MAYO CLINIC LABORATORIES

Test Definition: NGSFX

Reanalysis of Acute Myeloid Leukemia 4- or 11- Gene Panels, Additional Genes

Overview

Useful For

Comprehensive reanalysis of a larger set of genes/gene regions when a more targeted gene panel was previously performed in this laboratory

Evaluation of known or suspected hematologic neoplasms, specifically of myeloid origin (eg, acute myeloid leukemia, myelodysplastic syndrome, myeloproliferative neoplasm, myelodysplastic/myeloproliferative neoplasm, unexplained cytopenias) at the time of diagnosis or possibly disease relapse, to help determine diagnostic classification and provide prognostic or therapeutic information for helping guide clinical management

Determine the presence of new clinically important gene mutation changes at relapse

Genetics Test Information

This test includes next-generation sequencing to evaluate the following 47 genes and select intronic regions: *ANKRD26*, *ASXL1, BCOR, BCORL1, BRAF, CALR, CBL, CEBPA, CSF3R, DDX41, DNMT3A, ELANE, ETNK1, ETV6, EZH2, FLT3, GATA1, GATA2, IDH1, IDH2, JAK2, KDM6A, KIT, KRAS, MPL, NF1, NPM1, NRAS, PHF6, PPM1D, PTPN11, RAD21, RUNX1, SETBP1, SH2B3, SF3B1, SMC3, SRSF2, STAG2, STAT3, TERT, TET2, TP53, U2AF1, UBA1, WT1,* and *ZRSR2.*

Testing Algorithm

Only orderable as a reflex. Reflex testing is available upon request within 6 months of original NGAMT / MayoComplete Acute Myeloid Leukemia, Therapeutic Gene Mutation Panel (*FLT3*, *IDH1*, *IDH2*, *TP53*), Next Generation Sequencing, Varies or NGAML / MayoComplete Acute Myeloid Leukemia, 11-Gene Panel, Varies sample submission.

This is a bioinformatics and variant review only for the added gene regions.

For a list of genes and exons targeted by this test see Targeted Genes Interrogated by Myeloid Neoplasms, Comprehensive OncoHeme Next-Generation Sequencing.

Special Instructions

- Hematopathology Patient Information
- Targeted Genes Interrogated by Myeloid Neoplasms, Comprehensive OncoHeme Next-Generation Sequencing

Method Name

Only orderable as a reflex. For more information see:
-NGAMT / MayoComplete Acute Myeloid Leukemia, Therapeutic Gene Mutation Panel (*FLT3*, *IDH1*, *IDH2*, *TP53*) Next-Generation Sequencing, Varies
-NGAML / MayoComplete Acute Myeloid Leukemia, 11-Gene Panel, Varies

Next-Generation Sequencing (NGS)

NY State Available

Yes

Specimen

Specimen Type
Varies

Specimen Required
No additional specimen is required. This is a bioinformatics review of additional gene regions not analyzed in the previously ordered NGAMT / MayoComplete Acute Myeloid Leukemia, Therapeutic Gene Mutation Panel (*FLT3*, *IDH1*, *IDH2*, *TP53*), Next-Generation Sequencing, Varies or NGAML / MayoComplete Acute Myeloid Leukemia, 11-Gene Panel, Varies. Call 800-533-1710 for assistance with ordering.

Forms
1. Hematopathology Patient Information (T676)
2. If not ordering electronically, complete, print, and send an Hematopathology/Cytogenetics Test Request (T726) with the specimen.

Reject Due To

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies	14 days	

Clinical & Interpretive

Clinical Information
Next-generation sequencing is a comprehensive molecular diagnostic methodology that can interrogate multiple regions of genomic tumor DNA in a single assay. Many hematologic neoplasms are characterized by morphologic or phenotypic similarities but can have characteristic somatic mutations in many genes that enable more specific categorization. In addition, many myeloid neoplasms lack a clonal cytogenetic finding at diagnosis (normal karyotype) but can be diagnosed or confirmed and classified according to the gene mutation profile. Patients with unexplained cytopenias may harbor acquired genetic alterations in hematopoietic cells (clonal cytopenias of uncertain significance), which may carry the risk of developing overt myeloid malignancies. The presence and pattern of gene mutations in known or suspected myeloid neoplasm can provide critical diagnostic, prognostic, and therapeutic information to help guide management for the patient's physician.

Reference Values
Only orderable as a reflex. For more information see:

-NGAMT / MayoComplete Acute Myeloid Leukemia, Therapeutic Gene Mutation Panel (*FLT3*, *IDH1*, *IDH2*, *TP53*), Next-Generation Sequencing, Varies
-NGAML / MayoComplete Acute Myeloid Leukemia, 11-Gene Panel, Varies

Interpretation

Detailed variant assessment and interpretive comments will be provided for all reportable genetic alterations.

If this test is ordered in the setting of erythrocytosis and suspicion of polycythemia vera, interpretation requires correlation with a concurrent or recent prior bone marrow evaluation.

Cautions

This test is a targeted next-generation sequencing (NGS) assay that encompasses 47 genes with variable full exon, partial region (including select intronic or non-coding regions), or hot spot coverage (depending on specific locus). Therefore, this test will not detect other genetic abnormalities in genes or regions outside the specified target areas. The test detects single base substitutions (ie, point mutations) as well as small insertion or deletion type events, but it does not detect gene rearrangements (ie, translocations), gene fusions, copy number alterations, or large scale (segmental chromosome region) deletions and complex changes.

