

Patient Name		Affix barcode label of Patient's sample here
Date of Birth		

Test Information

Genomic Inform® is a whole genome sequencing test that informs the healthy individual of known genetic health risks that have actionability for the individual (e.g. reproductive decisions, cancer/cardiac monitoring) and adult/late onset diseases. The test reports known pathogenic variants in the individual's genome (both nuclear and mitochondrial) and known and predicted pathogenic variants in focused gene lists that include: medically actionable ACMG recommended secondary findings, genetic risk factors, predisposition to cancer and cardiac diseases, carrier status of a list of genes known to be associated with severe early onset recessive disease (see below), variants in genes related to various additional conditions such as Factor V Leiden and Prothrombin. This test also includes variants in genes associated with adult/late onset neurological diseases.

The benefits and risks of Genomic Inform® are explained below. It is recommended that you receive genetic counseling from a licensed healthcare provider who can answer your questions about genetic testing and provide information about alternatives. Information about genetic counselors in your area is available at <https://www.nsgc.org/>.

This test does not utilize the individual's clinical personal and family history in the assessment, reporting, and/or interpretation of variants. If a diagnostic test is preferred, please contact Variantyx for alternative analyses.

Background

The purpose of genetic testing is to identify changes in the DNA sequence that are the cause of an affected individual's condition, that increase the likelihood of an individual developing a condition and/or passing a condition on to offspring. The resulting data is subjected to in-silico analyses optimized for small sequence changes (single nucleotide variants and deletion/insertions), structural variants in chromosomes (deletions, duplications, copy number variants), short tandem repeats (STRs) and mitochondrial variants (single nucleotide variants and small deletion/insertions and large deletions).

Technical Limitations

Genetic testing is accurate, but may not always identify a genetic variant. This test attempts to evaluate the entire DNA sequence, but may not be able to detect all DNA changes due to limitations in current technology. Certain regions of the DNA may not be well covered. Certain variant types may not be detectable such as methylation abnormalities, variants in genes with highly homologous pseudogenes, and variants in regions that are difficult to assay based on current technology. Unusual circumstances including bone marrow transplantation, blood transfusion, and variants that exist in only a small fraction of cells (mosaicism) may interfere with variant identification. Additionally, interpretation of the results is limited by the current medical understanding of disease and available scientific information. This test does not consider somatic mutations.

This test requires high-quality DNA. In some cases, an additional sample may be needed if the volume, quality and/or condition of the initial sample is not adequate. Samples submitted as genomic DNA will only be processed if the extraction was performed in a CLIA/CAP accredited laboratory.

Reporting Limitations

This test will not report variants related to infertility and carrier status of autosomal recessive or X-linked diseases that are outside of the list of genes below or not known to be pathogenic or likely pathogenic. Variants are not confirmed unless stated and confirmations are not part of the test turn-around time.

Possible Test Results

Test results will be issued as a single clinical report for the patient. The following types of variants will be considered: variants in genes consistent with predisposition to cancer, variants in genes consistent with predisposition to cardiac disorders, early- and late-onset conditions (see below), variants in genes consistent with risk factors and variants in genes consistent with carrier risk.

Report summary results - this section will include pathogenic or likely pathogenic variants that are associated with clinically significant findings.

Cancer predisposition - A result indicates that one or more genetic variants were identified that are consistent with cancer predisposition.

Cardiovascular predisposition - A result indicates that one or more genetic variants were identified that are consistent with predisposition to cardiovascular disorder.

Risk factor finding result - A result indicates that one or more genetic variants were identified that are consistent with increased risk of developing one or more late-onset conditions.

Carrier status - A result indicates that one or more genetic variants were identified that are associated with severe early onset recessive disorder, indicating carrier status for these diseases.



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Adult onset neurological disorders -

- Disorders due to repeat expansions: This test is limited to the following genes: *AR, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, C9ORF72, CACNA1A, CNBP, CSTB, DMPK, FMR1, FXN, HTT, JPH3, PP2R2B, PABPN1*.
- Disorders due to other variant types like small sequence changes, copy numbers variants across the genome: This also includes *APOE* genotyping testing for the risk of developing Alzheimer disease (AD).

Uncertain result / Variant of uncertain significance (VUS) - Variants of uncertain significance are not reported in this test.

Indeterminate result - An indeterminate result indicates that there were relevant genetic variant(s) identified in the analysis, but that there is uncertainty as to whether they are true variants or artifacts. Furthermore, it is considered that a repeat test will not resolve the technical uncertainty and orthogonal confirmation is necessary to resolve the result.

Inconclusive result - A technically inconclusive result indicates that there was an issue with the patient sample that resulted in data that the lab cannot interpret. It is considered that a repeat test will likely resolve the technical uncertainty and therefore a repeat sample is recommended to complete the analysis.

All reportable variants in the clinical report will be categorized as pathogenic or likely pathogenic utilizing the American College of Medical Genetics and Genomics (ACMG) / Association for Molecular Pathology (AMP) guidelines as published by Richards et al 2015 (for more information see: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4544753/>). Short tandem repeat variants with an expanded number of repeats will be reported as full mutation or premutation. Variants with an intermediate number of repeats will not be reported unless specifically requested. Even if this test finds DNA changes that are indicative of increased risk of developing a condition, the testing may not completely predict the severity of the condition, possible future problems, or response to treatment.

Single Nucleotide Variants

Genome-wide single nucleotide variants and small deletion/insertions (<50 bp) are reported if they are pathogenic or likely pathogenic.

Structural Variants

Structural variants are considered genome-wide and are reported if the structural variant is considered pathogenic/likely pathogenic.

