Canadian Open Genetics Repository (COGR)

SurveyMonkey

1. A2) Are you a lab director?

		Response Percent	Response Count
Yes		**61.3%**	**19**
No		38.7%	12
		answered question	**31**
		skipped question	**1**

2. A3) Is your laboratory certified for clinical diagnostic testing services?

		Response Percent	Response Count
Yes		**80.8%**	**21**
No		19.2%	5
		answered question	**26**
		skipped question	**6**

3. A4) Does your laboratory conduct genetic testing for research purposes?

		Response Percent	Response Count
Yes		**57.7%**	**15**
No		42.3%	11
		answered question	**26**
		skipped question	**6**

4. A5) Are you a practicing clinician who orders testing or otherwise utilizes the services of a clinical genetic testing lab?

	Response Percent	Response Count
Yes	24.1%	7
No	**75.9%**	**22**
	answered question	**29**
	skipped question	**3**

5. A6) Are you a genetic counsellor or other healthcare provider who works in a clinic that sees patients and orders diagnostic genetic testing?

	Response Percent	Response Count
Yes	24.1%	7
No	**75.9%**	**22**
	answered question	**29**
	skipped question	**3**

6. B6) Which of the following represent concerns for data submission? Check all that apply.

	Response Percent	Response Count
Attribution must be provided for each variant submitted.	45.8%	11
All links to patient data must be removed.	**87.5%**	**21**
My staff will require some training in the transfer procedures.	75.0%	18
Any incremental lab costs we incur must be approved or recovered.	70.8%	17
The study team would need to provide assistance in the process.	66.7%	16
	answered question	**24**
	skipped question	**8**

7. B7) How do you envisage your laboratory or clinic will use the database as described above? (Check all that apply):

	Response Percent	Response Count
Refer to the knowledge base to see if there is a consensus about variant classification relative to a particular disease	**92.6%**	**25**
Refer to the knowledge base to see how individual labs interpret variants	74.1%	20
Use information from other labs to provide interpretation of a variant identified in your lab	77.8%	21
Use information from the database to provide better advice to your patients	63.0%	17
Use the database to identify genes reported to be associated with a particular disease by individual laboratories	63.0%	17
Will not use this database	0.0%	0
Other	0.0%	0
	answered question	**27**
	skipped question	**5**

8. B8) Even if you do not plan to share your own data, you can still have access to the database resources. How do you envisage your laboratory or clinic would use the database in this circumstance? (Check all that apply):

	Response Percent	Response Count
Refer to the knowledge base to see if there is a consensus about variant classification relative to a particular disease	**94.7%**	**18**
Refer to the knowledge base to see how individual labs interpret variants	73.7%	14
Use information from other labs to provide interpretation of a variant identified in your lab	84.2%	16
Use information from the database to provide better advice to your patients	63.2%	12
Use the database to identify genes reported to be associated with a particular disease by individual laboratories	57.9%	11
Will not use this database	0.0%	0
Other	0.0%	0
	answered question	**19**
	skipped question	**13**

9. C1) What kinds of genetic variant data do you collect as a matter of routine in your lab? (Choose all that apply):

	Response Percent	Response Count
Chromosome	57.1%	12
Strand	9.5%	2
Genome assembly	38.1%	8
Genomic coordinates	52.4%	11
Protein Ids (NP, UniProt, etc)	19.0%	4
Gene symbol	71.4%	15
Gene ids (Ex, HGNC, OMIM)	42.9%	9
Transcript ID (NM, Ensembl, UCSC)	57.1%	12
Variant Type (ex substitution, insertion)	76.2%	16
Variant coding effect (ex. Missense, nonsense, frameshift)	81.0%	17
Variant spectrum at the gene level	33.3%	7
Variant location (exon 1, intron 3)	76.2%	16
cDNA nomenclature	**90.5%**	**19**
Protein nomenclature	85.7%	18
Distance to nearest splice site	19.0%	4
Splice Site Consensus Scores (NNSPLICE, SpliceSiteFinder, etc)	23.8%	5
Protein domain	9.5%	2
Lab classification	47.6%	10
dbSNP I.D.	52.4%	11
dbSNP validation	14.3%	3

