

Snapshot: SARS-CoV-2 Sequencing, Polygenic Risk Scores, and Precision Medicine: High-throughput Solutions

Author

Christian Bixby, Sampled

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contact@sampled.com

Background

By accessing high-throughput solutions, researchers can gain more meaningful and in-depth data, which opens the door to more opportunities in the future. Here, two examples of situations where high-throughput testing was utilised to gain valuable insights will be explored.

SARS-CoV-2 sequencing

In April 2021, CDC contracted labs across the United States of America as part of a SARS-CoV-2 variant surveillance program. The samples had to be sequenced no more than 10 days from the time of collection to track the variants' progress in as real time as possible. This presented several challenges for Sampled to overcome:

Challenges to overcome	Solutions brought by Sampled
There would be a need to increase staff and equipment to scale from being able to complete 600 genomic sequences per week to 6000 within just 30 days.	Added 4 liquid handlers (Perkin Elmer G3 NGSx Sciclone) Increased staffing by 300%, split into two shifts
Sampled would need to select a high-throughput NGS library preparation kit that could meet this tight turnaround time.	Illumina COVIDSeq library prep kit was selected which enabled sequencing to be completed within 72hrs
A selection criterion would need to be established for the samples.	qPCR Cycle threshold cut-off was set to 30Ct.

These agile and proactive steps taken to meet these challenges enabled Sampled to contribute meaningful data to the CDC allowing the organisation to track emerging variants and their respective trends from April 2021 onwards.

Polygenic Risk Scores and Precision Medicine

Polygenic risk scores (PRS) are utilised to predict the likelihood of a patient developing polygenic diseases such as Alzheimer's disease, breast cancer, heart disease, and many more. Single nucleotide variants (SNV's) associated with the disease are investigated across the patient's whole genome. These genomic variants combined with clinical information (age, sex, weight, ethnicity, etc.) create an accurate risk percentage of the patient developing the disease. Sampled believes that this type of precision medicine will be utilized more frequently in the future, which is why we have invested in high-throughput solutions including:

- Microarrays and Next Generation sequencing instruments that can sequence thousands of samples a week and can genotype over a million SNVs.
- RT-PCR and qPCR instruments that can target up to a 100 SNVs and can process thousands of samples weekly.
- · As well as developing fully or semi-automated protocols and strong relationships with healthcare providers.

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