

Sample ID:	2870
Sample Type:	Blood
Patient's Name:	218794 b
Gender/Age:	Female / NA



RNA FUSION PANEL

Data Uploaded on:	15/08/2025 5:12PM	Report Generated on:	15/08/2025 8:35PM
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Cancer Type:- Myleoid

Results:-

CLINICALLY RELEVANT FUSIONS

Fusion	Breakpoint 1	Breakpoint 2
RUNX1-RUNX1T1	RUNX1- 21:34859474	RUNX1T1- 8:92017363

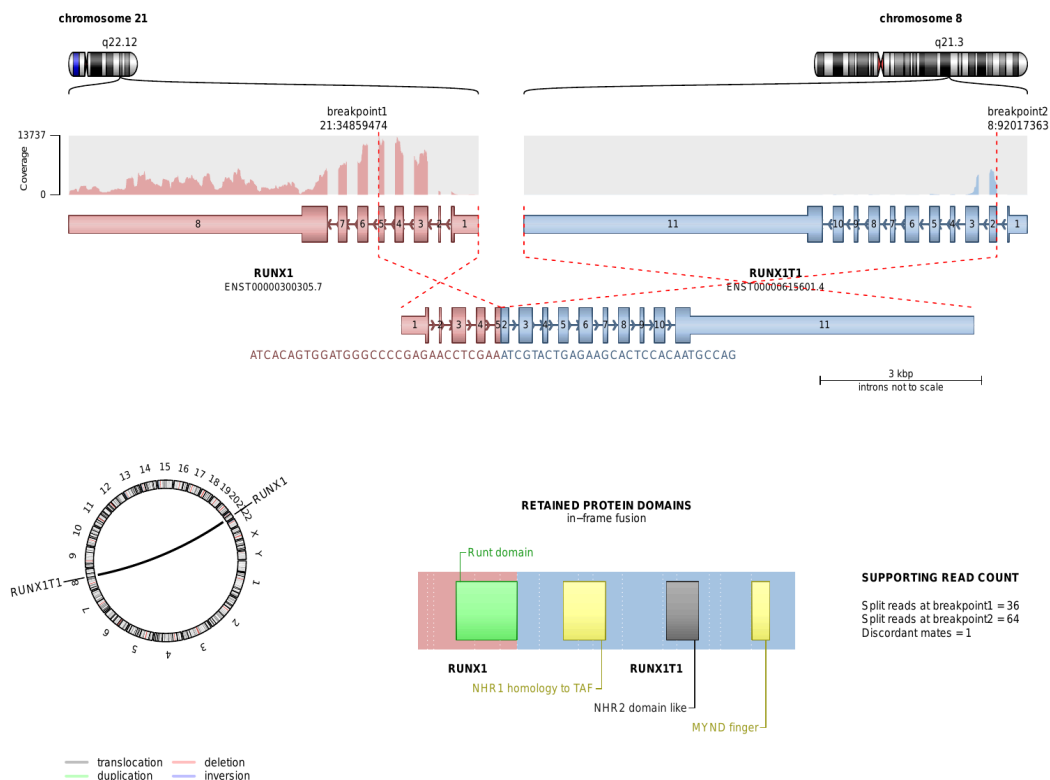


Fig :- Identification and Visualization of Gene Fusions via RNA-Targeted Sequencing

RECOMMENDATIONS

- Genetic counseling is advised for interpretation on the consequences of the results.
- If results obtained do not match the clinical findings, additional testing should be considered as per referring clinician's recommendation.
- Genetic test results are reported based on the recommendations of Association for Molecular Pathology, American Society of Clinical Oncology, and College of American Pathologists
- Data revaluation performed upon the up gradation of databases used and results may vary in accordance.

METHODOLOGY

The Genes2Me RNA Fusion NGS panel was used for sequencing that screens clinically relevant fusions in blood cancer with a hybridization-based target capture technique.

Sample type: Peripheral blood or bone marrow collected in EDTA tube

Extraction and Library Preparation:

RNA was isolated using standard extraction protocols. For the RNA Pre- capture library preparation 1st Strand Synthesis, 2nd Strand synthesis, End Repair A tailing, Ligation of the Adaptors, and attachment of unique barcodes followed by cleanups are performed. Then these precapture libraries are pooled as per the required ng input and kept for targeted gene capture.

Data Analysis:

Sequencing data were processed using G2M's internally developed and analytically validated bioinformatics pipeline optimized for gene fusion detection. Only gene fusions supported by ≥ 1 high-confidence split reads were considered for reporting, as established by internal validation metrics.

Disclaimer:

This test was developed and its performance characteristics were determined by G2M. The assay is validated for clinical use under applicable laboratory regulations.

