Variant Callers

Variations in nucleotide sequence, in the form of single nucleotide variants (SNVs) and insertion and deletion events (INDELs), can exist within the germline or can be acquired by somatic alterations. Partek Flow provides pipeline creation tools to identify both SNVs and INDELs using aligned reads generated from targeted, whole exome, or whole genome DNA-Seq (or RNA-Seq) data. Detection of these variants can be performed by comparison against either the reference sequence utilized for alignment or among paired samples in a project. Tools for variant detection are performed on either *Aligned reads* or *Filtered reads* data nodes (Figure 1), and the *Detect variants* task node will produce a *Variants* data node. The *Variants* data node will contain Variant Call Format (vcf) files for each sample in the project. Three detection tools, each employing unique algorithms to identify variants in aligned sequence data, are available under the *Variant callers* section of the context sensitive menu:

- SAMtools
- FreeBayes
- LoFreq

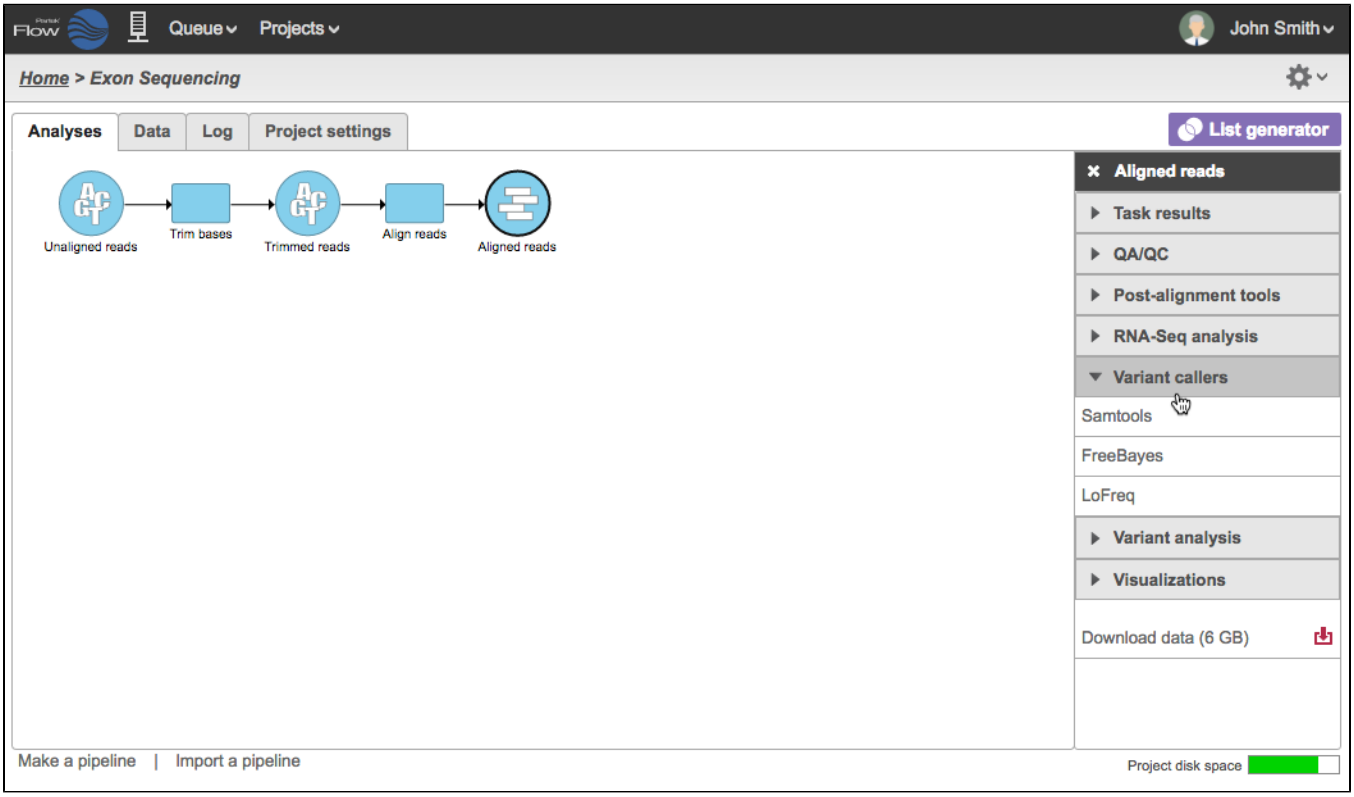

Figure 1. Showing Variant callers from an aligned reads node

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