Patient Information

Patient Name:	Yanxin Lu
Date of Birth:	10/17/1989
Gender:	Male
Ethnicity:	Not Provided
Patient ID:	300751462
Medical Record #:	202192870
Collection Kit:	40886880-2-C
Reference ID:	43607050-2-C
Accession ID:	676807716
Case File ID:	15637925

Test Information

Ordering Physician:	Erica T Wang, MD
Clinic Information:	Cedars Sinai-Fertility & Reproductive Medicine Center
Phone:	310-423-9964
Report Date:	03/14/2025
Sample Collected:	03/10/2025
Sample Received:	03/11/2025
Sample Type:	Blood

horizon™ natera carrier screen

CARRIER SCREENING REPORT

ABOUT THIS SCREEN: Horizon™ is a carrier screen for specific autosomal recessive and X-linked diseases. This information can help patients learn their risk of having a child with specific genetic conditions.

ORDER SELECTED: **Tay-Sachs Enzyme** was ordered for this patient.

FINAL RESULTS SUMMARY:

NEGATIVE FOR Tay-Sachs Disease (enzyme only)

Please see following page for Hexosaminidase A values.

RECOMMENDATIONS

Individuals who would like to review their Horizon report with a Natera Laboratory Genetic Counselor may schedule a telephone genetic information session by calling 650-249-9090 or visiting naterasession.com. Clinicians with questions may contact Natera at 650-249-9090, 855-866-6478 (toll free) or email support@natera.com.

Reviewed by: Yang Wang, Ph.D., FACMGG, Laboratory Director
CLIA Laboratory Director: J. Dianne Keen-Kim, Ph.D., FACMG

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DISEASES SCREENED

Below is a list of all diseases screened and the result. Certain conditions have unique patient-specific numerical values, therefore, results for those conditions are formatted differently.

Autosomal Recessive

T
Tay-Sachs Disease (enzyme only)
Negative: Normal Hexosaminidase Activity. WBC: 3789.00 nmol/hr/mg; Hex A %. WBC 69.30.

Testing Methodology, Limitations, and Comments:

Tay-Sachs Disease Enzyme Analysis

A small percentage (<0.7 %) of Tay-Sachs disease carriers may be identified as non-carriers by this assay (Triggs-Raine et al. NEJM 1990). In addition, Tay-Sachs disease patients or carriers with certain genetic variants such as AB variant (OMIM 272750) and B1 variant (OMIM 272800) will not be detected by this method. Methodology: This enzyme assay determines total hexosaminidase and hexosaminidase A activities in leukocytes. The hexosaminidase activities are measured before and after heat inactivation using a fluorescence-generating 4-methylumbelliferyl-N-acetyl-β-D-glucosaminide substrate. Thermal fractionation of hexosaminidase is calculated to differentiate Tay-Sachs disease carriers from non-carriers. This test was performed by Baylor Miraca Genetics Laboratories, DBA Baylor Genetics, 2450 Holcombe Blvd, Ste O104, Houston, TX 77021 (CLIA ID 45D0660090). The performance characteristics of this test were developed by Baylor Miraca Genetics, DBA Baylor Genetics. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research.

Tay-Sachs Disease (Hex A % Carrier Ranges)

Specimen	Carrier Range (%)	Non-Carrier Range (%)
White Blood Cells (WBC)	< 49	55.0-75.0

Negative Results

A negative carrier screening result reduces the risk for a patient to be a carrier of a specific disease but does not completely rule out carrier status. Any patient with a family history for a specific genetic disease will have a higher carrier risk prior to testing and if the disease-causing variant in their family is not included on the test, their carrier risk remains unchanged. Genetic counseling is recommended for patients with a family history of genetic disease so that risk figures based on actual family history can be determined and discussed along with potential implications for reproduction.

Additional Comments

Horizon carrier screening (3.2.1) has been developed to identify the reproductive risks for monogenic inherited conditions. Even when one or both members of a couple screen negative for pathogenic variants in a specific gene, the disease risk for their offspring is not zero. There is still a low risk for the condition in their offspring due to a number of different mechanisms that are not detected by Horizon, including but not limited to, pathogenic variant(s) in the tested gene or in a different gene not included on Horizon, pathogenic variant(s) in an upstream regulator, uniparental disomy, de novo mutation(s), or digenic or polygenic inheritance. Infrequent large genetic deletions or duplications are not detected unless they have been specifically targeted for carrier testing.

This test was developed and its performance characteristics determined by Baylor Miraca Genetics Laboratories DBA Baylor Genetics (CAP# 2109314/ CLIA# 45D0660090). Data review and reporting were performed by Natera and NSTX, 13011 McCallen Pass, Building A, Suite 110, Austin, TX 78753 (CLIA ID: 45D2093704). These tests have not been cleared or approved by the U.S. Food and Drug Administration (FDA). These analyses generally provide highly accurate information regarding the patient's carrier status; however, there are many potential sources of diagnostic error, including misidentification of samples, polymorphisms, or other rare genetic variants that interfere with analysis. Families should understand that rare diagnostic errors may occur for these reasons.

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DETAILED RESULTS AND INTERPRETATIONS

TAY-SACHS ENZYME

Tay Sachs Disease Carrier Testing

Sample	%HexosaminidaseA	Total Activity nmoles/hr/mg protein
Lu,Yanxin	69.3	3789
Normal Range	55.0-75.0	1023-1961
Carrier Range	34.0-49.0	870-1705

INTERPRETATION: Non-Carrier: Within the limits of this test this patient is NOT a carrier for Tay Sachs Disease.

Results Date: 03/14/2025