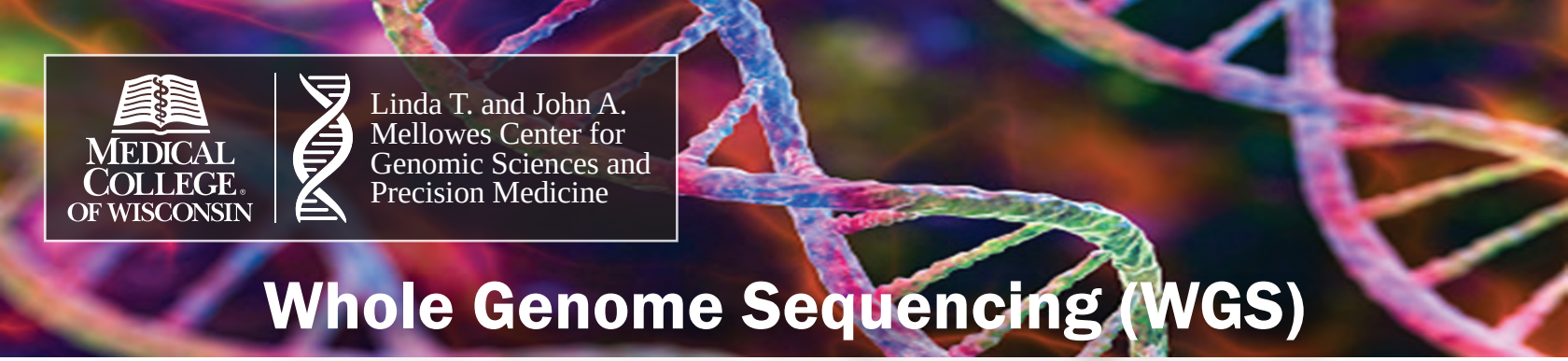

MEDICAL COLLEGE OF WISCONSIN

Linda T. and John A. Mellowes Center for Genomic Sciences and Precision Medicine

Whole Genome Sequencing (WGS)

Test Description

Whole Genome Sequencing (WGS) is a next-generation sequencing method used to comprehensively consider the DNA base pairs of a sample and evaluate variants. Variants are compared to clinical and research databases that annotate the role of base pair changes as pathogenic, benign, or unknown significance. The role of genomic variants in disease phenotypes and outcomes can be further understood by evaluating transcript expression, protein structure and dynamics, and epigenetic regulation. WGS is available for any species of interest including, but not limited to, human, mice, rats, mammals, plants, microbes and more.

Sample Types and Requirements

Whole Blood: 2-4mL in an EDTA tube (purple top)
Genomic DNA: 5-10µg with a minimum concentration of 75ng/µL, and 260/280 ratio of 1.75-2.0
Saliva, Cell Pellets, FFPE, Tissue: Consultation with the lab is requested, quality and quantity of DNA will be verified at the Mellowes Center prior to initiation of library preparation

Recommended Sequencing Depth

Equal distribution of total reads among all samples in the pool, aiming for ~35x depth of coverage, paired end, 2x150 base pair sequencing

Submission Requirements

Sample Intake Form and iLabs request. Contact lab for drop off or shipping requirements.

Turnaround Time

4-6 weeks for fastq files only
2-3 additional weeks for bioinformatics analysis

Deliverables

DNA-seq report (html with linked documents)
Variant call files (VCFs) with annotation and population frequency, BAM, fastq.gz files
All NGS data files will be delivered via Mellowes Center portal

Test Methodology

Isolated genomic DNA is fluorescently quantified using the BioTek Synergy LX or Qubit and, if necessary, quality assessed using the Agilent Fragment Analyzer. WGS libraries are then prepared according to the Illumina DNA preparation with tagmentation. The quality and quantity of the DNA library is checked by fragment analysis and qPCR respectively. The pooled library is sequenced on the Illumina NovaSeq.

Bioinformatic Core Analysis

WGS report includes:

- Quality control and sequencing metrics (FastQC)
- Filtering to focus on variants
- Comprehensive identification of alterations including single nucleotide variants (SNV), insertions and deletions (Indels)
- Annotation with germline and somatic variant resources
- Classification workflow that separates variant calls into pathogenic or unknown significance (VUS) groups
- Comparison among genetically or disease related samples to identify shared or novel variants in genes and pathways
- Additional analysis can evaluate the potential for the variant to alter protein structure, function, dynamics, or expression

Contact & Submission

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