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/DNLSex_ServiceRequest.pdf](http://www.mcw.edu/FileLibrary/Groups/HMGC/Forms/DNLSex_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**

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<th><strong>Reference Range:</strong></th>
<th>An interpretive report will be provided.</th>
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<tr>
<td><strong>Critical Values:</strong></td>
<td>N/A</td>
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<td><strong>Limitations:</strong></td>
<td>N/A</td>
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<td><strong>Methodology:</strong></td>
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<td><strong>References:</strong></td>
<td><a href="https://example.com">Medical College of Wisconsin</a> January 2017</td>
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