[0015] FIG. 10 is another flowchart of an example method for performing a real-time secondary analysis.

[0016] FIGS. 11A and 11B compare an existing variant caller (FIG. 11A) to a variant caller that uses high confidence, low processing path as described herein (FIG. 11B).

DETAILED DESCRIPTION

[0017] In the following detailed description, reference is made to the accompanying drawings, which form a part hereof. In the drawings, similar symbols typically identify similar components, unless context dictates otherwise. The illustrative embodiments described in the detailed description, drawings, and claims are not meant to be limiting. Other embodiments may be utilized, and other changes may be made, without departing from the spirit or scope of the subject matter presented herein. It will be readily understood that the aspects of the present disclosure, as generally described herein, and illustrated in the Figures, can be arranged, substituted, combined, separated, and designed in a wide variety of different configurations, all of which are explicitly contemplated herein.

[0018] Disclosed herein are systems and methods for performing secondary analyses of nucleotide sequencing data in a time-efficient manner. In some embodiments, the method comprises performing a secondary analysis iteratively while sequence reads are generated by a sequencing system. Secondary analyses can encompass both alignment of sequence reads to a reference sequence (e.g., the human reference genome sequence) and utilization of this alignment to detect differences between a sample and the reference. Secondary analyses can enable detection of genetic differences, variant detection and genotyping, identification of single nucleotide polymorphisms (SNPs), small insertions and deletion (indels) and structural changes in the DNA, such as copy number variants (CNVs) and chromosomal rearrangements.

[0019] By performing secondary analyses while sequence reads are generated, the system and method can determine preliminary variant calls iteratively in real-time (or with zero or low latency). Final results of variant determinations may be available soon after (or immediately after) the end of a sequencing run. Alternatively, a sequencing run can be terminated early if variant calls are available with sufficient confidence during the run. In some embodiments, only information related to variant determinations (e.g., variant calls) is transferred off the sequencing system. This can decrease, or minimize,