Test Definition: WESPR

Panel to Whole Exome Sequencing Reflex Test, Varies

Overview

Useful For

Serving as a second-tier test for patients in whom previous genetic testing was negative or inconclusive

Identifying causative variants in genes that were not included on panel testing which can allow for:
-Better understanding of natural history/prognosis
-Targeted management (anticipatory guidance, management changes, specific therapies)
-Predictive testing of at-risk family members
-Testing and exclusion of disease in siblings or other relatives
-Recurrence risk assessment

Additionally, this testing may be useful in the context of a patient's evolving clinical features.

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
_STR1	Comp Analysis using STR (Bill only)	No, (Bill only)	No
_STR2	Add'l comp analysis w/STR (Bill Only)	No, (Bill only)	No
CULFB	Fibroblast Culture for Genetic Test	Yes	No
MATCC	Maternal Cell Contamination, B	Yes	No
G237	Number of Comparators for WESPR	No	No

Genetics Test Information

Whole exome sequencing utilizes next-generation sequencing (NGS) to detect variants within the protein-coding regions of approximately 20,000 genes. In patients who have had negative or inconclusive postnatal hereditary gene panel testing, analysis of previously generated sequencing data, expanded to include the whole exome has the potential to identify new or additional variants associated with the patient's phenotype and increase the diagnostic yield of testing.

This test is available for patients who have had hereditary gene panel testing performed on a postnatal sample via NGS utilizing the Integrated DNA Technologies chemistry performed by Mayo Clinic Laboratories and would like to expand to whole exome sequencing. Most hereditary gene panels performed at Mayo Laboratories since 2023 are eligible for this test to be added. In addition, some hereditary gene panels performed between 2021 and 2023 are eligible. To confirm that it is possible to add this test for a specific patient, contact the laboratory at 800-533-1710.

It is strongly recommended that testing is performed alongside specimens submitted from the patient's biological mother and the patient's biological father as part of a trio analysis. However, testing for singletons (patient only), duos (patient and one relative to be used as a comparator), and non-traditional trios (patient and two relatives to be used as comparators) will also be accepted if the patient's biological parents are not available for testing.

This test may be ordered by the healthcare professional who ordered the original hereditary NGS panel or by a new healthcare professional if the patient is currently under their care. Results will be sent only to the individual who placed the current order.

Testing Algorithm

Skin biopsy or cultured fibroblast specimens:
For skin biopsy or cultured fibroblast specimens, a fibroblast culture will be performed at an additional charge. If viable cells are not obtained, the client will be notified.

Cord blood:
For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge.

Special Instructions

- Whole Exome Sequencing: Ordering Checklist
- Blood Spot Collection Card-Spanish Instructions
- Blood Spot Collection Card-Chinese Instructions
- Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm
- Blood Spot Collection Instructions
- Whole Exome and Genome Sequencing Information and Test Ordering Guide

Highlights

This test expands variant analysis from the targeted regions initially evaluated in panel testing previously performed by Mayo Clinic Laboratories using next-generation sequencing (NGS) to include protein-coding regions of approximately 20,000 genes (the exome). Expanding to whole exome sequencing offers a potentially cost-effective alternative to establishing a molecular diagnosis compared to performing multiple independent molecular assays.

Method Name

Reanalysis of Whole Exome Next-Generation Sequencing (NGS) followed by Sanger Sequencing or Quantitative Polymerase Chain Reaction (qPCR), as needed

NY State Available

Yes

Specimen

Specimen Type

Varies

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)

This test is only appropriate for patients who have had hereditary gene panel testing performed on a postnatal sample via next-generation sequencing (NGS) utilizing the Integrated DNA Technologies chemistry performed by Mayo Clinic Laboratories. To confirm that this test is possible for a specific patient, contact the laboratory at 800-533-1710.

If the patient has not had an appropriate test previously performed by Mayo Clinic Laboratories but whole exome sequencing is desired, order either WESDX / Whole Exome Sequencing for Hereditary Disorders, Varies or WESMT / Whole Exome and Mitochondrial Genome Sequencing, Varies. If whole genome sequencing is desired, order WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies. A new specimen may be required.

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

This test cannot support detection of deep intronic variants, trinucleotide repeat variants, or variants in the mitochondrial genome.
-If separate mitochondrial genome testing is needed, order MITOP / Mitochondrial Full Genome Analysis, Next-Generation Sequencing (NGS), Varies.
-For detection of single nucleotide variants, small insertions and deletions, copy number variants, deep intronic variants, trinucleotide repeat variants, and mitochondrial genome variants, order WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies.

This test is **not appropriate for** identification of somatic mutations in solid tumors. If this testing is needed, order MCSTP / MayoComplete Solid Tumor Panel, Next-Generation Sequencing, Tumor. A new specimen may be required.

