
Lab Dept: **Anatomic Pathology**

Test Name: **RAPID (EXPEDITED) WHOLE EXOME
SEQUENCING - TRIO (WES)**

General Information

Lab Order Codes: RWES

Synonyms: N/A

CPT Codes: 81415 - Exome Sequence analysis

Test Includes: * DNA extraction (if necessary), DNA integrity testing, Library preparation, data generation
* Variant analysis of sequence data files using the Carpe Novo system or commercial equivalent
* Confirmation of variants via Sanger sequencing
* Interpretation of results

Logistics

Test Indications: Whole Exome Sequencing (WES) is used to detect variants in a patient's exome in order to determine the role of genomic variants in disease outcomes. The exome is a little more than 1% of the genome that codes for protein. The patient's exome will be sequenced to an average depth of 100X with a minimum depth of coverage of 85X. Over 97% of the exome will be sequenced to the depth of 10X. The mitochondrial genome of the patient will be sequenced to a minimum depth of 20X.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Medical College of Wisconsin

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Performed Monday – Friday, 30-45 days (expedited)

Special Instructions: Complete and submit Sample Submission form with specimen:
http://www.mcw.edu/FileLibrary/Groups/HMGC/Forms/DNLSeq_ServiceRequest.pdf

Specimen

Specimen Type:	Whole blood
Container:	Lavender (EDTA) tube
Draw Volume:	2 – 4 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	Lab Staff: Invert specimen several times to mix blood. Sent specimen in original tube. Ship overnight at room temperature in an insulated container within 5 days of collection. Indicate on the Sample submission form: RAPID WESTRIO.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimen

Interpretive

Reference Range:	An interpretive report will be provided.
Critical Values:	N/A
Limitations:	N/A
Methodology:	Exome sequencing
References:	Medical College of Wisconsin January 2017