Consent form - *Genomic Health Screen*

You have expressed interest in pursuing Alamya Health's Genomic Health Screen testing. Results of this testing can be used to inform your health management strategies and to guide preventative health surveillance. The information on this form briefly summarizes information about genomic testing. If you have any questions or concerns about this testing, please contact the Alamya Health team or your healthcare provider to discuss further.
As well, if you decide to proceed with genomic testing, we recommend that you review all of the findings in this report with your healthcare professional.

Purpose

Alamya Health offers cutting edge, comprehensive whole genome sequencing, called the Genomic Health Screen. This test includes two complementary approaches for sequencing the whole genome: short-read sequencing and long-read sequencing. Briefly, whole genome sequencing looks for changes or variations throughout one's DNA. Such DNA variations can cause a gene or a group of genes to function improperly. Alamya Health's extensive testing approach and thorough data analyses can provide valuable information about one's health now and for the future along with possible management options. For more information on specific testing and analytic components, see www.alamyahealth.com

How it is performed

Genomic testing is performed on DNA that is typically obtained from a small blood sample. Analysis and reporting of the testing data may be guided by detailed clinical information and family history information along with up to date published medical and scientific information. If, during evaluation, you report clinical findings suggestive of a genetic condition, additional diagnostic testing might be offered in conjunction with the Genomic Health Screen. In some cases, samples from other family members might be recommended to help interpret a finding. In such situations, the choice of whether or not to pursue further testing is up to you.

Alamya Health will provide you with a test report, and we encourage you to share this with your healthcare provider. If the Genomic Health Screen is ordered by your healthcare provider, the test report will also be forwarded to that individual directly. Your healthcare provider can help you understand and contextualize your test results and manage follow up investigations, consultations, treatments. The team at Alamya Health are available to discuss test findings and answer some of your questions but we do not manage any follow up investigations or health concerns.

Types of results

Positive genetic result: A genetic change was found that could be associated with or pose a risk for a specific health concern(s). Such genetic changes are termed pathogenic variants or likely pathogenic variants. Genomic Health Screen focuses on the following three categories:

- The American College of Medical Genetics (ACMG) has recommended the reporting of genetic variants in 81 select genes associated with various genetic disorders that are considered to be medically actionable in childhood or in adulthood. There are recommended guidelines for monitoring and treating each of these conditions (https://www.acmg.net/PDFLibrary/Secondary-Findings-v3.1.pdf).
- Pharmacogenetic findings that can guide prescription approaches for certain common medications
- Determining carrier status for approximately 1,300 genetic disorders that could pose risks for offspring if both parents are carriers

Positive transcriptome result: Sequencing of RNA can provide support for the pathogenicity of a genetic variant.

Positive pharmacogenomic result: Specific changes can indicate the potential for an altered response to certain medications. As with any positive test finding, this should be discussed with your healthcare provider(s) especially as your response to a medication can be influenced by multiple factors.

Negative: No pathogenic or likely pathogenic variants were found in the select genes queried (as noted above). Negative findings might be due to current limitations in scientific and medical knowledge and/or testing technology. Such results do not, however, completely rule out the possibility of an underlying genetic disease. Reanalysis or retesting in future might be indicated and you should discuss this with your healthcare provider (see below section on "Sample retention, data storage, and database participation").

OPTIONAL TEST RESULTS: - please initial your choice of whether or not to include these options

1. Repeat expansion disorders: a number of genes contain repeated sequences of DNA. The number of repeats can vary among individuals and across generations but they do not typically vary within an individual; that is, repeat sequence sizes remain relatively stable across one's lifespan. If one of these repeat sequences has expanded more than it should, it can lead to a specific disorder in that individual. These disorders often involve neurological impairment, and this impairment can be progressive. For a number of these disorders, for example Huntington disease, there may not be an effective treatment. The decision of whether or not to include repeat expansion test results in your report is entirely up to you.

I **do** want the results of repeat expansion testing in my report _____
I **do not** want the results of repeat expansion testing in my report _____

2. Polygenic risk scores: certain traits and relatively common conditions (e.g. cardiovascular disease, type 2 diabetes, some cancers, autoimmune disorders, etc.) can each be caused by variants in many genes along with environmental factors. Genetic testing can be used to generate polygenic risk scores (i.e. probabilities) to develop such conditions. This information can be used to make lifestyle decisions and in some cases, early screening and treatment. Knowledge about polygenic risk scores and their clinical usefulness continues to evolve and this can lead to modification of your scores with time, including adding new scores. The decision of whether or not to include polygenic risk scores in your report is entirely up to you.

   I **do** want polygenic risk score results in my report _____
   I **do not** want polygenic risk score results in my report _____

Risks and limitations

- There may be standard risks associated with blood draw (e.g., bruising, bleeding).

- Inaccurate results can occur due to:
  i) mislabeled samples regardless of strict clinical and laboratory protocols in place to minimize this risk
  ii) inaccurate or incomplete reporting of medical and family history information
  iii) rare technical errors
  iv) other reasons

- This test might reveal biological relationships that were previously unrecognized, such as non-paternity or non-maternity if familial testing is undertaken.

- Genomic data may provide ancestry information (e.g. ethnicity) which could be used for the sole purpose of aiding the analysis of this test.

- Due to limitations in current medical knowledge or testing technology, a genetic variant may not be identified even though one exists and may contribute to or cause health problems.

- Genetic testing may identify a potential health problem in you; however, the test cannot predict the severity and precise clinical course of the genetic disorder. The

Alamya Health team and your healthcare provider can discuss this further with you if relevant.

- Genetic test results can trigger difficult emotions and, in some cases, cause tension within a family since identifying a genetic diagnosis can have implications for other family members.

- Test results might impact insurance coverage and rates, as well as employment. There may be local laws that help protect individuals undergoing genetic testing against discrimination. Please consider discussing this with your healthcare provider.

- This testing currently comprises both clinically validated testing and research testing and is undertaken in an accredited laboratory. In some situations, follow up confirmatory testing may be recommended..

Sample retention, data storage and database participation

After testing, Alamya Health will store your biological sample, if available, and genomic data for the option of reanalysis and reinterpretation in the future. Upon request for reanalysis/reinterpretation, Alamya Health will determine whether reanalysis or retesting would be preferred.

Alamya Health and its partners adhere to rigorous standards for personal health data storage as guided by laboratory regulatory bodies.

Alamya Health will share de-identified genomic and phenotypic (i.e. clinical) information with select genomic data repositories in order to improve the medical and scientific community's understanding of genetic information. Although sharing such information could support identification of potential health risks and health management, this could also result in no personal benefit. See notices of privacy practice for more details relative to the sharing of genetic information.

After testing is complete, your sample and/or data may be de-identified and used for internal validation, quality improvement and product development.

Billing

This test is not currently covered by insurance or institutional health plans and is only available via private pay.

I have reviewed this consent form and have been given the opportunity to ask questions with the Alamya Health team and/or my healthcare provider. By signing below, I consent to testing by Alamya Health as described in this consent form.	
Name (print)	**Date of birth** (MM-DD-YYYY)
Signature	**Date** (MM-DD-YYYY)