This test 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. A new specimen may be required.

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

Additional Testing Requirements

Patient DNA is required to allow for confirmation of any new reportable variants, based on internal laboratory criteria. For most patients, stored DNA from the original panel test should be available for this testing. If a DNA sample is depleted or discarded, testing will proceed, however, a new sample will be requested from the ordering provider to

attempt any necessary confirmatory testing. If a new sample is not provided, any findings that require confirmation will be reported with a disclaimer that confirmation was not performed due to lack of a DNA specimen.

To order whole exome sequencing for the patient and family member comparator specimens after a negative or inconclusive hereditary next-generation sequencing gene panel performed at Mayo Clinic Laboratories, perform the following steps:

1. Order WESPR / Panel to Whole Exome Sequencing Reflex Test, Varies
2. Call Mayo Clinic Laboratories at 800-533-1710 to request that the remaining DNA specimen be added to the WESPR order.
3. Complete the required paperwork and informed consent: Whole Exome Sequencing: Ordering Checklist.
4. Attach clinic notes from specialists relevant to patient's clinical features, if available.
5. Attach pedigree, if available.
6. If submitting family member comparator samples, order CMPRE / Family Member Comparator Specimen for Exome Sequencing, Varies for each family member.
   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, non-first-degree 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.
7. If needed, collect specimens. Label specimens with full name and birthdate. **Do not** label family members' specimens with the proband's name.
8. Send paperwork to the laboratory along with the specimens. If not sent with the specimen or if no specimen is being sent, fax a copy of the paperwork to 507-284-1759, Attn: WES Genetic Counselors.

For more information see Whole Exome and Genome Sequencing Information and Test Ordering Guide.

Necessary Information

Whole Exome Sequencing: Ordering Checklist is required. Fill out one form for the family and send with the specimens or fax to 507-284-1759, Attn: WES Genetic Counselors.

Specimen Required

For most patients, a new specimen submission will not be required. Testing can be performed using stored DNA from the original whole exome sequencing test. To order testing on the stored specimen, see Additional Testing Requirements.

Patient Preparation: A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Green top (sodium heparin)
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot**.
3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information
Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days
Additional Information:
1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA are met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.
3. For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.

Specimen Type: Saliva
Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.
Supplies:
DNA Saliva Kit High Yield (T1007)
Saliva Swab Collection Kit (T786)
Container/Tube:
Preferred: High-yield DNA saliva kit
Acceptable: Saliva swab
Specimen Volume: 1 Tube if using T1007 or 2 swabs if using T786
Collection Instructions: Collect and send specimen per kit instructions.
Specimen Stability Information: Ambient (preferred) 30 days/Refrigerated 30 days
Additional Information: Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

Specimen Type: Blood spot
Supplies: Card-Blood Spot Collection (Filter Paper) (T493)
Container/Tube:
Preferred: Collection card (Whatman Protein Saver 903 Paper)
Acceptable: PerkinElmer 226 filter paper or blood spot collection card
Specimen Volume: 2 to 5 Blood spots
Collection Instructions:
1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see How to Collect a Dried Blood Spot Sample.
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. Blood spot specimens are acceptable but not recommended. Multiple extractions will be required to obtain sufficient yield for supplemental analysis, and there is significant risk for test failure due to insufficient DNA.
2. Due to lower concentration of DNA yielded from blood spot, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.
3. For collection instructions, see Blood Spot Collection Instructions
4. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
5. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

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: Ambient (preferred) <24 hours/Refrigerated <24 hours

Additional Information:

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblasts

Source: Skin or tissue

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin or tissue biopsy.

Specimen Stability Information: Ambient (preferred) <24 hours/Refrigerated <24 hours

Additional Information:

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts before genetic testing can occur.

Specimen Type: Extracted DNA

Container/Tube:

Preferred: Screw Cap Micro Tube, 2 mL with skirted conical base

Acceptable: Matrix tube, 1 mL

Collection Instructions:

1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
2. Include concentration and volume on tube.

Specimen Stability Information: Frozen (preferred) 1 year/Ambient/Refrigerated
Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

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. If not ordering electronically, complete, print, and send 1 of the following forms with the specimen:
-Neurology Specialty Testing Client Test Request (T732)
-Cardiovascular Test Request (T724)

Specimen Minimum Volume

See Specimen Required

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical & Interpretive

Clinical Information

Whole exome sequencing utilizes next-generation sequencing to assess patients with suspected underlying genetic disorders for variants within the protein-coding regions (exons and splice junctions) of approximately 20,000 genes simultaneously. Based on a meta-analysis, the diagnostic utility of whole exome sequencing is approximately 36%.(1)

This test expands variant analysis from the targeted regions initially evaluated via panel testing to include protein-coding regions of approximately 20,000 genes (the exome). Variants are evaluated for pathogenicity and reported if they are suspected to be related to the individual's reason for testing, or if they are a medically actionable secondary finding, unless the patient opts out of secondary findings via the consent form contained within the Whole Exome Sequencing: Ordering Checklist.

