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Office of Technology Management

ULTRA-RARE VARIANT DETECTION USING NEXT GENERATION SEQUENCING

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Next Generation Sequencing (NGS) allows rapid sequencing of an individual's entire genome or portions of the genome to identify thousands of genetic variants. Personalized genomic tests based on NGS technologies can be used to predict an individual's risk of disease or monitor disease progression or regression in the clinic.

The invention describes a complete workflow (sample preparation to data analysis) for the detection of ultra-rare sequence changes from human DNA at a 50 fold improvement in detection sensitivity using current low-cost NGS technologies.

Advantages

- Low cost, rapid test for ultra-rare sequence variants from human DNA
- Complete method that fits seamlessly into existing NGS workflows
- Detect leukemia in patient samples as early as 9 years before diagnosis
- Test for sequence variants from limiting DNA sources