dbSNP clinical significance	23.8%	5
dbSNP global Minor Allele Frequency	47.6%	10
ESP Global MAF	19.0%	4
ESP Population MAF	19.0%	4
ESP Coverage	4.8%	1
ESP Average Read Depth	9.5%	2
1000 Genomes Global MAF	33.3%	7
1000 Genomes Population MAF	33.3%	7
HGMD	42.9%	9
Ingenuity	9.5%	2
Other fee-based Databases	19.0%	4
Open-access databases	42.9%	9
ClinVar	33.3%	7
PubMed Search	81.0%	17
Internet Search (ex Google)	66.7%	14
Nucleotide Conservation (PhastCons, PhyloP)	42.9%	9
Amino Acid Conservation (Alamut, UCSC)	57.1%	12
Amino Acid Substitution Matrix (ex BLOSUM, PFAM)	23.8%	5
Amino acid biochemical changes (ex. Composition, polarity, volume)	23.8%	5
Grantham distance	38.1%	8
Functional Prediction (PolyPhen2, SIFT, MutationAssessor)	66.7%	14
Proband Phenotypes	42.9%	9
Proband Zygosity	23.8%	5

Segregation among affected individuals		47.6%	10
Non-segregation		42.9%	9
Literature Control Studies		47.6%	10
Compound heterozygosity with pathogenic variants		66.7%	14
De novo occurance		47.6%	10
Functional assays in literature		42.9%	9
Number of proband chromosomes with variant		28.6%	6
Number of control chromosomes with variant		28.6%	6
		answered question	**21**
		skipped question	**11**

10. C2) Do you use a formal system for tracking information on genetic variants derived from the scientific literature?

		Response Percent	**Response Count**
Yes, stored in Database		37.5%	9
Yes, stored in Flat file		8.3%	2
No		**54.2%**	**13**
		answered question	**24**
		skipped question	**8**

11. C3) How are genetic variant data stored in your lab? Choose the principal method from the list. (Choose all that apply):

	Response Percent	Response Count
Spread sheet	**73.7%**	**14**
Access database	42.1%	8
SQL database	15.8%	3
	answered question	**19**
	skipped question	**13**

12. Please specify the database or software names.

	Response Percent	Response Count
Open source	13.3%	2
Public database	13.3%	2
Commercial solution	**53.3%**	**8**
Other (specify)	33.3%	5
	answered question	**15**
	skipped question	**17**

13. C4) Do you use a particular software application to curate information about variants?

	Response Percent	Response Count
Yes	40.9%	9
No	**59.1%**	**13**
If yes, provide software name of method used:		9
answered question		**22**
skipped question		**10**

14. C5) Does your laboratory use the ACMG system below for classifying sequence variants for Mendelian disorders? Pathogenic Variant of uncertain significance - likely pathogenic Variant of uncertain significance Variant of uncertain significance - likely benign Benign

	Response Percent	Response Count
Yes	**63.2%**	**12**
No	36.8%	7
answered question		**19**
skipped question		**13**

15. C6) Does your laboratory (a) use a consistent set of classification terms or (b) are laboratory directors able to change the terms on a case-by-case basis?

	Response Percent	Response Count
(a)	**65.0%**	**13**
(b)	35.0%	7
answered question		**20**
skipped question		**12**

16. C7) How many classification tiers do you use in your laboratory?

		Response Percent	Response Count
1-5____		**78.9%**	**15**
6-10_____		10.5%	2
>10_____		5.3%	1
Other (please specify)		5.3%	1
		answered question	**19**
		skipped question	**13**

17. C9) Does your laboratory use a different classification terminology for somatic, pharmacogenomic or risk variants?

		Response Percent	Response Count
Yes		17.6%	3
No		**82.4%**	**14**
		answered question	**17**
		skipped question	**15**

18. If yes, please list terms used for:

	Response Percent	Response Count
Somatic	**100.0%**	**3**
Pharmacogenomic	0.0%	0
Risk	0.0%	0
	answered question	**3**
	skipped question	**29**

19. C10) Does your laboratory provide information on penetrance and/or expressivity for individual variants?

	Response Percent	Response Count
Yes	27.8%	5
No	**72.2%**	**13**
	answered question	**18**
	skipped question	**14**

20. C11) Regardless of the classification system you use, does your laboratory have written rules or documentation outlining the criteria necessary for the evidence-based classification of variants?

	Response Percent	Response Count
Yes (go to C12 and C13)	40.0%	8
No (go to C14)	**60.0%**	**12**
	answered question	**20**
	skipped question	**12**

21. C13) How often do you adhere to these written rules?

	Response Percent	Response Count
Nearly all the time, with limited additional judgment required	16.7%	1
Most of the time, though many variants require additional judgment to classify	**83.3%**	**5**
Most variants do not fit rules and require manual assignment	0.0%	0
Other (please specify)	0.0%	0
	answered question	**6**
	skipped question	**26**

22. C14) Do you use a numerical scoring matrix to arrive at your variant interpretations?

	Response Percent	Response Count
Yes	0.0%	0
No	**100.0%**	**19**
	answered question	**19**
	skipped question	**13**

23. C15) In addition to the classification of a particular variant, does your lab create text detailing the reasons for this classification?

