



## COSMOS: cloud enabled NGS analysis

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MEETING ABSTRACT

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# COSMOS: cloud enabled NGS analysis

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## Background

The dramatic fall of next generation sequencing (NGS) cost in recent years positions the price in range of typical medical testing, and thus whole genome analysis (WGA) may be a viable clinical diagnostic tool. Modern sequencing platforms routinely generate petabyte data. The current challenge lies in calling and analyzing this large-scale data, which has become the new time and cost rate-limiting step.

## Methods

To address the computational limitations and optimize the cost, we have developed COSMOS (<http://cosmos.hms.harvard.edu>), a scalable, parallelizable workflow management system running on clouds (e.g., Amazon Web Services or Google Clouds). Using COSMOS [1], we have constructed a NGS analysis pipeline implementing the Genome Analysis Toolkit - GATK v3.1 - best practice protocol [2,3], a widely accepted industry standard developed by the Broad Institute. COSMOS performs a thorough sequence analysis, including quality control, alignment, variant calling and an unprecedented level of annotation using a custom extension of ANNOVAR. COSMOS takes advantage of parallelization and the resources of a high-performance compute cluster, either local or in the cloud, to process datasets of up to the petabyte scale, which is becoming standard in NGS.

## Conclusion

This approach enables the timely and cost-effective implementation of NGS analysis, allowing for it to be used in a clinical setting and translational medicine. With COSMOS we reduced the whole genome data analysis cost under the \$100 barrier, placing it within a reimbursable cost point and in *clinical time*, providing a significant change

to the landscape of genomic analysis and cement the utility of cloud environment as a resource for Petabyte-scale genomic research.

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