

## CLINICAL MOLECULAR DIAGNOSIS IN THE ERA OF NEXTGEN SEQUENCING.

*Fan Chen, Steve S. Sommer*

*MEDomics, Azusa, California, USA*

NextGen sequencing is increasing throughput 1,000<sup>+</sup> fold, facilitating the second most important paradigm shift in the history of medicine. The focus of medicine will soon shift from treating the sick individual to managing the genetic risks in the family to delay or prevent disease. There are four NextGen sequencing platforms that are currently available (454, Solexa, SOLiD, and Helicos) and another should come to market soon (Pacific Biosystems). Each platform has its sweet spot. The advantages and disadvantages of each system will be reviewed in the context of their epidemiological and diagnostic applications in mitochondrial medicine and oncology.

### **Key words:**

NextGen sequencing, molecular diagnostics, mitochondrial medicine