CLAIMS

Claim 1. A method for performing incremental secondary analysis of nucleic acid sequence reads, the method comprising:

(i) obtaining by one or more processors of a nucleic acid sequencing device, first data describing a plurality of first reads generated by the nucleic acid sequencing device during a first read interval, wherein each of the first reads represents a first ordered sequence of nucleotides

(ii) obtaining by one or more processors of the nucleic acid sequencing device, second data describing a plurality of second reads generated by the nucleic acid sequencing device during a second read interval that is performed after the first read interval, wherein each read of the second reads represents a second ordered sequence of nucleotides, wherein while the second data is being obtained, the method further comprises:

(a) providing, by one or more processors of the nucleic acid sequencing device, the first data as an input to a mapping and alignment engine;

(b) receiving, by one or more processors of the nucleic acid sequencing device and from the mapping and alignment engine, alignment results that indicate a quality of an alignment of the first data;

(c) determining, by one or more processors of the nucleic acid sequencing device and based on the received alignment results of the first data, that the nucleic acid sequencing device is to terminate operations that obtain the second data; and

(d) causing, by one or more processors of the nucleic acid sequencing device, the nucleic acid sequencing device to terminate operations that obtain the second data.

Claim 2. The method of claim 1, wherein the mapping and alignment engine is included within the nucleic acid sequencing device.

Claim 3. The method of claim 1,

wherein one or more of the first reads includes data representing a first sample identifier, and

wherein one or more of the second reads includes data representing a second sample identifier.

Claim 4. The method of claim 3, further comprising:

while the second data is being obtained:

organizing by one or more processors of the nucleic acid sequencing device, the one or more first reads into respective groups based on at least a first sample identifier or a second sample identifier; and

generating by one or more processors of the nucleic acid sequencing device, organization statistics, the organization statistics indicating a number of first reads corresponding to each sample identifier.

Claim 5. The method of claim 1, the method further comprising:

providing, by one or more processors of the nucleic acid sequencing device, output data that represents the received alignment results of first data before or while aligning the second portion of the cluster of reads.

Claim 6. The method of claim 1, the method further comprising:

causing, by one or more processors of the nucleic acid sequencing device, the mapping and alignment engine to begin a subsequent alignment of the data representing the first plurality of reads to a reference sequence.

Claim 7. The method of claim 1, the method further comprising:

while obtaining the second data, causing, by one or more processors of the nucleic acid sequencing device, a secondary analysis engine to determine a set of likely variants for the first data representing the first plurality of reads that was aligned to a reference sequence.

Claim 8. The method of claim 1, wherein the at least a portion of the second data representing the second plurality of reads is aligned while at least a different portion of second data representing the second plurality of reads is being obtained.

Claim 9. The method of claim 1, wherein the mapping and alignment engine is instructed to begin alignment of the second data representing the second plurality of reads a predetermined number of sequencing cycles before the second data is completely obtained.

Claim 10. The method of claim 1, wherein the mapping and alignment engine is implemented using a field programmable gate array (FPGA).

Claim 11. The method of claim 1, wherein the mapping and alignment engine is implemented using an Application Specific Integrated Circuit (ASIC).

Claim 12. The method of claim 1, wherein the mapping and alignment engine is implemented by using one or more computers to execute software instructions.

Claim 13. A system for performing incremental secondary analysis of nucleic acid sequence reads, the system comprising:

a nucleic acid sequencing device; and

one or more memory devices storing instructions that, when executed by one or more processors of the nucleic acid sequencing device, cause the nucleic acid sequencing device to perform operations, the operations comprising:

(i) obtaining, by the one or more processors of the nucleic acid sequencing device, first data describing a plurality of first reads generated by a nucleic acid sequencing device during a first read interval, wherein each of the first reads represents a first ordered sequence of nucleotides;

(ii) obtaining, by the one or more processors of the nucleic acid sequencing device, second data describing a plurality of second reads generated by the nucleic acid sequencing device during a second read interval that is performed after the first read interval, wherein each of the second reads represents a second ordered sequence of nucleotides, wherein while the second data is being obtained, the operations further comprising:

(a) providing, by the nucleic acid sequencing device, the first data as an input to a mapping and alignment engine;

(b) receiving, by the one or more processors of the nucleic acid sequencing device and from the mapping and alignment engine, alignment results that indicate a quality of an alignment of the first data; and

(c) determining, by the one or more processors of the nucleic acid sequencing device and based on the received alignment results of the first data, that the nucleic acid sequencing device is to terminate operations that obtain the second data; and

(d) causing, by the one or more processors of the nucleic acid sequencing device, the nucleic acid sequencing device to terminate operations that obtain the second data.

Claim 14. The system of claim 13, wherein the mapping and alignment engine is included within the nucleic acid sequencing device.

