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High-throughput clinical NGS data analysis on the cloud

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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. To address the computational limitations and optimize the cost, we have developed COSMOS, a scalable, parallelizable workflow management system running on cloud services. Using COSMOS, we have constructed a NGS analysis pipeline implementing the Genome Analysis Toolkit - GATK v3.1 - best practice protocol, 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. 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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Regular aspirin use and stomach cancer risk in China

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Epidemiological studies on aspirin and stomach cancer have been inconclusive. The purpose of our study was to investigate the association between aspirin and stomach cancer in China. A 1:2 matched case-control study was conducted in four large medical centers. A self-designed questionnaire was used to collect information. Unconditional logistic regression was used to compute crude and adjusted Odds Ratios (ORs) with 95% Confidence Intervals (CIs). Our study indicated that risk of stomach cancer was greatly reduced for regular aspirin user (OR=0.62; 95% CI 0.42-0.80). Specifically, dosage and tablet-years of use were associated with lower risk (OR=0.54; 95% CI 0.29–0.84 for ≥7 tabs/week; OR=0.47; 95% CI 0.32-0.81 for ≥10 tablet years, respectively). Furthermore, the finding was strengthened by stratified studies of gender, smoking status, Body Mass Index (BMI) and *Helicobacter pylori*. Our study confirmed that regular aspirin use is a protective factor to stomach cancer.

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