Indications for whole exome sequencing include but are not limited to:(1,2)
-Patients with one or more congenital anomalies
-Patients with developmental delay or intellectual disability with onset prior to age 18 years
-Patients with a phenotype and/or family history that strongly suggests an underlying genetic cause, yet genetic tests for

that phenotype have failed to arrive at a diagnosis (diagnostic odyssey)
-Patients with a phenotype and/or family history that strongly suggests an underlying genetic cause, but the phenotype does not fit with one specific disorder (numerous individual genetic tests would be required for evaluation)
-Patients with a suspected genetic disorder that has numerous underlying genetic causes, making analysis of numerous genes simultaneously a more practical approach than single-gene testing (condition is genetically heterogeneous)
-Patients with a suspected genetic disorder for which specific molecular genetic testing is not yet available
-Patients with an atypical presentation of a genetic disorder

It is strongly recommended that specimens are also submitted from the patient's biological mother and biological father, which are used for comparison purposes (trio analysis). Based upon published reports, a diagnosis is identified in trio-based whole exome sequencing (WES) in approximately 25% to 37% of cases, with slightly lower diagnostic yield in non-trio WES.(3-5) However, testing for singletons (patient only), duos (patient and one family member to be used as a comparator), and non-traditional trios (patient and two family members to be used as comparators) will also be accepted if both biological parents are unavailable.

For more information see Whole Exome and Genome Sequencing Information and Test Ordering Guide.

Reference Values

An interpretive report will be provided.

Interpretation

Variants of interest are evaluated according to American College of Medical Genetics and Genomics (ACMG) recommendations.(4) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Variants are reported in one of the following categories:
-Likely Causative: Variants with a high degree of suspicion for causing the patient's reported clinical features
-Possibly Relevant: Variants that may be related to the patient's clinical features or variants in genes of uncertain significance
-Secondary Findings: Medically actionable variants unrelated to the indication for testing (see below for additional information)
-Previously Reported: Variants that were reported on the original panel test report but do not reach the reporting criteria to be included in the above categories.

It is possible that a variant may not be recognized as the underlying cause of disease due to incomplete scientific knowledge about the function of all genes in the human genome or the impact of variants in those genes.

Variants previously identified through testing performed at other laboratories, via different methodologies, or different ordered services may or may not be rereported in this analysis. The inclusion of such variants in the final report is at the discretion of the laboratory based on clinical relevance, current interpretation standards, and the context of the patient's phenotype. Variants may be re-reported using a different genome build or transcript that utilizes alternative nomenclature; similarly, reported breakpoints for copy number variants may differ due to differences in testing methodologies.

Secondary Findings:
Patients are evaluated for medically actionable secondary findings and these findings are reported in accordance with the most current ACMG recommendations, including the most up-to-date gene list.(5) Variants in these genes will not be evaluated or reported if the patient opts out of this evaluation unless they overlap with the patient's reported clinical phenotype.

The presence of a variant in family member comparator specimens is stated on the patient's (proband's) report unless family members opt out of secondary findings. If the patient (proband) opts out, secondary findings will not be reported for any family member.

Variants that are present in family members comparators but absent from the patient (proband) are not evaluated.

The absence of a reportable secondary finding does not guarantee that there are no disease-causing or likely disease-causing variants in these genes, as portions of the genes may not be adequately covered by this testing methodology.

Reanalysis and Raw Data Requests:
Patient data is not guaranteed to be stored indefinitely. Requests for reanalysis or release of raw data may not be possible, and a new whole exome sequencing order may be required if the original patient data is no longer available or no longer compatible with current bioinformatics processes or analysis tools.

Reanalysis of the patient's exome due to new patient clinical features, advances in genetic knowledge, or changes in testing methodology is available. See test WESR/ Whole Exome Sequencing Reanalysis, Varies or contact the laboratory at 800-533-1710 for more information.

Requests for the raw data obtained from whole exome sequencing should be directed to the laboratory. A separate fee may apply. Raw data will be released for individuals who complete a Mayo Clinic release of information form. If raw data for family member comparators is requested, it will only be released with an accompanying request for the proband's raw data. Contact the laboratory for instructions on completing the release of information form. The laboratory is not responsible for providing software or other tools needed to visualize, filter, or interpret this data.

