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 the platform?

	srNGS	lrNGS	RNAseq	Methylation
Yes	○	○	○	○
No	○	○	○	○
Not sure/Not evaluated	○	○	○	○

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

Your answer

- [ ] VCF
- [ ] GVCF
- [ ] CRAM
- [ ] BAM
- [ ] FASTQ
- [ ] BED
- [ ] Other
- [ ] Prefer not to answer

If you selected 'Other' for input file types, please describe.

Your answer

For which of the following variant types does the platform support primary generation?

- [ ] CNVs
- [ ] SVs
- [ ] Mitochondrial
- [ ] TREs
- [ ] Mosaicism
- [ ] Manually entered variants
- [ ] Prefer not to answer

platform generated calls?

- Yes
- 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

innovative/atypically well-supported)

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

limitations.

Your answer

innovative/atypically well-supported)

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

professional variant classification standards	○	○	○	○	○	○	○
Automated application of variant classification evidence types	○	○	○	○	○	○	○
Management of knowledgebase independent from case analysis	○	○	○	○	○	○	○
Submission to ClinVar	○	○	○	○	○	○	○
Access internal allele frequencies and artifacts	○	○	○	○	○	○	○
Access to internal case-level data	○	○	○	○	○	○	○
level data							

If you scored any Variant Curation Support function under 4, please explain limitations.

Your answer

innovative/atypically well-supported)

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

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

Your answer

innovative/atypically well-supported)

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

use in prioritization (use of EMR records, HPO terms, external platforms, etc)	○	○	○	○	○	○	○

If you scored any Filtering, Prioritization, and Decision Support function under 4, please explain limitations.

Your answer

innovative/atypically well-supported)

	1	2	3	4	5	Not evaluated/Not able to evaluate	Prefer not to answer
Sex concordance	○	○	○	○	○	○	○
Familial relatedness	○	○	○	○	○	○	○
Coverage metrics for test regions or genes	○	○	○	○	○	○	○
Genome metric outlier tracking (e.g. due to contamination)	○	○	○	○	○	○	○
Output low coverage regions as relevant to analysis	○	○	○	○	○	○	○

If you scored any Quality Control function under 4, please explain limitations.

Your answer

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)	○	○	○	○	○	○	○
Ability to update phenotype	○	○	○	○	○	○	○
Integration of old and new analysis	○	○	○	○	○	○	○

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

Your answer

innovative/atypically well-supported)

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

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

Your answer

innovative/atypically well-supported)

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

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	○	○	○	○	○	○	○
Useful release notes	○	○	○	○	○	○	○
Validation support/help	○	○	○	○	○	○	○
Teaching tutorials	○	○	○	○	○	○	○
Ability to run multiple versions in parallel	○	○	○	○	○	○	○

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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