**Reporting Title:** Whole Exome Sequencing  
**Performing Location:** Rochester  

**Ordering Guidance:**  
The American College of Medical Genetics and Genomics recommends that whole exome sequencing be considered as a first-tier or second-tier test for patients with one or more congenital anomalies, or developmental delay or intellectual disability with onset prior to age 18 years.(1)  

If a specific diagnosis is suspected, single gene or panel testing may be a more appropriate first-tier testing option.  

**This test is for affected patients (probands) only.** For family member specimens being sent as comparators, order CMPRE / Family Member Comparator Specimen for Exome Sequencing, Varies. If this test is ordered on a family member comparator, this test will be canceled and CMPRE performed as the appropriate test.  

This test cannot support detection of deep intronic variants, trinucleotide repeat variants, or variants in the mitochondrial genome.  
- For whole exome sequencing plus analysis of the mitochondrial genome, order WESMT / Whole Exome and Mitochondrial Genome Sequencing, Varies.  
- If separate mitochondrial genome testing is needed, order MITOP / Mitochondrial Full Genome Analysis, Next-Generation Sequencing (NGS), Varies.  

This test is not appropriate for identification of somatic variants in solid tumors. If this testing is needed, order MCSTP / MayoComplete Solid Tumor Panel, Next-Generation Sequencing, Tumor.  

This testing does not provide genotyping of patients for pharmacogenomic purposes. For an assessment for genes with strong drug-gene associations, order PGXQP / Focused Pharmacogenomics Panel, Varies.  

Targeted testing for familial variants (also called site-specific or known variant testing) is available for variants identified by this test. See FMTT / Familial Mutation, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.  

Prenatal specimens (amniocentesis or chorionic villi) are not currently accepted for this test.  

**Additional Testing Requirements:**  
To order whole exome testing for the patient and the family member comparator specimens, see the following steps:  
1. Order this test on the patient (proband).  
2. Order CMPRE / Family Member Comparator Specimen for Exome Sequencing, Varies on all family members’ specimens being submitted as comparators.  
   a. When available, the patient’s biological mother and biological father are the preferred family member comparators.  
   b. If one or both of the patient’s biological parents are not available for testing, specimens from other first-degree relatives (siblings or children) can be used as comparators. Contact the laboratory at 800-533-1710 for approval to send specimens from other relatives.  
   c. The cost of analysis for family member comparator specimens is applied to the patient’s (proband’s) test. Family members will not be charged separately.  
3. Collect patient (proband) and family member specimens. Label specimens with full name and birthdate. Do not label
family members’ specimens with the proband’s name.
5. If the patient wishes to opt-out of receiving secondary findings or change the DNA storage selection, select the appropriate boxes in the Informed Consent section.
6. Attach clinic notes from specialists relevant to patient’s clinical features, if available.
7. Attach pedigree, if available.
8. Send paperwork to the laboratory along with the specimens. If not sent with the specimen, fax a copy of the paperwork to 507-284-1759, Attn: WES Genetic Counselors.

For more information see Whole Exome Sequencing (WES): Questions and Answers for Providers.

Shipping Instructions:
Specimen preferred to arrive within 96 hours of collection.

Necessary Information:
1. Whole Exome Sequencing: Ordering Checklist is required. Fill out one form for the family and send with the specimens.
2. Prior Authorization is available for this test. Submit the required form with the specimen.

Specimen Requirements:
Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.
Specimen Stability Information: Ambient (preferred)/Refrigerated
Additional Information: If a cord blood specimen is received, MATCC / Maternal Cell Contamination, Molecular Analysis, Varies will be performed at an additional charge.

Specimen Type: Skin biopsy
Supplies: Fibroblast Biopsy Transport Media (T115)
Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.
Specimen Volume: 4-mm punch
Specimen Stability Information: Refrigerated (preferred)/Ambient
Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular
Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

**Specimen Type:** Cultured fibroblast  
**Container/Tube:** T-25 flask  
**Specimen Volume:** 2 Flasks  
**Collection Instructions:** Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.  
**Specimen Stability Information:** Ambient (preferred)/Refrigerated (<24 hours)  
**Additional Information:** A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

**Specimen Type:** Blood spot  
**Supplies:** Card-Blood Spot Collection (Filtration Paper) (T493)  
**Container/Tube:**  
  **Preferred:** Collection card (Whatman Protein Saver 903 Paper)  
  **Acceptable:** PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card  
**Specimen Volume:** 5 Blood spots  
**Collection Instructions:**  
1. An alternative blood collection option for a patient older than 1 year of age is fingerstick. For infants younger than 1 year, a heel stick should be used. See [How to Collect Dried Blood Spot Samples](#) via fingerstick.  
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.  
3. Do not expose specimen to heat or direct sunlight.  
4. Do not stack wet specimens.  
5. Keep specimen dry.  
**Specimen Stability Information:** Ambient (preferred)/Refrigerated  
**Additional Information:**  
1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.  
2. For collection instructions, see [Blood Spot Collection Instructions](#)  
3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)  
4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)

**Specimen Type:** Saliva  
**Patient Preparation:** Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.  
**Supplies:** Saliva Swab Collection Kit (T786)  
**Specimen Volume:** 1 Swab  
**Collection Instructions:** Collect and send specimen per kit instructions.  
**Specimen Stability Information:** Ambient 30 days  
**Additional Information:** Due to lower concentration of DNA yielded from saliva, it is possible that additional specimen may be required to complete testing.

**Forms:**  
1. [Whole Exome Sequencing: Ordering Checklist](#) is required.  
2. New York Clients-Informed consent is required, included in the above form. Document on the request form or electronic order that a copy is on file.
3. **Whole Exome Sequencing (WESDX) Prior Authorization Ordering Instructions**

4. If not ordering electronically, complete, print, and send a Neurology Specialty Testing Client Test Request (T732) with the specimen.

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**Result Codes:**

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LOINC® and CPT codes are provided by the performing laboratory.

**Supplemental Report:**

Supplemental

**CPT Code Information:**

- 81415-Patient only
- 81415, 81416-Patient and one family member comparator sample (duo) (as appropriate)
- 81415, 81416 x 2-Patient and two family member comparator samples (trio or non-traditional trio) (as appropriate)
- 81415, 81416 x 3-Patient and three family member comparator samples (quad) (as appropriate)

**Reflex Tests:**

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<th>CPT Units</th>
<th>CPT Code</th>
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<td>MATCC</td>
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**Result Codes for Reflex Tests:**

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Test Definition: WESDX
Whole Exome Sequencing for Hereditary Disorders, Varies

Reference Values:
An interpretive report will be provided.