#### Medical Genetics and Variation 2020 Multi-Product Survey

OMB Control Number: 0925-0648 Expiration Date: 05/31/2021

Public reporting burden for this collection of information is estimated to average 15 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.

Next



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

Bioinformatics Professiona     Computer Scientist / Software			~	Information Speci ce Researcher	alist	
Educator			9	Ithcare Profession	nal	
Genetic Counselor Geneticist			Physician     Student			
Laboratory Staff			2	amily of Patient		
Other (please specify)						
2. Pick one category that I	best describes	your organizatio	on			
Commercial or Industry						
Commercial or Industry     Hospital / Clinical / Medical	l Practice					
Non-Profit Organization						
Government						
Other (please specify)		1				
	V-791111					
B. How likely is it that you Not at all likely	would recomm	nend this resource	ce to a friend or	colleague?		Extremely likely
0 1	2 3	4	5 6	7	8 9	10
. Were you able to compl	ete your work	to your satisfact	ion today?			
Yes						
) No						
Not completed yet, but I ex Unsure	pect to be able to	complete				
Find / access research stu Find / access genetic tests Submit data Download data Other (please specify)						
Other (picase speeliy)		-,				
If you download data w	that are your re	acono 2 Places a	oloot all that an	nhe		
6. If you download data, was Integrate into an open sou			elect all that ap	piy.		
Integrate into my own pipel	line					
One-time download						
Other (please specify)						
		-				
Cloud computing Would not use Other (please specify)  B. How useful would each	of these variar	nt visualizations	/ graphs be?			
Variations in a gene	Extremely useful	Very useful	Moderately useful	Not as useful	Not at all useful	N/A
Variations in a gene		^	~		~	
organized by molecular consequence	O	0	0	O	0	O
Variations in a region  Variations related to a	0	0	0	0	0	0
disease / phenotype	0	0	0	0	0	0
Clinically significant variations in a gene	0	0	0	0	0	0
Clinically significant variations in a region	O	0	0	0	0	0
Variations annotated with research / publications	0	0	0	0	0	0
Variations annotated with	~					
RefSeq functional elements	0	0	0	0	.0	0
Other (please specify)						
9. What types of searches	would be usef	ul to you in this	resource?			
		-10				
10. Which resources do yo	ou use frequen	tly?				
dbGaP						
dbSNP	0.70					
Genetic Testing Registry (0	SIR)					
MedGen						
ClinVar						
Variation Viewer  Medical Genetics Summari	ies					
GeneReviews						
PheGenl						
OSIRIS						
1. Would you be willing to	o answer ques	tions about dbGa	aP today?			
Yes						
No						
		Prev	Next			

Prev Next

	Very easy	Moderately easy	Neither easy nor difficult	Moderately difficult	Very difficult	N/A
Finding the data	0	0	0	0	0	0
Applying for access	0	0	0	0	0	0
Downloading public FTP data	0	0	0	0	0	0
Downloading controlled- access data	0	0	0	0	0	0
Use of phenotype data in the format provided	0	0	0		0	0
Use of genotype data in the format provided	0	0	0	0	0	0
Determining the validity / accuracy of data	0	0	0	0	0	0
Using data across multiple studies	0	0	0	0	0	0
Getting answers to your questions	0	0	0	0	0	0
ther (please specify)						
ther (please specify)  3. If we provided a dbGa	P API for data	a access (public a	and controlled-ac	cess), what typ	es of data request	s would yo
	P API for data	a access (public a	ind controlled-ac	cess), what typ	es <mark>of data reques</mark> t	s would yo
3. If we provided a dbGa	P API for data	a access (public a	ind controlled-ac	cess), what typ	es of data request	s would yo
3. If we provided a dbGa	P API for data	a access (public a	ind controlled-ac	cess), what typ	es <mark>of data reques</mark> t	s would yo
3. If we provided a dbGa					es of data request	s would yo
3. If we provided a dbGa se that API for?					es of data request	s would yo
3. If we provided a dbGa se that API for? 4. Which of the following					es of data request	s would yo
3. If we provided a dbGase that API for?  4. Which of the following a lonly submit data	g is the best o				es of data request	s would yo

