

CLAIMS

~~We Claim:~~

1. A system comprising:

at least one processor; and

a non-transitory computer readable medium comprising instructions that, when executed by the at least one processor, cause the system to:

determine sequencing metrics for nucleotide-base calls of nucleotide reads corresponding to a genomic coordinate of a sample nucleotide sequence;

generate, utilizing a call-recalibration-machine-learning model and based on the sequencing metrics, one or more variant-call classifications indicating an accuracy of identifying a variant at the genomic coordinate; and

determine a final nucleotide-base call for the genomic coordinate based on the one or more variant-call classifications.
2. The system of claim 1, further comprising instructions that, when executed by the at least one processor, cause the system to:

increase a base-call-quality metric for the final nucleotide-base call based on the one or more variant-call classifications;

determine the increased base-call-quality metric passes a quality filter; and

generate a variant call file that includes the final nucleotide-base call based on the increased base-call-quality metric.

3. The system of claim 1, further comprising instructions that, when executed by the at least one processor, cause the system to:

decrease a base-call-quality metric for the final nucleotide-base call based on the one or more variant-call classifications;

determine that the decreased base-call-quality metric does not pass a quality filter; and

generate a post-filter variant call file that excludes the final nucleotide-base call based on the decreased base-call-quality metric not passing the quality filter.

4. The system of [any one of claims 1-3](#), further comprising instructions that, when executed by the at least one processor, cause the system to determine the final nucleotide-base call for the genomic coordinate by:

determining an initial nucleotide-base call for the genomic coordinate utilizing a call-generation model based on one or more sequencing metrics; and

modifying one or more data fields corresponding to a variant call file and the initial nucleotide-base call based on the one or more variant-call classifications from the call-recalibration-machine-learning model.

5. The system of [any one of claims 1-4](#), further comprising instructions that, when executed by the at least one processor, cause the system to modify one or more data fields corresponding to a variant call file for one or more of call quality, genotype, or genotype quality based on the one or more variant-call classifications.

6. The system of [any one of claims 1-5](#), further comprising instructions that, when executed by the at least one processor, cause the system to determine the sequencing metrics for the nucleotide-base calls by determining one or more of read-based sequencing metrics, call-model-generated sequencing metrics, or externally sourced sequencing metrics.

7. The system of claim 6, further comprising instructions that, when executed by the at least one processor, cause the system to determine the call-model-generated sequencing metrics by determining variant-caller sequencing metrics and mapping-and-alignment sequencing metrics from a call-generation model.

8. The system of [any one of claims 1-7](#), further comprising instructions that, when executed by the at least one processor, cause the system to determine the sequencing metrics for the nucleotide-base calls by determining re-engineered sequencing metrics derived from other sequencing metrics for the nucleotide-base calls.

9. The system of [any one of claims 1-8](#), further comprising instructions that, when executed by the at least one processor, cause the system to generate the one or more variant-call classifications by generating one or more of a false-positive classification, a genotype-error classification, or a true-positive classification.

10. The system of [any one of claims 1-9](#), further comprising instructions that, when executed by the at least one processor, cause the system to:

determine contribution measures for the sequencing metrics indicating respective measures of impact that the sequencing metrics have on the final nucleotide-base call; and

provide, for display on a client device, a visualization of the contribution measures corresponding to one or more of the sequencing metrics.

11. A non-transitory computer readable medium comprising instructions that, when executed by at least one processor, cause a computing device to:

determine sequencing metrics for nucleotide-base calls of nucleotide reads corresponding to a genomic coordinate of a sample nucleotide sequence;

generate, utilizing a call-recalibration-machine-learning model and based on the sequencing metrics, one or more variant-call classifications indicating an accuracy of identifying a variant at the genomic coordinate; and

determine a final nucleotide-base call for the genomic coordinate based on the one or more variant-call classifications.

12. The non-transitory computer readable medium of claim 11, further comprising instructions that, when executed by the at least one processor, cause the computing device to determine the final nucleotide-base call as part of a variant call comprising a single nucleotide polymorphism, a deletion, or an insertion corresponding to the genomic coordinate.

13. The non-transitory computer readable medium of claim 11 [or 12](#), further comprising instructions that, when executed by the at least one processor, cause the computing device to determine the final nucleotide-base call for the genomic coordinate by changing a

genotype of an initial nucleotide-base call to an updated genotype of an updated nucleotide-base call.

14. The non-transitory computer readable medium of [any one of claims 11-13](#), further comprising instructions that, when executed by the at least one processor, cause the computing device to generate the one or more variant-call classifications by utilizing the call-recalibration-machine-learning model to generate one or more classifications corresponding to a genotype for the final nucleotide-base call for the genomic coordinate.

15. The non-transitory computer readable medium of [any one of claims 11-14](#), further comprising instructions that, when executed by the at least one processor, cause the computing device to:

determine the sequencing metrics by determining variant-caller sequencing metrics and mapping-and-alignment sequencing metrics from a call-generation model; and

generate the one or more variant-call classifications based on the variant-caller sequencing metrics and the mapping-and-alignment sequencing metrics utilizing the call-recalibration-machine-learning model.

16. A computer-implemented method comprising:

determining sequencing metrics for nucleotide-base calls of nucleotide reads corresponding to a genomic coordinate of a sample nucleotide sequence;

generating, utilizing a call-recalibration-machine-learning model and based on the sequencing metrics, one or more variant-call classifications indicating an accuracy of identifying a variant at the genomic coordinate; and

determining a variant call for the genomic coordinate based on the one or more variant-call classifications.

17. The computer-implemented method of claim 16, further comprising:
updating a base-call-quality metric for the variant call based on the one or more variant-call classifications;

determining that the base-call-quality metric for the variant call passes a quality filter; and
generating a variant call file comprising the variant call based on the base-call-quality metric passing the quality filter.

18. The computer-implemented method of claim 16 or 17, wherein generating the one or more variant-call classifications comprises utilizing a machine-learning classifier to generate one or more of:

a false-positive probability that the variant call is a false positive;
a genotype-error probability that a genotype for the variant call is incorrect; or
a true-positive probability that the variant call is a true positive.

19. The computer-implemented method of any one of claims 16-18, wherein determining the sequencing metrics comprises determining one or more of read-based sequencing metrics comprising metrics derived from the nucleotide reads of the sample nucleotide sequence, call-model-generated sequencing metrics generated via a call-generation model, or externally sourced sequencing metrics identified from one or more external databases.

~~20.—The computer-implemented method of claim 16, further comprising determining contribution measures for the sequencing metrics indicating respective measures of impact that the sequencing metrics have on the variant call for the genomic coordinate.~~

~~20~~²¹. The computer-implemented method of any one of claims 16-19, wherein the call-recalibration-machine-learning model comprises one or more of a neural network, a deep-learning transformer, a gradient boost decision tree, a random forest model, a support vector machine, a linear regression, or a logistic regression.