Clinical Genomes Diagnostics survey

The purpose of this survey is to assess routine usage of tools for rare disease diagnosis, rather than experiences specific to the CAGI7 Clinical Genomes Challenge; nonetheless, answers should refer to the tool you used to complete the CAGI7 challenge. Data collected using this survey will be used for research purposes to assess general platform usability and services such as filtering, reporting, and reanalysis.

By completing this survey, you are agreeing to participate in the study. You can skip any question you do not wish to answer. No direct personal identifiers will be collected.

Ingestion and analysis of which of the following data types is supported by th	е
platform?	

	srNGS	IrNGS	RNAseq	Methylation
Yes	0	0	0	0
No	0	0	0	0
Not sure/Not evaluated	0	0	0	0

If you selected 'Other' for supported data types, please describe.

Your answer



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	VCF		
	GVCF		
	CRAM		
	ВАМ		
	FASTQ		
	BED		
	Other		
	Prefer not to answer		
	If you selected 'Other' for input file types Your answer	s, please describe.	
	For which of the following variant types generation? CNVs	does the platform supp	port primary
	SVs		
	Mitochondrial		
	TREs		
	Mosaicism		
	Manually entered variants		
	Prefer not to answer		

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	piationii gonoratoa cano.		
	○ Yes		
	O No		
	Not evaluated/Not able to evaluate		
	Prefer not to answer		
	Once data is generated, which call types	is the platform able to	support?
	CNVs		
	SVs		
	Mitochondrial		
	TREs		
	Mosaicism		
	Manually entered variants		
	Prefer not to answer		



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	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer
PGx alleles	0	0	0	0	0	0	0
HLA alleles	0	0	0	0	0	0	0
Mitochondrial haplotypes	0	0	0	0	0	0	0
RNA expression levels, splicing, intronic retention	0	0	0	0	0	0	0
Support for problematic genes (SMN1/2, GBA, CYP21A2, KCNE1, RNU genes)	0	0	0	0	0	0	0
Loss of heterozygosity	0	0	0	0	0	0	0
Ploidy - chromosomal and regional	0	0	0	0	0	0	0

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	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer	
Variant nomenclature (HGVS, ISCN, VRS)	0	0	0	0	0	0	0	
MANE Select and MANE Plus Clinical transcripts	0	0	0	0	0	0	0	
Specification of custom transcripts (non-MANE, etc)	0	0	0	0	0	0	0	
Literature search - facilitate search with variant or gene strings	0	0	0	0	0	0	0	
Database integration (e.g. ClinVar, HGMD, OMIM, VLM network)	0	0	0	0	0	0	0	
Tool integration (SpliceAI, Missense predictors, etc)	0	0	0	0	0	0	0	0
Tracking of								\perp

proressional variant classification standards OOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOO	Preview mode				⊘ Pı	ıblished	Cop	y responder li
application of variant classification evidence types O O O O O O O O O O O O O O O O O O O	variant classification	0	0	0	0	0	0	0
knowledgebase independent from case analysis O O O O O O O O O O O O O O O O O O	application of variant classification	0	0	0	0	0	0	0
ClinVar Access internal allele frequencies and artifacts Access to internal case-level data OOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOOO	knowledgebase independent from case	0	0	0	0	0	0	0
allele frequencies and artifacts Access to internal case- level data O O O O O O O O O O O O O O O O O O	ClinVar	0	0	0	0	0	0	0
internal case- level data	allele frequencies	0	0	0	0	0	0	0
level data	internal case-	0	0	0	0	0	0	0
	Your answer							



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	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer
Clear and informative reports	0	0	0	0	0	0	0
Flexibility in formats	0	0	0	0	0	0	0
User modifiable report templates	0	0	0	0	0	0	0
Report automation through data autopopulation	0	0	0	0	0	0	0

If you scored any Reporting Capabilities function under 4, please explain limitations.

Your answer



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	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer
Useful variant prioritization standardized filters	0	0	0	0	0	0	0
Customizable variant filtration parameters	0	0	0	0	0	0	0
Use of phenotype in prioritization	0	0	0	0	0	0	0
Support for family based analysis (duos, trios, quads, larger pedigrees)	0	0	0	0	0	0	0
Support for panel analysis (all variants)	0	0	0	0	0	0	0
Phenotype intake and use in prioritization (use of EMR records, HPO terms, external platforms,							

etc)

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prioritization (use of EMR records, HPO terms, external platforms, etc)	0	0	0	0	0	0	0	
If you scored any please explain lin		•	ization, a	nd Decisi	on Suppor	t function u	nder 4,	
Your answer								

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	1	2	3	4	5	evaluated/Not able to	Prefer not to nswer
Sex concordance	0	0	0	0	0	0	0
Familial relatedness	0	0	0	0	0	0	0
Coverage metrics for test regions or genes	0	0	0	0	0	0	0
Genome metric outlier tracking (e.g. due to contamination)	0	0	0	0	0	0	0
Output low coverage regions as relevant to analysis	0	0	0	0	0	0	0
If you scored any	If you scored any Quality Control function under 4, please explain limitations.						
Your answer							
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innovative/aty	innovative/atypically well-supported)								
	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer		
Automated detection of new findings (new genes, variants newly classified as P/LP)	0	0	0	0	0	0	0		
Ability to update phenotype	0	0	0	0	0	0	0		
Integration of old and new analysis	0	0	0	0	0	0	0		
If you scored any Reanalysis function under 4, please explain limitations. Your answer									

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	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer	
User interface	0	0	0	0	0	0	0	
Speed and responsiveness of interface	0	0	0	0	0	0	0	
Supports team use (e.g. communication, notes)	0	0	0	0	0	0	0	
User management (onboarding, permissions, role- based access control)	0	0	0	0	0	0	0	
Ease of out-of- platform internet/database searching	0	0	0	0	0	0	0	

If you scored any Ease of Use and Workflow Efficiency function under 4, please explain limitations.

Your answer

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	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer		
Data Input Flexibility (vcf, FASTQ, CRAM, BAM)	0	0	0	0	0	0	0		
LIMS integration ease	0	0	0	0	0	0	0		
Configuration of custom bioinformatic workflows	0	0	0	0	0	0	0		
Availability of APIs	0	0	0	0	0	0	0		
Private (self- managed) deployment option (on prem or bring your own cloud)	0	0	0	0	0	0	0		
Import/Export of data capability (for platform transition, research use)	0	0	0	0	0	0	0		
Visualization tools	0	0	0	0	0	0	0		

compliance

If you scored any Integration, Data Input Flexibility, and Test Type Compatibility function under 4, please explain limitations.

Your answer

New releases

How well are new releases supported? (1 = not supported, 5 = innovative/atypically well-supported)

	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer	
Flexible launch timelines	0	0	0	0	0	0	0	
Useful release notes	0	0	0	0	0	0	0	
Validation support/help	0	0	0	0	0	0	0	
Teaching tutorials	0	0	0	0	0	0	0	
Ability to run multiple versions in parallel	0	0	0	0	0	0	0	

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Your answer

Please list all regulatory or security frameworks under which your system operates.

Your answer

Thank you for filling out the survey! Your input is extremely valuable.

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