Nanofabrication and Genomics: How Patterning is Enabling a Revolution in Healthcare

M. Shane Bowen

Advanced Engineering Division, Department of Research and Technology Development
Illumina, Inc.
San Diego, CA USA 92122

E-mail: sbowen@illumina.com

According to the American Cancer Society, men have a 39.66% probability of developing cancer in their lifetime. For women, the odds are slightly lower at 37.65%. As cancer is a disease of the genome, having the ability to read the genome accurately is critical to not only gain and develop an understanding of the disease but also, it sets the foundation for identifying where the disease is affecting an individual and then being able to treat it.

In this presentation, I will provide an overview of illumina's sequencing technology and provide an example of how the power of nanofabricated substrates analogous to those being discussed throughout this conference, have effectively driven the technology to a realm of broad applicability and accessibility to the healthcare industry.

Over the duration of the human genome project (HGP) from 1990 to 2003, more than $2.7 billion dollars was spent across 20 global institutions resulting in the cataloging a fraction of a human genome at 8 -9 X coverage. This was a monumental achievement completed through the use of hierarchical shotgun method, which set the foundation upon which modern methods of genetic analysis stand. Since the completion of the HGP, numerous efforts have been put forth to increase the accuracy, simplify the process, reduce the amount of time and cost associated with measuring the sequence of any organism. This technology has become the foundation of modern medicine and is changing the way not only healthcare is performed but also revolutionizing agriculture, forensics and pharmaceutical development around the world.

Over the years of 2009 through 2014, I was part of a team of research scientists at illumina who developed a method to sequence a human genome in less than 48 hours at a coverage of 30X and at the price of $1000. During this talk, I will give an overview of the technologies developed to make this happen as well as set the stage for the next iteration of innovations paving the path to the $100 genome, being delivered to the public with the NovaSeq platform commercialized by illumina in 2017. We will also use this time to look at complementary substrate formats that can similarly be realized with micro and nanofabrication methodologies that are becoming similarly important for the health industry in order to usher in the age of precision medicine.

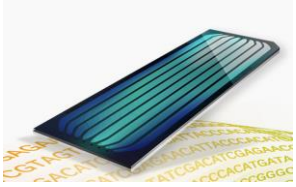

Figure 1. illustration of a HiSeq X, illumina's first patterned flowcell commercialized in 2014 and

brought to market the $1000 genome

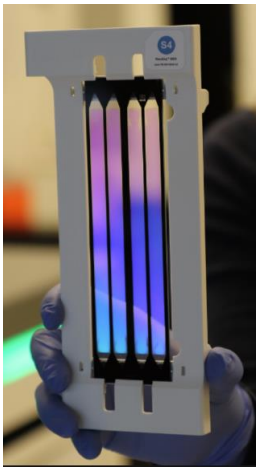

Figure 2. Photograph of the NovaSeq 6000 patterned flowcell, commercialized in 2017, and on the trajectory to deliver the $100 genome.