Expanded Carrier Screen
Pathology Request Form

genomic diagnostics
A Healius company

AUTHORIZED DISTRIBUTOR
natera | Horizon™ Advanced carrier screening

Lab ID

Patient Information

Surname: .....

First Name: .....

Address: .....

..... Postcode: .....

Email: .....

Mobile: .....

DOB: D D M M Y Y Y Y Age: ..... M ☐ F ☐

Patient Ethnicity - Tick One Box Only

- ☐ African American/Black
- ☐ Hispanic/Latin American
- ☐ Mediterranean
- ☐ Caucasian/Non-Hispanic White
- ☐ Ashkenazi Jewish
- ☐ Sephardic Jewish
- ☐ French Canadian
- ☐ East Asian
- ☐ Southeast Asian
- ☐ South Asian*
- ☐ Other*.....

*When these ethnicities are checked, the residual risk for negative results on the report will reflect a pan-ethnic residual risk number.

Requesting Clinician

Name: .....

Address: .....

..... Postcode: .....

Phone: .....

Email: .....

Provider No. .....

Signature: .....

Report Copy

Name: .....

Address: .....

..... Postcode: .....

Phone: .....

Email: .....

Reason for Test

- ☐ Supervision of other normal pregnancy, 1st trimester
- ☐ Supervision of other normal pregnancy, 2nd trimester
- ☐ Female: genetic disease carrier status
- ☐ Male: genetic disease carrier status
- ☐ Supervision of normal 1st pregnancy, 1st trimester
- ☐ Supervision of normal 1st pregnancy, 2nd trimester
- ☐ Encounter for genetic counselling
- ☐ Family history of intellectual disabilities
- ☐ Family history of other diseases of the musculoskeletal system and connective tissue
- ☐ Family history of carrier of genetic disease
- ☐ Family history of other specified conditions

Comments Please describe any relevant family history or prior testing

Clinical Details

Is the patient above currently pregnant? ☐ Yes ☐ No

Gestational age: Weeks: ..... Days: .....

If patient is pregnant, consider simultaneous tandem testing of partner for fastest return of results.

Couple Reporting:

Do you wish to merge this patient with a partner?

☐ Yes ☐ No Check if a couple test ☐

Partner Information:

First Name: .....

Last Name: .....

DOB: D D M M Y Y Y Y

I would like to share my Horizon™ test results with my partner and his/her health care provider for treatment purposes. I have obtained necessary authorisation to provide my partner's information above.

Patient Signature: .....

Date: .....

☐ Do not send reports to My Health Record

DATA ENTRY INSTRUCTIONS: BILL CODE: COFS **PANEL CODE:** CFC/CFO (QML)

Collection Information

PLEASE CONFIRM PATIENT HAS PAID FOR THIS TEST BY CHECKING THE RECEIPT BOX over page.

Sample Type: Whole blood required - 1 x 6mL pink top EDTA tube.

PERSON COLLECTING SPECIMEN TO COMPLETE:

I certify I established the identity of the patient named on this request, collected and immediately labelled the accompanying specimen with the patient's details.

Initials ..... ACC Code / Location: ..... Date of draw: D D M M Y Y Y Y Time: ☐☐:☐☐ am / pm

Informed Consent Natera Horizon™ Expanded Carrier Screen

Please review this information carefully and then indicate with your signature if you wish to move forward with testing. This is a voluntary test. You may wish to undertake a genetic information session prior to signing this form.

Purpose

- The Horizon™ Carrier Screen is designed to determine whether you carry genetic changes, called pathogenic variants, that could cause serious genetic conditions in your children.
- For most of the conditions on the panel, both parents must carry a pathogenic variant in the same gene for their children to be at risk of developing symptoms. However, there are certain conditions on the Horizon™ Carrier Screen for which only the mother needs to carry a pathogenic variant for her children to be at risk of developing symptoms.

Benefits

- Your Horizon™ Carrier Screen results can help you and your partner make more informed decisions regarding your family, particularly if screening is performed prior to pregnancy.
- If it is early in your pregnancy, you can pursue further testing to determine if the pregnancy is affected, and receive guidance from your healthcare provider about how best to plan and prepare for birth.
- Your Horizon™ Carrier Screen results may also benefit your other family members. If you test positive, your biological relatives are more likely to test positive for the same pathogenic variant(s), thereby allowing them to discover previously unknown conditions and risks.