LIMITATIONS

- Genetic testing is an important part of the diagnostic process however it may not always give a definitive answer. Accurate interpretation of test results is dependent on the availability of biological & medical information (clinical history) of the family, failing to this may leads to incorrect result interpretation and diagnosis.
- Test results are interpreted in the context of clinical findings, available scientific evidences, family history and other laboratory data.
- Genetic testing is highly accurate but rarely inaccurate results may occur for various reasons like mislabelling of samples, inaccurate clinical/medical family history, tumour purity, rare technical errors or unusual circumstances such as bone marrow transplantation,

DISCLAIMER

- The interpretation result in this report is performed to the best knowledge of the scientific & medical information available at the time of reporting. The classification of variants is based on AMP-ASCO-CAP guidelines.
- As of the inherent technological limitations of the sequencing assay, some of the coding regions can't be properly sequenced, so, variations in these regions may not be identified & interpreted. It is possible that variants not identified by the assay may be associated with the provided phenotypes of the patient.
- Genes2Me clarify that the generated report(s) doesn't provide any kind of diagnosis or opinion or recommendation for any disease and its cure in any manner. It is therefore recommended that the patient and/or the guardian(s) of the patient must take the consultation of the clinician or a certified physician or doctor for further course of action.
- If the provided material quality and/or quantity not up to the desired level, further procedures will be completed only after getting confirmation from referring clinician/physician only, so, in that case, test(s) result(s) may be misleading or even wrong, therefore, Genes2Me hereby disclaims all liability arising in this connection with the test(s) and report(s).
- The analysis pipeline is developed in-house and the performance characteristics of this analysis are determined by Genes2Me only.
- This test result should be used as a reference by the healthcare provider for diagnosis and development of treatment plan.
- The clinically significant mutations enlisted in this report are provided as a professional service, and are not reviewed or approved by the FDA.

REFERENCES

- Hoffmeister, L.M., Suttrop, J., Walter, C. et al. Panel-based RNA fusion sequencing improves diagnostics of pediatric acute myeloid leukemia. *Leukemia* 38, 538–544 (2024). <https://doi.org/10.1038/s41375-023-02102-9>
- Barua S, Wang G, Mansukhani M, Hsiao S, Fernandes H. Key considerations for comprehensive validation of an RNA fusion NGS panel. *Pract Lab Med*. 2020 Jun 8;21:e00173. doi: 10.1016/j.plabm.2020.e00173. PMID: 32613069; PMCID: PMC7322345.
- Heyer, E.E., Deveson, I.W., Wooi, D. et al. Diagnosis of fusion genes using targeted RNA sequencing. *Nat Commun* 10, 1388 (2019)
- Kumar, S., Vo, A., Qin, F. et al. Comparative assessment of methods for the fusion transcripts detection from RNA-Seq data. *Sci Rep* 6, 21597 (2016).
- Davila, J.I., Fadra, N.M., Wang, X. et al. Impact of RNA degradation on fusion detection by RNA-seq. *BMC Genomics* 17, 814 (2016).

APPENDIX

FDA-approved drugs targeting gene fusion proteins

Fusion Genes	Cancer Type	Drugs	Classification
BCR-ABL1	B-Lymphoblastic Leukemia/Lymphoma	Dasatinib, Imatinib, Ponatinib	Tier 1
BCR-ABL1	Chronic Myelogenous Leukemia	Asciminib, Bosutinib, Dasatinib, Imatinib, Nilotinib	Tier 1
BCR-ABL1	B-Lymphoblastic Leukemia/Lymphoma	Bosutinib, Nilotinib	Tier 1
ALK	Anaplastic Large-Cell Lymphoma	Crizotinib	Tier 1
ALK	Non-Small Cell Lung Cancer	Alectinib, Brigatinib, Ceritinib, Ensartinib, Lorlatinib	Tier 1
BRAF	Low-Grade Glioma	Tovorafenib	Tier 2
FGFR1	Myeloid/Lymphoid	Pemigatinib	Tier 1
FGFR2	Cholangiocarcinoma	Pemigatinib, Futibatinib	
NRG1	Non-Small Cell Lung	Zenocutuzumab	
NRG1	Pancreatic Adenocarcinoma	Zenocutuzumab	
NTRK1	All Solid Tumors	Entrectinib, Larotrectinib, Repotrectinib	Tier 1
NTRK2	All Solid Tumors	Entrectinib, Larotrectinib, Repotrectinib, Selpercatinib	
RET	All Solid Tumors	Selpercatinib	Tier 1
RET	Non-Small Cell Lung Cancer	Pralsetinib, Selpercatinib	Tier 1
RET	Thyroid Cancer	Pralsetinib, Selpercatinib	Tier 1
ROS1	Non-Small Cell Lung Cancer	Crizotinib, Entrectinib, Repotrectinib	Tier 2
BCR-ABL1	B-Lymphoblastic Leukemia/Lymphoma	Bosutinib, Nilotinib	Tier 1
ALK	Inflammatory Myofibroblastic Tumor	Alectinib, Brigatinib, Ceritinib, Lorlatinib	Tier 1
JAK2	Myeloid/Lymphoid	Fedratinib, Ruxolitinib	Tier 1
RET	Non-Small Cell Lung Cancer	Cabozantinib	Tier 1
ROS1	Non-Small Cell Lung Cancer	Ceritinib, Lorlatinib	Tier 2
BRAF	Melanoma	Cobimetinib, Trametinib	Tier 2
BRAF	Ovarian Cancer	Cobimetinib, Trametinib	Tier 2
NRG1	All Solid Tumors	Zenocutuzumab	
NRG1	Non-Small Cell Lung Cancer	Seribantumab, Vandetanib, Taletrectinib	
MET	All Solid Tumors	Crizotinib	
NTRK1	All Solid Tumors	Zurietrectinib	Tier 1
NTRK2	All Solid Tumors	Zurietrectinib	
NTRK3	All Solid Tumors	Zurietrectinib	

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***For any further technical queries please contact at contact@genes2me.com**