



Hybrid-variant recognition is a process of identifying and classifying variants in a sample based on their sequence profile. This process involves several steps, including initial screening of the sample for specific variants, followed by sequencing and analysis of the variants to determine their characteristics and potential clinical significance. The final output of this process is a report that includes a detailed description of the variants found in the sample, along with their estimated frequency and any associated clinical information.

- **Initial screening:** This step involves identifying the presence of specific variants in the sample. This can be done using a variety of methods, including PCR, sequencing, and bioinformatics analysis. The goal of this step is to quickly identify any variants that may be present in the sample, so that they can be further analyzed.
- **Sequencing:** Once specific variants have been identified, the next step is to sequence them. This involves using a sequencing machine to determine the sequence of the variants. The goal of this step is to obtain a high-quality sequence of the variants, so that they can be analyzed in detail.
- **Analysis:** The final step in the hybrid-variant recognition process is to analyze the variants. This involves using bioinformatics tools to determine the characteristics of the variants, such as their estimated frequency and any associated clinical information. The goal of this step is to provide a detailed report that includes a description of the variants found in the sample, along with their estimated frequency and any associated clinical information.

Variant Type	Sequencing Step	Analysis Step
Known variants	Initial screening	Analysis
Novel variants	Initial screening	Analysis
Indeterminate variants	Initial screening	Analysis
Unknown variants	Initial screening	Analysis

- **Known variants:** These are variants that have been previously identified and characterized. They are typically found in the initial screening step, and their presence is often indicated by a high frequency of the variant in the sample.
- **Novel variants:** These are variants that have not been previously identified or characterized. They are typically found in the initial screening step, and their presence is often indicated by a low frequency of the variant in the sample.
- **Indeterminate variants:** These are variants that have not been previously identified or characterized, but their presence is indicated by a frequency of the variant in the sample that is not high enough to be considered a known variant.
- **Unknown variants:** These are variants that have not been previously identified or characterized, and their presence is indicated by a frequency of the variant in the sample that is not high enough to be considered a known variant.

Hybrid-variant recognition is a process of identifying and classifying variants in a sample based on their sequence profile.



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Kathleen Suttles Nehmer

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