	Response Percent	Response Count
Yes (go to C16)	**57.9%**	**11**
No (go to C17)	42.1%	8
	answered question	**19**
	skipped question	**13**

24. C16) Is this text saved and reused when the variant is identified in another individual or is the text regenerated each time?

	Response Percent	Response Count
Text is standardized and infrequently updated	18.2%	2
Text is saved and repeated, though edits may be made each time	**72.7%**	**8**
Text is not saved but is regenerated for each individual	9.1%	1
	answered question	**11**
	skipped question	**21**

25. C19) Are your variant data linked to patient information?

	Response Percent	Response Count
Yes (go to C20, C21, C22)	**84.2%**	**16**
No (go to C23)	15.8%	3
	answered question	**19**
	skipped question	**13**

26. C20) Are your variant data linked to all patients in your database who have the variant? (i.e. are you able to retrieve a list of all patients who have a variant?)

	Response Percent	Response Count
Yes	**66.7%**	**12**
No	33.3%	6
	answered question	**18**
	skipped question	**14**

27. C21) Are variant data linked directly to the disease type your patients present with?

	Response Percent	Response Count
Yes	**61.1%**	**11**
No	38.9%	7
	answered question	**18**
	skipped question	**14**

28. C22) Do you provide additional levels of phenotypic detail for variants associated with disease?

	Response Percent	Response Count
Yes	44.4%	8
No	**55.6%**	**10**
	answered question	**18**
	skipped question	**14**

29. C23) Do you maintain a database that tracks the number of clinical reports or individuals associated with a particular variant?

	Response Percent	Response Count
Yes	**68.2%**	**15**
No	27.3%	6
Dont' know	4.5%	1
	answered question	**22**
	skipped question	**10**

30. C24) Do you maintain a database that tracks the number of families associated with a particular variant?

	Response Percent	Response Count
Yes	**54.5%**	**12**
No	45.5%	10
	answered question	**22**
	skipped question	**10**

31. C25) Does your laboratory track new knowledge from the literature about the association of genes with different diseases for genes where disease causality may not have been firmly established? E.g. Association of MYH6 variants with cardiomyopathy (limited data) vs known relationship of MYH7 with cardiomyopathy.

	Response Percent	Response Count
Yes	30.0%	6
No	**70.0%**	**14**
	answered question	**20**
	skipped question	**12**

32. C26) What is your policy for reassessment of variants?

		Response Percent	Response Count
(1) Reassess every variant at a routine frequency regardless of identification in additional patients		0.0%	0
(2) Reassess variants every time they are seen in a new patient		**65.0%**	**13**
(3) Reassess variants if seen in a new patient and have not be assessed within a set period		30.0%	6
(4) Do not reassess the pathogenicity of variants		5.0%	1
	If you selected (1) or (3), indicate time period between assessments.		5
		answered question	**20**
		skipped question	**12**

33. C27) What do you do when a previously reported variant changes categories? (Check all that apply):

	Response Percent	Response Count
Amend and report out all cases with this variant	15.0%	3
Report out and amend current case only	25.0%	5
Call each clinician who has a patient with this variant	5.0%	1
Send an email to each clinician who has a patient with this variant	5.0%	1
Automatically alert clinicians via other electronic communication	5.0%	1
We do not re-contact clinicians or patients but they may contact us to obtain an update on a given variant	**65.0%**	**13**
Not applicable. We do not reassess variants	10.0%	2
	answered question	**20**
	skipped question	**12**

34. D2) There are several options for loading variant-level information into GeneInsight. All of these options would enable each lab or end-user to input micro-attribution of variant-level information. Of the following options, please select all that you would be likely to choose:

	Response Percent	Response Count
I can provide variant-level information in a common format for batch upload.	38.9%	7
I need assistance extracting variant information and saving it in a common format for batch upload.	**55.6%**	**10**
In addition to batch uploads, I would like to manually edit individual variant interpretations from my laboratory within the centralized GeneInsight database.	50.0%	9
I would like GeneInsight to be more directly integrated into my lab and/or be the database for my lab's internal variant classifications.	33.3%	6
I annotate and store variants in Alamut and would like to use this software for batch upload and/or for submitting my variants individually.	33.3%	6
	answered question	**18**
	skipped question	**14**