

Medical Genetics and Human Variation 2019 ClinVar Search Survey

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

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

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
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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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*** 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	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Begin new submission	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Understand and gather required materials for submission	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Follow instructions and enter data into forms	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Upload files	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Understand and correct errors	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Understand submission, status, steps, and timeline	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Ability to test files and get immediate error checks and recommended corrections	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Faster responses about whether submitted data were acceptable or not and why	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Diagram illustrating step-by-step submission process	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Videos describing the step-by-step submission process	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Examples of successful submissions	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Required materials checklist	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
FAQ organized by submission steps	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
More instructions on template use for your situation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Condition for which the variant was interpreted	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Interpretation of clinical significance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The method used to collect the data for the interpretation (e.g. clinical testing vs research)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Allele origin (e.g. germline vs somatic)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Affected status of individuals with the variant	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Publications about the variant	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The date the clinical significance of the variant was last evaluated	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
A summary explaining why the submitter interpreted the variant as they did	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Evidence	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Ability to test files and get immediate error checks and recommended corrections	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Faster responses about whether submitted data were acceptable or not and why	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Diagram illustrating step-by-step submission process	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Videos describing the step-by-step submission process	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Examples of successful submissions	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Required materials checklist	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
FAQ organized by submission steps	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
More instructions on template use for your situation	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Other (please specify)

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

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.

⋮	<input type="text"/>	Access to as many variants as possible, even if they have limited information
⋮	<input type="text"/>	More variants that have complete or nearly complete information
⋮	<input type="text"/>	More variants submitted from known/trusted sources, even if the information is minimal
⋮	<input type="text"/>	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

NM_000059.3(BRCA2):c.53G>A (p.Arg18His)**Interpretation:****Benign****Review status:**

★★★★☆ reviewed by expert panel

Submissions:

10 (Most recent: Apr 24, 2019)

Last evaluated:

Aug 10, 2015

Accession:

VCV000051855.2

Variation ID:

51855

Description:

single nucleotide variant

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)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Condition for which the variant was interpreted	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Interpretation of clinical significance	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The method used to collect the data for the interpretation (e.g., clinical testing vs research)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Allele origin (e.g., germline vs somatic)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Affected status of individuals with the variant	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Publications about the variant	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
The date the clinical significance of the variant was last evaluated	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
A summary explaining why the submitter interpreted the variant as they did	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Evidence	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Preview what I may be downloading before I start	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Download variants by gene	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Download variants by region	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Download variants by disease	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Download only those variants that have changed since the last time I downloaded	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Download variants by population (e.g., ethnic, geographic region, etc.)	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Other (please specify)	<input type="text"/>					

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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	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
LOVD	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Varsome	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
ClinGen	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
dbSNP	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

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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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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