ClinVar Submissions Enhancements Survey 2024

Start of Block: Intro Question Block

OMB Control Number: **0925-0648**
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Public reporting burden for this collection of information is estimated to average **10** minutes per response, including the time for reviewing instructions, searching existing data sources, gathering and maintaining the data needed, and completing and reviewing the collection of information. An agency may not conduct or sponsor, and a person is not required to respond to, a collection of information unless it displays a current valid OMB control number. Send comments regarding this burden estimate or any other aspect of this collection of information, including suggestions for reducing this burden, to NIH, Project Clearance Branch, 6705 Rockledge Drive, MSC 7974, Bethesda, MD 20892-7974, ATTN: PRA (0925-0648). Do not return the completed form to this address.

All questions are optional, and you may exit the survey at any time.

Please select one professional category that describes you best.

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

Please select one type that describes your organization best.

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

How likely are you to recommend ClinVar to a friend or colleague?

Not at all likely										Extremely likely
0	1	2	3	4	5	6	7	8	9	10

If you have had difficulty understanding error messages related to your ClinVar submission, please describe the types of errors and what additional / different information would have been helpful.

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If you have had difficulty getting answers to your questions about submission to ClinVar, please describe the type of questions that you had and include anything you'd like to tell us about your experience.

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Do you submit copy number variants (CNVs) to ClinVar?

- ○ Yes
- ○ No

End of Block: Intro Question Block

Start of Block: COPY NUMBER VARIATION (CNV) DATA ENHANCEMENTS

COPY NUMBER VARIATION (CNV) DATA ENHANCEMENTS

Do you agree with how ClinVar proposes standardizing the definition of CNVs below?

- CNVs and structural variants are considered synonymous
- CNVs are any variants that are larger than 1000 base pairs (bp)
- CNVs may span one or more genes
- CNVs include exon deletions and / or duplications, but we will provide a way to differentiate

- ○ Yes
- ○ Maybe
- ○ No

Display This Question:

If Do you agree with how ClinVar proposes standardizing the definition of CNVs below? CNVs and struc... = Maybe

Or Do you agree with how ClinVar proposes standardizing the definition of CNVs below? CNVs and struc... = No

Please elaborate on how you disagree with the ClinVar's proposed definition of CNVs. Feel free to include a source link for a definition that you use.

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Please rank the following data types from easiest to submit for CNVs to most difficult to submit for CNVs.

_____ Genomic location
_____ Exon numbers for exon deletions/duplications
_____ Variant type, e.g. copy number gain/loss, deletion, duplication
_____ Reference and observed copy number
_____ The disease for the classification
_____ The patient's phenotype
_____ Citations supporting the classification
_____ Free text comment explaining the classification
_____ Other (please specify)

What is the most important feature we can provide to improve the submission of CNVs?

- ○ Provide better instructions specific for CNVs
- ○ Provide a submission form specific for CNVs
- ○ Integrate submission with the software I use to evaluate CNVs (please specify the software) _____
- ○ Other (please specify) _____

End of Block: COPY NUMBER VARIATION (CNV) DATA ENHANCEMENTS

Start of Block: Functional Data Enhancements Yes/No

Many laboratories are developing functional assays to assess the impact of a variant on the transcript or protein. The functional data that are produced by these assays are critical to classification of variants, particularly Variants of Uncertain Significance (VUS). Functional data can be submitted to ClinVar today, and we are interested in how we can improve its representation.

Would you like to answer questions related to enhancements to functional data for variants in ClinVar?

- ○ Yes
- ○ No

End of Block: Functional Data Enhancements Yes/No

Start of Block: FUNCTIONAL DATA ENHANCEMENTS

FUNCTIONAL DATA ENHANCEMENTS

How important is it for ClinVar to support submission of functional data for variants?

- ○ Not at all important
- ○ Slightly important
- ○ Moderately important
- ○ Very important
- ○ Extremely important

Please explain your answer to the previous question.

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Should functional data be submitted on its own, or should it always be provided in support of a germline or somatic classification?

- ○ Functional data should be submitted on its own
- ○ Functional data should always be provided in support of a germline or somatic classification
- ○ Functional data could be provided both on its own and/or in support of a germline or somatic classification

Please explain your response to the previous question.

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Display This Question:

If Should functional data be submitted on its own, or should it always be provided in support of a g... = Functional data should be submitted on its own

Or Should functional data be submitted on its own, or should it always be provided in support of a g... = Functional data could be provided both on its own and/or in support of a germline or somatic classification

If functional data is submitted on its own, would you want to know the strength of functional evidence (i.e., high quality, low quality, etc.) using pre-defined criteria?

- ○ Yes
- ○ Sometimes
- ○ No

Display This Question:

If If functional data is submitted on its own, would you want to know the strength of functional evi... = Yes

Or If functional data is submitted on its own, would you want to know the strength of functional evi... = Sometimes

Please explain your response to the previous question.

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Please rank the following items related to functional data for variants from most important to least important to you.

_____ A description of the assay
_____ The scoring system for the assay
_____ The disease or drug response that the assay informs
_____ The result of the assay for a specific variant
_____ A citation describing the assay
_____ Links to other databases with additional information about the assay or the result
_____ Other (please specify)

End of Block: FUNCTIONAL DATA ENHANCEMENTS

Start of Block: Pharma Enhancements Yes/No

Pharmacogenomic variants affect how an individual responds to certain drugs. Results from pharmacogenomic testing may be submitted to ClinVar today, and we are interested in how we can improve its representation.

Would you like to answer questions about enhancements related to pharmacogenomic variants in ClinVar?

- ○ Yes
- ○ No

End of Block: Pharma Enhancements Yes/No

Start of Block: PHARMACOGENOMIC (Pharma) DATA ENHANCEMENTS

PHARMACOGENOMIC (Pharma) DATA ENHANCEMENTS

How important is it for ClinVar to support variant classifications specific to pharmacogenomic variants?

- ○ Not at all important
- ○ Slightly important
- ○ Moderately important
- ○ Very important
- ○ Extremely important

Please explain your answer to the previous question.

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Please rank the following aspects of pharmacogenomic data from most importance to least important to you

_____ Standard terms for pharmacogenomic classifications
_____ Description of a single variant (e.g., a single nucleotide variant (SNV) classified for drug efficacy and toxicity
_____ Description of a haplotype or genotype classified for drug efficacy and toxicity
_____ Pharmacogenomic literature
_____ Study parameters such as study size, ethnicity, allele frequency and statistics (e.g., P value and odds ratio)
_____ Functional data supporting pharmacogenomic classifications
_____ Links to other databases with additional information about pharmacogenomic variants
_____ Other (please specify)

End of Block: PHARMACOGENOMIC (Pharma) DATA ENHANCEMENTS

Start of Block: Survey Wrap Up

Please share what ClinVar means to you.

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What is the one word that comes to mind when you think of ClinVar?

_____

Please enter your contact information if you would be willing to share additional feedback about ClinVar with us.

Name

_____

Email Address

_____

End of Block: Survey Wrap Up