This assay does not distinguish between somatic and germline alterations in analyzed gene regions, particularly with variant allele frequencies near 50% or 100%. If nucleotide alterations in genes associated with germline variant syndromes are present and there is a strong clinical suspicion or family history of malignant disease predisposition, additional genetic testing and appropriate counseling may be indicated. A low incidence of gene mutations associated with myeloid neoplasms can be detected in nonmalignant hematopoietic cells in individuals with advancing age (clonal hematopoiesis of indeterminate potential), and these may not be clearly distinguishable from tumor-associated mutations. Some apparent mutations classified as variants of uncertain significance may represent rare or low-frequency polymorphisms.

Prior treatment for hematologic malignancy could affect the results obtained in this assay. In particular, a prior allogeneic hematopoietic stem cell transplant may cause difficulties in resolving somatic or polymorphic alterations or assigning variant calls correctly to donor and recipient fractions if pertinent clinical or laboratory information (eg, chimerism engraftment status) is not provided.

The finding of a genetic alteration does not necessarily indicate the presence of a myeloid neoplasm. Correlation with clinical, histopathologic, and additional laboratory findings is required for final interpretation of NGS results and is the responsibility of the managing physician.

Clinical Reference

1. Pollyea DA, Altman JK, Assi R, et al. Acute Myeloid Leukemia, Version 3.2023, NCCN Clinical Practice Guidelines in Oncology. J Natl Compr Canc Netw. 2023;21(5):503-513. doi:10.6004/jnccn.2023.0025
2. Gerds AT, Gotlib J, Ali H, et al. Myeloproliferative Neoplasms, Version 3.2022, NCCN Clinical Practice Guidelines in Oncology. J Natl Compr Canc Netw. 2022;20(9):1033-1062. doi:10.6004/jnccn.2022.0046
3. Greenberg PL, Stone RM, Abaza Y, et al. NCCN Guidelines Insights: Myelodysplastic Syndromes, Version 2.2025. J Natl Compr Canc Netw. 2025;23(3):66-75. doi:10.6004/jnccn.2025.0013
4. He R, Chiou J, Chiou A, et al. Molecular markers demonstrate diagnostic and prognostic value in the evaluation of myelodysplastic syndromes in cytopenia patients. Blood Cancer J. 2022;12(1):12. doi:10.1038/s41408-022-00612-w

5. Malcovati L, Galli A, Travaglino E, et al. Clinical significance of somatic mutation in unexplained blood cytopenia. Blood. 2017;129(25):3371-3378. doi:10.1182/blood-2017-01-763425
6. DiNardo CD, Stein EM, de Botton S, et al. Durable remissions with ivosidenib in IDH1-mutated relapsed or refractory AML. N Engl J Med. 2018;378(25):2386-2398. doi:10.1056/NEJMoa1716984
7. Stein EM, DiNardo CD, Fathi AT, et al. Molecular remission and response patterns in patients with mutant-IDH2 acute myeloid leukemia treated with enasidenib. Blood. 2019;133(7):676-687. doi:10.1182/blood-2018-08-869008
8. Dohner H, Estey E, Grimwade D, et al. Diagnosis and management of AML in adults: 2017 ELN recommendations from an international expert panel. Blood. 2017;129(4):424-447. doi:10.1182/blood-2016-08-733196
9. Smith CC. The growing landscape of FLT3 inhibition in AML. Hematology Am Soc Hematol Educ Program. 2019;2019(1):539-547. doi:10.1182/hematology.2019000058
10. Kennedy JA, Ebert BL. Clinical implications of genetic mutations in myelodysplastic syndrome. J Clin Oncol. 2017;35(9):968-974. doi:10.1200/JCO.2016.71.0806
11. Daver N, Schlenk RF, Russell NH, Levis MJ. Targeting FLT3 mutations in AML: review of current knowledge and evidence. Leukemia. 2019;33(2):299-312. doi:10.1038/s41375-018-0357-9
12. Swerdlow SH, Campo E, Harris NL, et al. WHO classification of tumours of hematopoietic and lymphoid tissues. IARC Press; 2017

Performance

Method Description
This analysis requires either NGAMT / MayoComplete Acute Myeloid Leukemia, Therapeutic Gene Mutation Panel (*FLT3*, *IDH1*, *IDH2*, *TP53)*, Next-Generation Sequencing, Varies or NGAML / MayoComplete Acute Myeloid Leukemia, 11-Gene Panel, Varies to have been previously performed at Mayo Clinic Laboratories within the last 6 months. An extended bioinformatics analysis is performed on the original data by a bioinformatics pipeline, and a variant call file is generated for final analysis and reporting of any additional disease-causing variants within genomic target regions present in the larger NGSHM / MayoComplete Myeloid Neoplasms, Comprehensive OncoHeme Next-Generation Sequencing, Varies.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
16 to 21 days

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

81450

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
NGSFX	Reanalysis, AML 4 or 11 Gene Panel	99961-5

Result ID	Test Result Name	Result LOINC® Value
MP043	Specimen Type	31208-2
NFXID	Diagnosis/Indication	29308-4
601695	NGSFX Result	No LOINC Needed
601687	Pathogenic Mutations Detected	82939-0
601686	Interpretation	69047-9
601688	Clinical Trials	82786-5
601689	Variants of Unknown Significance	93367-1
601690	Additional Notes	48767-8
601691	Method Summary	85069-3
601693	NGSFX Panel Gene List	36908-2
601694	Reviewed By:	18771-6
601692	Disclaimer	62364-5