### 15. How easy are each of these tasks during a dbGaP submission? Niether easy nor Moderately Very difficult Moderately easy difficult difficult N/A Very easy Register study Provide consents and Data Use Certification (DUC) Submit study summary descriptions Submit phenotype data Submit genotype data Understand what was already uploaded by other submitters of the study Obtain a copy of the latest version of your uploaded data Get answers to your questions Other (please specify) 16. How useful would each of these be in your dbGaP submission? Extremely useful Very useful Somewhat useful Not so useful Not at all useful N/A Use of an API (Application Programming Interface) Simpler submission process and/or file format More help material Help during registration process Help during file preparation Better communication about submission status Other (please specify) Prev Next

### dbGaP last page

7. Do you have any other	
9 Would you be willing t	anguer questions shout dheND today?
~	answer questions about dbSNP today?
8. Would you be willing to Yes	answer questions about dbSNP today?
Yes	answer questions about dbSNP today?

### dbSNP General

### 19. How easy are each of these tasks in dbSNP?

	Very easy	Moderately easy	Neither easy nor difficult	Moderately difficult	Very difficult
Finding the data	0	0	0	0	0
Downloading data	0	0	0	0	0
Use of data in the format provided	0	0	0	0	0
Determining the validity / accuracy of the data	0	0	0	0	0
Other (please specify)					
		)			
20. What would make dbS	NP data downlo	ad easier for you (e.g	g., API, different f	ormat, etc.)?	
21. Do you submit data to	dbSNP?				
Yes					
○ No					
		Prev	Next		

	Very easy	Moderately easy	Neither easy / nor Difficult	Moderately difficult	Very difficult	N/A
Prepare submission data	0	0	0	0	0	C
Submit data using the submission portal	0	0	0	0	0	C
Submit data using a file	0	0	0	0	0	C
Submit via FTP	0	0	0	0	0	C
Get answers to your questions	0	0	0	0	0	C
ther (please specify)						
3. How useful would ea	ch of these be i	in a dbSNP subr	nission ? Somewhat Useful	Not so Useful	Not at all Useful	N//
API (Application Programming Interface)	0	0	0	0	0	С
VCF format	0	0	0	0	0	C
BED format	0	0	0	0	0	
Tab delimited format	0	0	0	0	0	(
Simpler submission process and/or file format	0	0	0	0	0	
More help material	0	0	0	0	0	(
Help during file preparation	0	0	0	0	0	C
Better communication about submission status	0	0	0	0	0	Ç
ther (please specify)						
4. Indicate your level of	f interest in sub Extremely Interested	mitting any of th	nese other types Moderately interested	of data to us? Slightly Interested	Not at all Interested	N/
Genotype allele frequency	0	0	0	0	0	C
Haplotypes	0	0	0	0	0	(
Other annotations	0	0	0	0	0	(
ther (please specify)						

# dbSNP last page

25. Do you have any other su			
26 Would you be willing to a		the Constin Testing Design	- /CTD
26. Would you be willing to a	nswer some questions about	the Genetic lesting Regist	ry (G IR
Yes	nswer some questions about	the Genetic Testing Regist	ry (G IK
	nswer some questions about	the Genetic Testing Regist	ary (GTK
Yes	nswer some questions about	the Genetic Testing Regist	ary (GTK

	Very Easy	Moderately Easy	Neither Easy nor Difficult	Moderately Difficult	Very Difficult	N/A
Finding the data					O	
Comparing tests	0	0	0	0	0	
Downloading data		0	0	0	0	
Use of data in the format provided	0	0	0	0	0	C
Determining the validity / accuracy of data	0	0	0	0	0	С
Other (please specify)						
28. How useful would ea						
Direct to	Extremely useful	Very useful	Somewhat useful	Not so useful	Not at all useful	N/A
Direct to consumer tests	0	0	O	.0	0	
Exome tests with all the test targets and coverage information	0	0	0	0	0	C
Pathogen genetic tests	0	0	0	0	0	C
Non-disease related tests (e.g., nutrition, sports aptitude, etc.)	0	0	0	0	0	C
Other (please specify)						
29. How useful would ea	ach of these be i	n GTR?				
	Extremely useful	Very useful	Somewhat useful	No so useful	Not at all useful	N/A
		100		7.7		(60)
Lab staff credentials	0	0	0	0		
Lab staff credentials  Test turn-around time	0	0	0	0	Ö	C
	0	0	0	0	0	C
Test turn-around time	0	0	0	0	0	000
Test turn-around time Test price Test codes		0 0 0		0		000
Test turn-around time Test price		0000				000
Test turn-around time Test price Test codes Other (please specify)	O O O					000
Test turn-around time Test price Test codes Other (please specify)	∩ ○ ○					CCC
Test turn-around time Test price Test codes Other (please specify)	TR?					
Test turn-around time Test price Test codes Other (please specify)  30. Do you submit to G1	TR?	Prev	Next			