What you might learn

- Carrier (Positive): A positive result means that a change(s) has been identified in a specific gene indicating that a person is at least a carrier for the screened condition. Natera's policy is to report pathogenic and likely pathogenic variants. Likely pathogenic variants are those that based on review of the available literature and American College of Medical Genetics and Genomics guidelines are likely—but not certain—to have deleterious effects. Natera's Horizon Carrier Screen does not detect all possible disease-causing pathogenic variants in a given gene. Carriers usually do not experience symptoms of the condition. For some conditions, carriers may require changes to medical management.
- No pathogenic variants detected (Negative): A negative carrier screening result reduces but does not eliminate, the risk to be a carrier for the genetic conditions screened. Carrier frequencies and detection rates vary based on ethnicity. The residual carrier risk for each specific disease given negative screening results can be accessed through the link on the Horizon report or via the link at https://www.natera.com/panel-option/h-all/

Procedure

- The Horizon™ Carrier Screen can be done before pregnancy or early in pregnancy, as ordered by your healthcare provider.
- A small blood sample is taken and sent to Natera™ for screening.

Risks

- Genetic testing may reveal sensitive information about your health and that of your family members.
- This test may provide information that can have an impact on your medical decisions.

Limitations

- The Horizon™ Carrier Screen is not intended to detect all genetic pathogenic variants.
- As with all medical screening tests, there is a chance of error, including a false positive or false negative result.
- A "false positive" refers to identifying a pathogenic variant that is not present.
- A "false negative" is the failure to detect a pathogenic variant that is present in the sample.
- Certain factors, such as having blood cancer, prior blood transfusions, or previous bone marrow transplants can affect the accuracy of Horizon™ Carrier Screen results. Be sure to discuss your medical history with your healthcare provider.
- Occasionally it may not be possible to provide a result. A repeat specimen may be requested.

Privacy

- If you and your partner are receiving simultaneous Horizon™ Carrier Screen testing, each of your test results may be revealed to one another and to each other's ordering providers.
- Your Horizon™ Carrier Screen results will be reported to your healthcare provider or his/her agent.
- By agreeing to testing and signing this consent, you hereby authorise Genomic Diagnostics to share your Horizon™ Carrier Screen results with other authorised representatives that you've identified to us or your healthcare provider, or as otherwise allowed by law.
- Natera™ may find information that is not included in the original test requested by your healthcare provider and may report these additional results, if clinically relevant. You authorise Genomic Diagnostics to share these results with you and your healthcare provider.
- Please refer to Natera's™ Notice of Privacy Policy, available on the Natera™ website, for additional information about Natera's™ privacy practices, including how your protected health information (including your samples and genetic information) may be shared with third-party vendors and service providers that they partner with to provide testing services to you.
- Please refer to Genomic Diagnostics Privacy Policy at genomicdiagnostics.com.au.

Sample Processing Authorisation

My sample and related data will be sent outside of Australia for performance of the ordered test(s) by Natera and/or its contractor(s), and there will be no receipt of any payments, benefits, or rights to any resulting products or discoveries for myself or my heirs.

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I have read or have had read to me and understand all of the above information and have had an opportunity to ask questions about the purpose, procedure, risks, benefits and limitations of testing.

I HAVE DECIDED TO PURSUE TESTING and to be bound by the terms of this Consent and any policies referenced herein.

Patient Name	Date of Birth	Patient Signature	Date

Ordering Healthcare Provider Name	Ordering Healthcare Provider Signature	Date

PAYMENT INFORMATION

Patient's Name: .....

PATIENT AUTHORISATION: I understand this test requires prepayment of $699 per person and $1350 per couple tested before my blood is collected* ☐

RECEIPT NUMBER FOR COUPLE TESTING MUST BE PROVIDED ON BOTH REQUEST FORMS.

Please visit genomicdiagnostics.com.au to prepay or call 1800 822 999.

RECEIPT NO.

QR PAY

INDIVIDUAL TEST

COUPLE TEST

* Pricing is valid at June 2024 and subject to change without notice.

For more information, contact us at info@genomicdiagnostics.com.au

1800 822 999 **genomicdiagnostics.com.au**