

## Machine Learning 2018: Using genomics cloud platform and machine learning for genome variant analysis- Tilila El Moujahid-Microsoft

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Cutting edge Sequencing (NGS) permits performing hugely paralleled DNA sequencing and is as of now reforming organic investigations. Rather than sequencing a specific set of qualities exclusively, NGS permits to grouping a more extensive bit of the genome (even an entire genome), which opens the entryway for a more extensive investigation of organic pathways inside a person. Analysts have at no other time gotten to such an abundance of genomic information which holds the guarantee of unveiling the privileged insights of the most overwhelming diseases of the century, for example, malignant growth. It additionally accompanies its own arrangement of difficulties for the administration and investigation of Big Data to remove important and significant bits of knowledge. Distributed computing and AI do have the ability to explain this test. In this discussion we will show how Cloud based Genomics stage permits to oversee petabytes of genomic information just as encourage quick and spry Secondary Analysis. We will likewise additionally use genomics explicit AI bundles to perform Tertiary Analysis on quality variations information and representation apparatuses to uncover and impart the outcomes to the scientific network. We will start the discussion with prologue to the genomics field and the usually utilized genomic examination process and will introduce down to earth uses of the above administrations and investigation.

The ability to sequence DNA provides researchers with the power to “read” the genetic blueprint that directs all the activities of a living organism. To provide context, the central dogma of biology is summarized as the pathway from DNA to RNA to Protein. DNA is composed of base pairs, based on 4 basic units (A, C, G and T) called nucleotides: A pairs with T, and C pairs with G. DNA is organized

into chromosomes and humans have a complete of 23 pairs.

Chromosomes are further organized into segments of DNA called genes which make or encode proteins. The sum of genes that an organism possess is named the genome. Humans have roughly 20,000 genes and three billion base pairs. Interestingly, only about 2 percent of the human genome encodes protein and this is often a key area of focus in research and therefore the business of genomics.

Genomics is closely related to Precision medicine. With a market size projected to reach \$87 billion by 2023, the field of Precision Medicine (also known as personalized medicine) is an approach to patient care that encompasses genetics, behaviors and environment with a goal of implementing a patient or population-specific treatment intervention; in contrast to a one-size-fits-all approach. For example, to scale back the danger of complications, a private who needs a transfusion would be matched to a donor who shares an equivalent blood group rather than a randomly selected donor.

Currently, there are two main barriers to greater implementation of precision medicine: High costs and technology limitations. To tackle the vast amount of patient data that has got to be collected and analyzed, and to assist hamper on costs many researchers are implementing machine learning techniques. Fortunately for researchers and genomics companies, the value of sequencing a genome continues to drop year-over-year – even after a huge relative plunge in cost between 2007 and 2012.

Biography:

Tilila is currently a Data Scientists and a Technical Evangelist for data and AI working at Microsoft. She accompanies partners in architecting and building their cloud based, AI powered solutions. She was previously a Technology Strategist for enterprise accounts including Education and healthcare industry. She's also a Fulbright scholar who earned, in 2012, a master in Computer Science and Business from San Francisco State university where her focus was on Bioinformatics including Genomics and Biomedical Image Analysis.

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