Cautions

Clinical Correlations:
Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

To discuss the availability of additional testing options or for assistance in the interpretation of these results, contact Mayo Clinic Laboratories genetic counselors at 800-533-1710.

Technical Limitations:
Whole exome sequencing may not detect all types of genomic variants. In rare cases, false-negative or false-positive results may occur. The depth of coverage may be variable for some target regions. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis (as applicable) as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. Confirmation of select reportable variants will be performed by alternate methodologies based on internal laboratory criteria.

If a DNA specimen is no longer available for confirmation, Mayo Clinic Laboratories will contact the client and ask that they submit a new sample to be used for this purpose. If no sample has been received by the time testing is ready to be reported, results will be reported with a disclaimer that confirmation studies were not performed.

This test is not designed to detect low levels of mosaicism or differentiate between somatic mutations and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent non-leukocyte reduced blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reclassification of Variants:
The classification of all previously reported variants will be reassessed at the time of reporting. Once reported, it is not standard practice for the laboratory to systematically review previously classified variants on a regular basis. The laboratory encourages healthcare professionals to contact the laboratory at any time to learn how the classification of a particular variant may have changed over time. Due to broadening genetic knowledge, it is possible that the laboratory may discover new information of relevance to the patient. Should that occur, the laboratory may issue an amended report.

Variant Evaluation:
Evaluation and categorization of variants are performed using published American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology recommendations as a guideline.(4) Other gene-specific guidelines may also be considered. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. Variants classified as benign or likely benign are not reported.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and periodic updates to these tools may cause predictions to change over time. Results from in silico evaluation tools should be interpreted with caution and professional clinical judgment.

Rarely, incidental or secondary findings outside of the genes recommended by the ACMG may implicate another predisposition or presence of active disease. These findings will be carefully reviewed to determine whether they will be reported.

Data Sharing:
Deidentified variant information may be shared in public genetic databases, such as GeneMatcher or ClinVar

Clinical Reference

1. Clark MM, Stark Z, Farnaes L, et al. Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. NPJ Genom Med. 2018;3:16. doi:10.1038/s41525-018-0053-8
2. Deignan JL, Chung WK, Kearney HM, et al. Points to consider in the reevaluation and reanalysis of genomic test results: A statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2019;21(6):1267-1270
3. Tan NB, Stapleton R, Stark Z, et al. Evaluating systematic reanalysis of clinical genomic data in rare disease from single center experience and literature review. Mol Genet Genomic Med. 2020;8(11):e1508
4. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015;17(5):405-424
5. Miller DT, Lee K, Gordon AS, et al. Recommendations for reporting of secondary findings in clinical exome and genome sequencing, 2021 update: a policy statement of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021;23(8):1391-1398. doi:10.1038/s41436-021-01171-4

Performance

Method Description

Next-generation sequencing (NGS) data previously generated on the proband is reprocessed to assess for the presence of variants in the coding regions and intron/exon boundaries of approximately 20,000 genes. If comparator samples are submitted, NGS is performed on DNA extracted from the submitted comparator samples and this data is used in conjunction with the proband's previously generated data. The human genome reference GRCh37/hg19 build is used for sequence read alignment. Variants are called using an optimized bioinformatics package. At least 99% of the bases are covered at a read depth over 30X. Sensitivity is estimated at above 99% for single nucleotide variants, above 94% for deletion-insertions (delins) less than 40 base pairs (bp), and above 95% for deletions up to 75 bp and insertions up to 47 bp. This assay also detects most copy number variants (deletions/duplications) involving three or more exons. In some instances, copy number variants less than three exons may be detected, however, the reliability of this detection is variable due to isolated reduction in sequence coverage or inherent genomic complexity. Resulting variants are filtered and annotated using public and proprietary resources and presented for analysis and interpretation using a vended interpretation tool. Confirmation of select reportable variants in the proband and submitted comparator samples may be performed by alternate methodologies based on internal laboratory criteria.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis as a result of technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences.(Unpublished Mayo method)

PDF Report

Supplemental

Day(s) Performed

Varies

Report Available
56 to 70 days

Specimen Retention Time
Whole blood: 28 days (if available); Saliva: 30 days (if available); Extracted DNA: 3 months; Blood spots: 1 year (if available)

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
81417-Patient only
81417, 81416-Patient and one family member comparator sample (duo) (as appropriate)
81417, 81416 x 2-Patient and two family member comparator samples (trio or non-traditional trio) (as appropriate)
81417, 81416 x 3-Patient and three family member comparator samples (quad) (as appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
WESPR	Whole Exome Sequencing Panel Reflex	86205-2

Result ID	Test Result Name	Result LOINC® Value
621327	Interpretation	69047-9
621328	Specimen	31208-2
621329	Source	31208-2
621330	Released By	18771-6