

INVITED SPEAKER PRESENTATION

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# Helicos single molecule sequencing: unique capabilities and importance for molecular diagnostics

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## Background

Helicos Single Molecule Sequencing provides a unique view of genome biology through direct sequencing of cellular nucleic acids in an unbiased manner, providing both quantitative and accurate sequence information. The simple sample preparation involves no ligation or PCR amplification, allowing direct sequencing of targeted DNA or RNA molecules. DNA and RNA can be directly hybridized to the flow cell, eliminating many intermediary steps that can introduce sample loss or bias. From these methods, a diverse array of applications have been successfully demonstrated on the HeliScope Sequencer. For example, human genome sequencing for accurate variant detection; multiplexing of DNA for targeted resequencing; copy number variation studies from both fresh tumor tissue and formalin-fixed paraffin-embedded tissue samples; RNA-seq studies demonstrating the unexplored biology that exists in the transcriptome; small RNA studies leading to the identification of new classes of RNAs [1]; and the direct capture and sequencing of nucleic acid from as few as 400 cells with the end goal of single cell measurements [2]. Current efforts now focus on applying these unique attributes to develop novel substrate for molecular diagnostic tests.

## Ongoing programs

Recent studies identified the attributes of the Helicos Single Molecule Sequencing technology that provide unique features for molecular diagnostic application including combining the simplicity of sample preparation with sequencing, the scalability of the workflow and the quantitative power of the sequencing. Initially, Helicos is developing a molecular diagnostic test that will

identify gene mutations indicative of a woman's increased risk of developing hereditary breast or ovarian cancer. We are also investigating longer-term studies to examine the quantitative capabilities of our core technology in order to develop molecular diagnostic tests based on the detection and quantification of foreign DNA/RNA circulating in the bloodstream, such as a non-invasive prenatal diagnostic test.

## Acknowledgements

The work presented represents the collective, dedicated contributions of numerous Helicos scientists and collaborators.

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## References

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2. Ozsolak F, *et al*: Amplification-free Digital Gene Expression Profiling from Minute Cell Quantities. *Nature Methods* 2010, **7**:619-621.

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