TITLE: Implementation of whole genome sequencing at the Health and Environmental Testing Laboratory

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BACKGROUND: With the cost of sequencing rapidly falling below $500 per sample, the use of whole genome sequencing (WGS) to detect and characterize bacterial and viral infections is becoming more commonplace. There is a major push by American Society of Microbiology (ASM), Federal CDC, American Public Health Laboratory (APHL), Food and Drug Administration (FDA), American College of Medical Genetics and Genomics (ACMG), Clinical Laboratory Improvement Amendments (CLIA), College of American Pathologists (CAP), to both implement and regulate whole genome sequencing in the clinical laboratory setting. WGS is a paradigm shift in laboratory testing; instead of a multitude of equipment and protocols, WGS is a diagnostic one stop shop for infectious disease and cancer screening.

METHODS: Total chromosomal and plasmid nucleic acid were extracted via the Qiagen DNA mini kit from previously qualified bacterial isolates. Genomic material from gram-positive bacteria was extracted using an in-house method. Whole genome sequencing was performed on the Illumina MiSeq sequencing platform using Illumina's NexteraXT protocols and reagents. Multiple plug-n-play bioinformatics workflows for the characterization of bacterial isolates; serotyping, MLST, resistance genes, virulence genes, and bacterial taxonomy were validated using Clinical Laboratory Improvement Amendments (CLIA) and The American College of Medical Genetics and Genomics (ACMG) guidelines.

RESULTS: HETL has validated the use of the Illumina MiSeq whole genome sequencer and implemented its use for the molecular epidemiological analysis using a variety of web based bioinformatics suites for the detection of infectious disease with a focus on bacterial isolates.

CONCLUSIONS: HETL now routinely uses whole genome sequencing for *E.coli* and *Salmonella* serology, bacterial taxonomy, Pulsenet foodborne surveillance and, antibiotic resistance and virulence factor genes. This technology has supplanted or enhanced traditional methods of serotyping, bacterial identification, and other strain typing methods.