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**Lab Dept:**                   **Anatomic Pathology**

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

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***General Information***

**Lab Order Codes:**       WESTR

**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

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***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, 90 days maximum (regular TAT).

**Special Instructions:**    Complete and submit Sample Submission form with specimen:  
[http://www.mcw.edu/FileLibrary/Groups/HMGC/Forms/DNLSeq\\_ServiceRequest.pdf](http://www.mcw.edu/FileLibrary/Groups/HMGC/Forms/DNLSeq_ServiceRequest.pdf)

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***Specimen***

<b>Specimen Type:</b>	Whole blood.
<b>Container:</b>	Lavender (EDTA) tube
<b>Draw Volume:</b>	2-4 mL (Minimum 1 mL) blood
<b>Processed Volume:</b>	Same as Draw Volume
<b>Collection:</b>	Routine venipuncture
<b>Special Processing:</b>	Lab Staff: Invert specimen several times to mix blood. Send specimen in original tube. Ship overnight at room temperature in an insulated container within 5 days of collection.
<b>Patient Preparation:</b>	None
<b>Sample Rejection:</b>	Mislabeled or unlabeled specimens

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### ***Interpretive***

<b>Reference Range:</b>	An interpretive report will be provided.
<b>Critical Values:</b>	N/A
<b>Limitations:</b>	N/A
<b>Methodology:</b>	Exome Sequencing
<b>References:</b>	<a href="#">Medical College of Wisconsin</a> January 2017