specify)

8. How easy were each of the following tasks? If you have made multiple submissions, please evaluate your most recent completed submission.

	Very difficult	Difficult	Neither easy nor difficult	Easy	Very easy	N/A
Complete NCBI login process	○	○	○	○	○	○
Begin new submission	○	○	○	○	○	○
Understand and gather required materials for submission	○	○	○	○	○	○
Follow instructions and enter data into forms	○	○	○	○	○	○
Upload files	○	○	○	○	○	○
Understand and correct errors	○	○	○	○	○	○
Understand submission, status, steps, and timeline	○	○	○	○	○	○

Other (please specify)

9. How useful would these enhancements to the ClinVar be in making submissions easier?

	Extremely useful	Very useful	Somewhat useful	Not so useful	Not at all useful	N/A
An API (Application Programming Interface) for submission	○	○	○	○	○	○
Ability to test files and get immediate error checks and recommended corrections	○	○	○	○	○	○
Faster responses about whether submitted data were acceptable or not and why	○	○	○	○	○	○
Diagram illustrating step-by-step submission process	○	○	○	○	○	○
Videos describing the step-by-step submission process	○	○	○	○	○	○
Examples of successful submissions	○	○	○	○	○	○
Required materials checklist	○	○	○	○	○	○
FAQ organized by submission steps	○	○	○	○	○	○
More instructions on template use for your situation	○	○	○	○	○	○

Other (please specify)

10. Indicate how difficult is it for you to provide us the following information?

	Very difficult	Difficult	Neither easy nor difficult	Easy	Very easy	N/A
A description of the variant (including HGVS expressions, chromosomal coordinates, and cytogenetic descriptions)	○	○	○	○	○	○
Condition for which the variant was interpreted	○	○	○	○	○	○
Interpretation of clinical significance	○	○	○	○	○	○
The method used to collect the data for the interpretation (e.g. clinical testing vs research)	○	○	○	○	○	○
Allele origin (e.g. germline vs somatic)	○	○	○	○	○	○
Affected status of individuals with the variant	○	○	○	○	○	○
Publications about the variant	○	○	○	○	○	○
The date the clinical significance of the variant was last evaluated	○	○	○	○	○	○
A summary explaining why the submitter interpreted the variant as they did	○	○	○	○	○	○
Evidence	○	○	○	○	○	○

11. If we were to require more data in a submission, would you be less likely to submit?

- ○ Yes
- ○ No
- ○ Unsure

12. What else can we do to make submission to ClinVar easier for you?

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7. How did you find out about ClinVar submissions?

- ( ) Google or other search engine
- ( ) NCBI (www.ncbi.nlm.nih.gov) or NIH home page (www.nih.gov) search
- ( ) Submission portal page (https://submit.ncbi.nlm.nih.gov/clinvar/)
- ( ) ClinVar home page (https://ncbi.nlm.nih.gov/clinvar/)
- ( ) Following a link from a publication
- ( ) Following a link from another website
- ( ) Colleague / word-of-mouth
- ( ) Email
- ( ) Other (please specify)

8. Which one of these best describes your primary goal for visiting us today?

- ( ) Understand how to begin a submission
- ( ) Prepare data to make a submission later
- ( ) Begin new submission
- ( ) Complete a started submission
- ( ) Assist a colleague with their submission
- ( ) Other (please specify)

9. How useful would these enhancements to the ClinVar be in making submissions easier?

	Extremely useful	Very useful	Somewhat useful	Not so useful	Not at all useful	N/A
An API (Application Programming Interface) for submission	( )	( )	( )	( )	( )	( )
Ability to test files and get immediate error checks and recommended corrections	( )	( )	( )	( )	( )	( )
Faster responses about whether submitted data were acceptable or not and why	( )	( )	( )	( )	( )	( )
Diagram illustrating step-by-step submission process	( )	( )	( )	( )	( )	( )
Videos describing the step-by-step submission process	( )	( )	( )	( )	( )	( )
Examples of successful submissions	( )	( )	( )	( )	( )	( )
Required materials checklist	( )	( )	( )	( )	( )	( )
FAQ organized by submission steps	( )	( )	( )	( )	( )	( )
More instructions on template use for your situation	( )	( )	( )	( )	( )	( )

Other (please specify)

10. What else can we do to make submission to ClinVar easier for you?

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6. How would you like to be able to search in ClinVar? Choose all that apply.

