MedGen Survey

Start of Block: Default Question Block

OMB Control Number: **0925-0648**
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Public reporting burden for this collection of information is estimated to average **8** 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.

- ○ Laboratory staff
- ○ Genetic counselor
- ○ Physician
- ○ Nurse
- ○ Other healthcare professional
- ○ Bioinformatics professional
- ○ Researcher
- ○ Patient or family
- ○ Educator
- ○ Student
- ○ Other (please specify) _____

Please select one type that describes your organization best.

- ○ Hospital / Clinic / Medical Practice
- ○ Laboratory
- ○ Payer (insurance company)
- ○ College or University
- ○ Government
- ○ Other (please specify) _____

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

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

Did you find what you were looking for?

- ○ Yes
- ○ No
- ○ Partially

Please tell us why you were not able to find what you were looking for.

_____

_____

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How often do you typically use MedGen?

- ○ Daily
- ○ Once a week
- ○ Once a month
- ○ Once in 6 months
- ○ Once a year
- ○ This is my first visit

What are you looking for in MedGen today? Please select all that apply.

- ☐ Find information about a specific disease / condition
- ☐ Find identifiers for a disease (e.g., UMLS, HPO, Mondo, OMIM, Orphanet)
- ☐ Find diseases caused by a gene(s)
- ☐ Find diseases with a set of findings/clinical features
- ☐ Find specific GeneReviews articles
- ☐ Other (please specify) _____

Where does your MedGen journey typically start?

- ○ I am a clinician and am looking for a disease/condition in a lab report
- ○ I am a GTR / ClinVar submitter and I am looking for the ID for the disease/phenotype I want to submit
- ○ I/My family member was diagnosed with a genetic disease and I am looking for more information
- ○ Followed link from NLM webpage
- ○ Search engine result (Google, Bing, etc.)
- ○ Other (please specify) _____

Please rank the terms you use to search for a disease/condition within a resource like MedGen from most used to least used.

_____ Disease name
_____ Disease family name
_____ Gene
_____ UMLS CUI (ID)
_____ HPO ID
_____ MONDO ID
_____ Orphanet ID
_____ OMIM ID
_____ SNOWMED ID
_____ LOINC Code
_____ MeSH Term
_____ GARD ID
_____ MedGen ID
_____ Other (please specify)

Please rank the information that you typically review for a disease/condition from most important to least important.

_____ Disease characteristics
_____ Gene-disease relationships
_____ Clinical features
_____ Disease attributes like prevalence and age of onset
_____ Disease hierarchy
_____ Professional guidelines
_____ Clinical studies
_____ Latest research articles on diagnosis, therapy, or prognosis
_____ Genetic variation information
_____ Links to consumer resources like Genetic Alliance, Medlineplus Genetics
_____ Other, (please specify)

Please rank the usefulness of these currently unavailable features from most important to least important.

_____ List of disease indications for a drug in drug response records
_____ Search results displayed as hierarchies to clearly see disease families
_____ Disease prevalence and age of onset
_____ Other, (please specify)

Tell us about anything else that is important to you, that currently is not provided in MedGen or is hard to find.

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When you search for a disease, would you like to see related records? (Some examples include parent terms and related sub-types, all phenotypes with the same causative gene, pharmacogenetic drug class members or drugs metabolized by the same gene.)

- ○ Yes
- ○ No
- ○ Not sure

Please rank the type of related records you would like to see when searching for a disease from most important to least important.

_____ No, show me only what matches my search.
_____ Related disease(s) (by overlapping phenotypes)
_____ Related diseases by gene(s) (same gene causing different disease)
_____ Diseases related by cellular process/pathway (e.g., Autophagy defects, RASopathies, Laminopathies, Ciliopathies, etc.)
_____ Related drug responses (diseases, genes)
_____ Other (please specify)

How would you like the MedGen information display to be organized? Please select all that apply.

- ☐ By disease family / condition category (e.g., cardiovascular, musculoskeletal, etc.)
- ☐ By gene
- ☐ By Chromosomes & mtDNA
- ☐ By disease attributes (inheritance, growth, etc.)
- ☐ Other (please specify)

_____

MedGen recently changed its display of practice guidelines for a disease. Before, MedGen provided links to a select few guidelines manually curated for a small number of diseases; now nearly all diseases have a curated link to relevant guidelines in PubMed. Is this change useful to you?

- ○ Yes
- ○ No
- ○ Partially, please specify _____
- ○ I did not notice a change

Other than MedGen, what other medical genetics resources do you use?

_____

What do you like or dislike about the features available in other medical genetics resources?

_____

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Do you have any other suggestions for improving MedGen?

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

Name

_____

Email Address

_____

End of Block: Default Question Block