### **GTR Submission**

	Very <mark>eas</mark> y	Moderately easy	Neither easy nor difficult	Moderately difficult	Very difficult	N/A
Prepare submission data	0	0	0	0	0	0
Use Excel file	0	0	0	0	0	0
Submit via submission portal	0	0	0	0	0	0
Get answers to your questions	0	0	0	0	0	0
Other (please specify)						
2. How useful would e	ach of these be i		sion? Somewhat useful	Not so useful	Not at all useful	N/A
An API (Application	Extremely useful	Very useful	Somewhat userui	Not so userui	Not at all useful	IVA
Programming Interface)	0	0	0	.0	0	0
Excel format	0	0	0	0	0	0
XML format	0	0	0	0	0	0
Simpler submission	0	0	0	0	0	0
process and/or file format				-		
	0	0	0	0	0	0
More help materials	0	0	0	0	0	0
More help materials Help during file	0	0	0	0	0	0
More help materials  Help during file preparation  Better communication	0	0	0	0		0

# GTR last page

4. Would you be willing to	answer some questions about dbVar?
4. Would you be willing to Yes	answer some questions about dbVar?
	answer some questions about dbVar?
Yes	answer some questions about dbVar?

	Very easy	Moderately easy	Neither easy / nor difficult	Moderately difficult	Very difficult	N/A
Finding the data	0	0	0	0	0	C
Downloading data	0	0	0	0	0	C
Use of data in the format provided	0	0	0	0	0	С
Determining the validity / accuracy of data	0	0	0	0	0	C
Other (please specify)						
Other (please specify)						
other (please specify) 6. What would make db	Var data down	nload easier for yo	ou (e.g., API, diffei	rent format, etc	:.)?	
	Var data down	nload easier for yo	ou (e.g., API, diffei	rent format, etc	:.)?	
6. What would make db		nload easier for yo	ou (e.g., API, diffei	rent format, etc	:.)?	
6. What would make db		nload easier for yo	ou (e.g., API, diffei	rent format, etc	:.)?	
6. What would make db		nload easier for yo	ou (e.g., API, diffei	rent format, etc	:.)?	
6. What would make db		nload easier for yo	ou (e.g., API, diffei	rent format, etc	:.)?	

	Very Easy	Moderately Easy	Niether Easy / Nor Difficult	Moderately Difficult	Very Difficult	N/A
Prepare submission data	0	0	0	0	0	0
Submit via FTP	0	0	0	0	0	0
Use Excel file	0	0	0	0	0	0
Get answers to your questions	0	0	0	0	0	0
ther (please specify)						
9. How useful would ea	ch of these be in			useful	Not so useful	Not at all usefu
API (Application Programming Interface) for submission	0	0	0		0	0
VCF format	0	0	0		0	0
BED format	0	0	0		0	0
Tab delimited format for submission	0	0	0		0	0
Simpler submission process and/or file format	0	0	0		0	0
More help materials	0	0	0		0	0
Help during file preparation	0	0	0		0	0
Better communication about submission status	0	0	0		0	0
ther (please specify)						
ther (piease speelig)						

# dbVar last page

0. Do you have any other sug	
~	swer some questions about MedGen?
Yes	swer some questions about MedGen?
	swer some questions about MedGen?
Yes	swer some questions about MedGen?