- [ ] By HGVS expressions
- [ ] By gene symbols
- [ ] By disease or phenotype
- [ ] By ClinVar Variation ID
- [ ] By rs number
- [ ] By a region on a chromosome
- [ ] By submitter
- [ ] Other (please specify)

7. How can we help you find the information you need more efficiently?

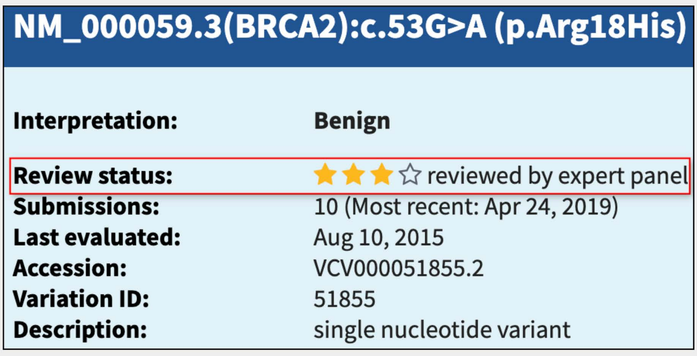
8. Please rank the following in order of importance to you.

- Access to as many variants as possible, even if they have limited information
- More variants that have complete or nearly complete information
- More variants submitted from known/trusted sources, even if the information is minimal
- More rare variants that are unlikely to be found elsewhere

9. Do you understand what the review status for a variant means as represented by the number of stars on the web and in the image below ? Do you find it useful?

- ( ) I understand it and find it useful
- ( ) I understand it but do not find it useful
- ( ) I do not understand the review status
- ( ) Unsure

Comments



10. Indicate how useful each of the items listed below is to you.

	Extremely useful	Very useful	Somewhat useful	Not so useful	Not at all useful	N/A
A description of the variant (including HGVS expressions, chromosomal coordinates, and cytogenetic descriptions)	( )	( )	( )	( )	( )	( )
Condition for which the variant was interpreted	( )	( )	( )	( )	( )	( )
Interpretation of clinical significance	( )	( )	( )	( )	( )	( )
The method used to collect the data for the interpretation (e.g., clinical testing vs research)	( )	( )	( )	( )	( )	( )
Allele origin (e.g., germline vs somatic)	( )	( )	( )	( )	( )	( )
Affected status of individuals with the variant	( )	( )	( )	( )	( )	( )
Publications about the variant	( )	( )	( )	( )	( )	( )
The date the clinical significance of the variant was last evaluated	( )	( )	( )	( )	( )	( )
A summary explaining why the submitter interpreted the variant as they did	( )	( )	( )	( )	( )	( )
Evidence	( )	( )	( )	( )	( )	( )

11. Indicate how useful the following features would be to you if you needed to download a large number of variants.

	Extremely useful	Very useful	Somewhat useful	Not so useful	Not at all useful	N/A
More help on how I can set up my tools to bulk download variant information	( )	( )	( )	( )	( )	( )
Preview what I may be downloading before I start	( )	( )	( )	( )	( )	( )
Download variants by gene	( )	( )	( )	( )	( )	( )
Download variants by region	( )	( )	( )	( )	( )	( )
Download variants by disease	( )	( )	( )	( )	( )	( )
Download only those variants that have changed since the last time I downloaded	( )	( )	( )	( )	( )	( )
Download variants by population (e.g., ethnic, geographic region, etc.)	( )	( )	( )	( )	( )	( )

Other (please specify)

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ClinVar, Other resources

12. Indicate the relative usefulness of the resources below that you use for similar purposes as ClinVar. (For each resource, choose one)

	Extremely useful	Very useful	Somewhat useful	Not so useful	Not at all useful	N/A
ClinVar	○	○	○	○	○	○
LOVD	○	○	○	○	○	○
Varsome	○	○	○	○	○	○
ClinGen	○	○	○	○	○	○
dbSNP	○	○	○	○	○	○

Other (please specify)

13. Considering the resources listed above, what does ClinVar do better? What do the other resources do better?

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2019 ClinVar Survey, More Feedback

Please write to us at suggest@ncbi.nlm.nih.gov if you would like to provide additional feedback on NCBI's Medical Genetics and Human Variation resources.

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