DEPLOYING LOW-COVERAGE WHOLE GENOME SEQUENCING IN THE PRACTICE: A TRANSLATIONAL JOURNEY FROM BIOINFORMATICS RESEARCH TO CLINICAL TEST

ABSTRACT

Low-Coverage Whole Genome Sequencing (LC-WGS) has recently been implemented in clinical laboratories at Mayo. It is one of the sequencing protocols that have been translated into clinical applications. The first successful test is the Non-Invasive Prenatal Test that is becoming routinely used to detect fetal trisomy. Other clinical applications are underway. I will describe the analytical workflows put in place to analyze LC-WGS data and report trisomy. I will also describe how the analytics has been extended to the stratification of ovarian tumors. This new application of LC-WGS is currently being translated into a clinical test.