CLAIMS

1. A genomic services system comprising:

a network interface through which are received genomic sequence reads derived from a biological sample obtained from a user; and

one or more processors configured to execute a bioinformatics processing pipeline including:

a read alignment module configured to generate observed sequence data by aligning the sequence reads relative to a reference sequence;

a variant calling module operative to identify observed variants in the observed sequence data;

a variant refinement module configured to produce a set of refined variants associated with the user based on the identified observed variants;

a variant imputation module configured to produce a set of predicted genotypes associated with the user based on population reference genotype data; and

a variant storage module configured to:

receive a first application programming interface (API) call from a network infrastructure of a partner application provider among multiple partner application providers, the received first API call requesting a notification that indicates whether the user consented to share a genomic window that includes selected ones of the refined variants or predicted genotypes with the partner application provider, the genomic window corresponding to the partner application provider;

respond to the received first API call by providing the network infrastructure of the partner application provider with the requested notification, the provided notification indicating that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider;

in response to the provided notification that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider, receive a second API call from the network infrastructure of the partner application provider, the received second API call requesting genomic information of the user and identifying the genomic window that corresponds to the partner application provider; and

based on the provided notification that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider, respond to the received second API call by returning the genomic window that corresponds to the partner application provider and includes the selected ones of the refined variants or predicted genotypes to the network infrastructure of the partner application provider identified by the received second API call.

2. The genomics services system of claim 1 wherein the received second API call comprises a unique identifier specifying a user and a partner application, and wherein the selected ones of the refined variants or predicted genotypes correspond to a genomics service being provided by the partner application provider identified by the received second API call.

3. The genomics services system of claim 2, wherein the variant storage module is further configured to use the unique identifier included in the received second API call to perform a look-up of a genomic data file, the genomic data file defining the genomic window that corresponds to the partner application provider.

4. The genomic services system of claim 1, wherein the variant storage module is further configured to store a record of all genomic information of the user provided to the partner application provider.

5. The genomic services system of claim 1, wherein the selected ones of the refined variants or predicted genotypes are returned asynchronously in relation to the request.

6. The genomic services system of claim 1, wherein:

the variant calling module is further operative to:

output first data including information on all sites in the observed sequence data; and

output second data including posterior genotype likelihoods for variants for each variant in the observed sequence data; and

the variant imputation module is further configured to merge the first data with the second data to produce the set of refined variants.

7. The genomic services system of claim 1, wherein the variant imputation module is configured to:

impute variants based on phased high-quality haplotypes;

boost probability information relating to low-quality genotypes to produce boosted genotypes; and

combine the high-quality haplotypes and the boosted genotypes to create the set of predicted genotypes.

8. The genomic services system of claim 1, wherein the set of predicted genotypes are not directly assayed in the biological sample obtained from the user.

9. The genomic services system of claim 3, wherein genomic information within the genomic window that corresponds to the partner application provider is accessible by the partner application provider or the partner application, and wherein genomic information outside of the genomic window that corresponds to the partner application provider is inaccessible by the partner application provider and the partner application.

10. The genomic services system of claim 2, wherein the unique identifier preserves anonymity of the user in relation to the partner application provider.

11. A method comprising:

receiving genomic sequence reads derived from a biological sample obtained from a user;

generating observed sequence data by aligning the sequence reads relative to a reference sequence;

identifying observed variants in the observed sequence data;

producing a set of refined variants associated with the user based on the identified observed variants;

producing a set of predicted genotypes associated with the user based on population reference genotype data;

receiving a first application programming interface (API) call from a network infrastructure of a partner application provider among multiple partner application providers, the received first API call requesting a notification that indicates whether the user consented to share a genomic window that includes selected ones of the

refined variants or predicted genotypes with the partner application provider, the genomic window corresponding to the partner application provider;

responding to the received first API call by providing the network infrastructure of the partner application provider with the requested notification, the provided notification indicating that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider;

in response to the provided notification that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider, receiving a second API call from the network infrastructure of the partner application provider, the received second API call requesting genomic information of the user and identifying the genomic window that corresponds to the partner application provider; and

based on the provided notification that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider, responding to the received second API call by returning the genomic window that corresponds to the partner application provider and includes the selected ones of the refined variants or predicted genotypes to the network infrastructure of the partner application provider identified by the received second API call.

12. The method of claim 11, wherein the received second API call comprises a unique identifier specifying a partner application, and wherein the selected ones of the refined variants or predicted genotypes correspond to a genomic service being provided by the partner application provider identified by the received second API call.

13. The method of claim 12, further comprising performing, based on the unique identifier included in the received second API call, a look-up of a genomic data file, the genomic data file defining the genomic window that corresponds to the partner application provider.

14. The method of claim 11, further comprising storing a record of all genomic information of the user provided to the partner application provider.

15. The method of claim 11, wherein the selected ones of the refined variants or predicted genotypes are returned asynchronously in relation to the request.

16. A non-transitory machine-readable medium comprising instructions that, when executed by a machine, cause the machine to perform operations comprising:

receiving genomic sequence reads derived from a biological sample obtained from a user;

generating observed sequence data by aligning the sequence reads relative to a reference sequence;

identifying observed variants in the observed sequence data;

producing a set of refined variants associated with the user based on the identified observed variants;

producing a set of predicted genotypes associated with the user based on population reference genotype data;

receiving a first application programming interface (API) call from a network infrastructure of a partner application provider among multiple partner application providers, the received first API call requesting a notification that indicates whether the user consented to share a genomic window that includes selected ones of the refined variants or predicted genotypes with the partner application provider, the genomic window corresponding to the partner application provider;

responding to the received first API call by providing the network infrastructure of the partner application provider with the requested notification, the provided notification indicating that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider;

in response to the provided notification that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes with the partner application provider, receiving a second API call from the network infrastructure of the partner application provider, the received second API call requesting genomic information of the user and identifying the genomic window that corresponds to the partner application provider; and

based on the provided notification that the user consented to share the genomic window that includes the selected ones of the refined variants or predicted genotypes

with the partner application provider, responding to the received second API call by returning the genomic window that corresponds to the partner application provider and includes the selected ones of the refined variants or predicted genotypes to the network infrastructure of the partner application provider identified by the received second API call.

17. The non-transitory machine-readable medium of claim 16, wherein the received second API call comprises a unique identifier specifying a user and a partner application, and wherein the selected ones of the refined variants or predicted genotypes correspond to a genomics service being provided by the partner application provider identified by the received second API call.

18. The non-transitory machine-readable medium of claim 17, wherein the operations further comprise performing, based on the unique identifier included in the received second API call, a look-up of a genomic data file, the genomic data file defining the genomic window that corresponds to the partner application provider.

19. The non-transitory machine-readable medium of claim 16, wherein the operations further comprise storing a record of all genomic information of the user provided to the partner application provider.

20. The non-transitory machine-readable medium of claim 16, wherein the selected ones of the refined variants or predicted genotypes are returned asynchronously in relation to the request.