Claim 15. The system of claim 13,

wherein one or more of the first reads includes data representing a first sample identifier, and

wherein one or more of the second reads includes data representing a second sample identifier.

Claim 16. The system of claim 15, the operations further comprising:

while the second data is being obtained:

organizing, by the one or more processors of the nucleic acid sequencing device, the one or more first reads into respective groups based on at least a first sample identifier or a second sample identifier; and

generating, by the one or more processors of the nucleic acid sequencing device, organization statistics, the organization statistics indicating a number of first reads corresponding to each sample identifier.

Claim 17. The system of claim 13, the operations further comprising:

providing, by the one or more processors of the nucleic acid sequencing device, output data that represents the received alignment results of the data before or while aligning the second portion of the cluster of reads.

Claim 18. The system of claim 13, the operations further comprising:

causing, by the one or more processors of the nucleic acid sequencing device, the mapping and alignment engine to begin a subsequent alignment of the data representing the first plurality of reads to a reference sequence.

Claim 19. The system of claim 13, the operations further comprising:

while obtaining the second data, causing, by the one or more processors of the nucleic acid sequencing device, a secondary analysis engine to determine a set of likely variants for the first data representing the first plurality of reads that was aligned to a reference sequence.

Claim 20. The system of claim 13, wherein the at least a portion of the second data representing the second plurality of reads is aligned while at least a different portion of second data representing the second plurality of reads is being obtained.

Claim 21. The system of claim 13, wherein the mapping and alignment engine is instructed to begin alignment of the second data representing the second plurality of reads a predetermined number of sequencing cycles before the second data is completely obtained.

Claim 22. The system of claim 13, wherein the mapping and alignment engine is implemented using a field programmable gate array (FPGA).

Claim 23. The system of claim 13, wherein the mapping and alignment engine is implemented using an Application Specific Integrated Circuit (ASIC).

Claim 24. The system of claim 13, wherein the mapping and alignment engine is implemented by using one or more computers to execute software instructions.

Claim 25. A non-transitory computer-readable storage medium storing instructions that, when executed by one or more computers, causes the one or more processors of a nucleic acid sequencing device to perform operations, the operations comprising:

(i) obtaining first data describing a plurality of first reads generated by a nucleic acid sequencing device during a first read interval, wherein each read of the first reads represents a first ordered sequence of nucleotides

(ii) obtaining second data describing a plurality of second reads generated by the nucleic acid sequencing device during a second read interval that is performed after the first read interval, wherein each read of the second reads represents a second ordered sequence of nucleotides, wherein while the second data is being obtained, the operations further comprising:

(a) providing, by the nucleic acid sequencing device, the first data as an input to a mapping and alignment engine;

(b) receiving, from the mapping and alignment engine, alignment results that indicate a quality of an alignment of the first data; (c) determining, based on the received alignment results, whether to terminate sequencing operations; and

(d) causing the nucleic acid sequencing device to terminate operations that obtain the second data.

Claim 26. The computer-readable storage medium of claim 25,

wherein one or more of the first reads includes data representing a first sample identifier, and

wherein one or more of the second reads includes data representing a second sample identifier.

Claim 27. The computer-readable storage medium of claim 26, the operations comprising:

while the second data is being obtained:

organizing the one or more first reads into respective groups based on at least a first sample identifier or a second sample identifier; and

generating organization statistics, the organization statistics indicating a number of first reads corresponding to each sample identifier.

Claim 28. The computer-readable storage medium of claim 25, the operations further comprising:

providing output data that represents the received alignment results of first data before or while aligning the second portion of the cluster of reads.

Claim 29. The computer-readable storage medium of claim 25, the operations further comprising:

causing the mapping and alignment engine to begin a subsequent alignment of the data representing the first plurality of reads to a reference sequence.

Claim 30. The computer-readable storage medium of claim 25, the operations further comprising:

while obtaining the second data, causing a secondary analysis engine to determine a set of likely variants for the first data representing the first plurality of reads that was aligned to the reference sequence.

Claim 31. The computer-readable storage medium of claim 25, wherein the at least a portion of the second data representing the second plurality of reads is aligned while at least a different portion of second data representing the second plurality of reads is being obtained.

Claim 32. The computer-readable storage medium of claim 25, wherein the mapping and alignment engine is instructed to begin alignment of the second data representing the second plurality of reads a predetermined number of sequencing cycles before the second data is completely obtained.

Claim 33. The non-transitory computer-readable storage medium of claim 25, wherein the mapping and alignment engine is implemented using a field programmable gate array (FPGA).

Claim 34. The non-transitory computer-readable storage medium of claim 25, wherein the mapping and alignment engine is implemented using an Application Specific Integrated Circuit (ASIC).

Claim 35. The non-transitory computer-readable storage medium of claim 25, wherein the mapping and alignment engine is implemented by using one or more computers to execute software instructions.