		(25) 12 MAN	Neither easy nor	Moderately	2022 207429 1274	1200
	Very easy	Moderately easy	difficult	difficult	Very difficult	N/A
Finding the data	0	0	0	0	0	0
Downloading data	0	0	0	0	0	0
Use of data in the format provided	0	0	0	0	0	0
Determining the validity / accuracy of data	0	0	0	0		0
ther (please specify)		18				
3. How useful would ea	ch of these be i	n MedGen?				
	Extremely useful	Very useful	Somewhat useful	Not so useful	Not at all useful	N/A
Search results displayed in hierarchy format	0	0	0	0	0	0
Search using multiple parameters like genes and clinical features	0	0	0	0	0	0
Information organized by disease category (e.g. oncology, cardiology, pharmacogenetics)	0	0	0	0	0	0
List of support groups for a condition	0	0	0	0	0	0
A simplified disease page	0	0	0	0	0	0
More help on how to navigate the site	0	0	0	0	0	0
A quick summary on disease or condition	0	0	0	0	0	0
Clearer relationships and hierarchies in disease families	0		0	0	0	0
Up-to-date practice guidelines	0	0	0	0	0	0
ther (please specify)						
4. Do you have any oth	er suggestions	on how we can i	improve MedGen	?		
5. Would you be willing	to answer addi	tional questions	about ClinVar2			
	LO GIISWEI duul	aonai questions	about Omital:			
) Yes						
) No						

# ClinVar General

	Very easy	Moderately easy	Neither easy nor difficult	Moderately difficult	Very difficult	N/A
Finding the data	0	0	0	0	0	0
Downloading data	0	0	0	0	0	C
Use o <mark>f data in the format</mark> p <mark>rovided</mark>	0	0	0	0	0	С
Determining the validity / accuracy of data	0	0	0	0	0	C
other (please specify)						
other (please specify)						
	22 273 2				8 22	
	nVar data dow	vnload easier for y	ou (e.g., API, diff	erent format, e	tc.)?	
	nVar data dov	vnload easier for y	ou (e.g., API, diff	erent format, e	tc.)?	
7. What would make Cli		vnload easier for y	ou (e.g., API, diff	erent format, e	tc.)?	
7. What would make Cli 8. Do you submit data to		vnload easier for y	ou (e.g., API, diff	erent format, e	tc.)?	
7. What would make Cli  8. Do you submit data to		vnload easier for y	ou (e.g., API, diff	erent format, e	tc.)?	
7. What would make Cli 8. Do you submit data to		vnload easier for y	ou (e.g., API, diff	erent format, e	tc.)?	
7. What would make Cli  8. Do you submit data to		vnload easier for y	ou (e.g., API, diff	erent format, e	tc.)?	

		tasks during a C				
	Very easy	Moderately easy	Neither easy / nor difficult	Moderately difficult	Very difficult	N/A
Registering your organization	0	0	0	0	0	0
Uploading your data in the Submission Portal	0	0	0	0	0	0
Finding the status of your submission	0	0	0	0	0	0
Interpreting automated feedback (i.e., validation errors)	0	O	0	0	0	0
Interpreting submission reports and failure reports	0	0	0	0	0	0
Getting answers to your questions	0	0	0	0	0	0
Other (please specify)						
0. How useful would ea	ch of these be	in a ClinVar subr	nission?			
	Extremely useful		Somewhat useful	Not so useful	Not at all useful	N/A
API (Application Programming Interface) for submission	0	0	0	0	0	0
Excel format	0	0	0	0	0	0
XML format	0	0	0	0	0	0
Simpler submission process and/or file format	0	0	0	0		0
More help materials	0	0	0	0	0	0
Help during file preparation	0	0	0	0	0	0
Better communication about submission status	0	0	0	0	0	0
Other (please specify)						
51. Do you have any oth	er suggestions	for improving C	linVar?			
		-16				
		Prev	Next			

#### Medical Genetics and Variation overall feedback

52. Please tell us anything else that can help improve our resources for you.

Please write to us at <a href="mailto:suggest@ncbi.nlm.nih.gov">suggest@ncbi.nlm.nih.gov</a> if you would like to provide additional feedback on NCBI's Medical Genetics and Human Variation resources.

Prev

Done