Short Tandem Repeats

This test uniquely assesses short tandem repeats in genes for a number of disorders. The genes are organized into three sets. The first set includes genes (*AFF2, DIP2B, FMR1*) that are involved in early-onset intellectual disability. The second set includes genes (*AR, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, C9ORF72, CACNA1A, CNBP, CSTB, DMPK, FMR1, HTT, JPH3, FXN, PABPN1, PP2R2B*) that are involved in predominantly late-onset movement disorders, with or without cognitive involvement. Premutation variants which carry an increased risk of expanding to the full mutation size in offspring may also be identified.

The false negative rate for repeat expansions has not been determined for the following genes: *AFF2, ATXN10, CNBP, CSTB, DIP2B, JPH3, NOP56, NOTCH2NLC, PHOX2B, TBP*. The following genes can be assessed for normal repeat ranges only: *DIP2B, NOTCH2NLC*. Repeat counts above 45 will be reported as indeterminate.

Mitochondrial Variants

Mitochondrial variants are reported in the mitochondrial genome if they are pathogenic or likely pathogenic. Homoplasmic large deletions are reported, however duplications are not.

Cancer related genes:

ABRAXAS1; ANKRD26; APC; ATG2B; ATM; AXIN2; BAP1; BARD1; BLM; BMPR1A; BRCA1; BRCA2; BRIP1; CBL; CDC73; CDH1; CDK4; CDKN1B; CDKN1C; CDKN2A; CEBPA; CHEK2; CTNNA1; CTNNA3; DDX41; DICER1; EPCAM; ERCC4; ETV6; EXT1; EXT2; FH; FLCN; GATA2; GREM1; GSKIP; HOXB13; HRAS; KIT; MAX; MBD4; MECOM; MEN1; MET; MTF; MLH1; MSH2; MSH3; MSH6; MUTYH; NBN; NF1; NF2; NTHL1; PALB2; PAX5; PDGFRA; PHOX2B; PMS2; POLD1; POLE; POT1; PRKAR1A; PTCH1; PTEN; PTPN11; RAD51C; RAD51D; RB1; RECQL; RECQL4; RET; RPL5; RUNX1; SAMD9; SAMD9L; SDHA; SDHAF2; SDHB; SDHC; SDHD; SEC23B; SH2B3; SMAD4; SMARCA4; SMARCB1; SMARCE1; SOS1; SRP72; STK11; SUFU; TERC; TERT; TMEM127; TP53; TSC1; TSC2; VHL; WT1; WRN

Cardiovascular disorders related genes:

A2ML1; ABCC9; ACTA2; ACTC1; ACTN2; ACVRL1; AKAP9; ANK2; APOA5; APOB; BAG3; BMPR2; CACNA1C; CACNA2D1; CACNB2; CALM1; CALM2; CALM3; CASQ2; CAV1; CAV3; CBL; COL3A1; CRYAB; CSRP3; CTNNA3; DCHS1; DES; DMD; DOLK; DPP6; DSC2; DSG2; DSP; DTNA; EMD; ENG; EYA4; F2; F5; F9; FBN1; FHL1; FKTN; FLNC; GATA6; GATAD1; GDF2; GJA5; GLA; GPD1L; HCN4; JPH2; JUP; KCNA5; KCND3; KCNE1;



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KCNE2; KCNE3; KCNH2; KCNJ2; KCNJ5; KCNK3; KCNQ1; LAMA4; LAMP2; LDB3; LDLR; LDLRAP1; LMNA; MIB1; MYBPC3; MYH11; MYH6; MYH7; MYL2; MYL3; MYLK; MYLK2; MYO22; MYPN; NEXN; NKX2-5; NPPA; PCSK9; PKP2; PLN; PRDM16; PRKAG2; PRKG1; PROC; PROS1; RBM20; RYR2; SCN10A; SCN1B; SCN2B; SCN3B; SCN4B; SCN5A; SERPINC1; SGCD; SMAD3; SMAD4; SMAD9; SNTA1; SYNE1; TAZ; TBX1; TCAP; TGF2; TGF3; TGFBR1; TGFBR2; TMEM43; TNNC1; TNNT3; TNNT2; TPM1; TRDN; TRPM4; TTN; TTR; TXNRD2; VCL

Carrier status list of genes:

AAAS; ABCA12; ABCB11; ABCC6; ABCC8; ABCD1; ACAD8; ACAD9; ACADM; ACADS; ACADSB; ACADVL; ACAT1; ACOX1; ACSF3; ADA; ADA2; ADAMTS13; ADAMTS2; ADAR; ADGRG1; AFF2; AGA; AGL; AGPS; AGXT; AHCY; AHI1; AIRE; ALDH3A2; ALDH5A1; ALDH7A1; ALDOB; ALG6; ALMS1; ALPL; ALS2; ALX4; AMN; AMPD2; AMT; ANO5; ANOS1; AP1S1; AP3B1; AQP2; AR; ARG1; ARL13B; ARMC9; ARSA; ARSB; ARSL; ASAH1; ASL; ASNS; ASPA; ASPM; ASS1; ATM; ATP13A2; ATP6V0A2; ATP6V1B1; ATP7A; ATP7B; ATP8B1; ATR; ATRX; B3GALNT2; B4GAT1; B9D1; BBS1; BBS10; BBS12; BBS2; BBS4; BBS7; BBS9; BCKDHA; BCKDHB; BCS1L; BLM; BMP1; BSND; BTB; BTK; CANT1; CAPN3; CBS; CC2D2A; CCN6; CDH23; CEP104; CEP120; CEP290; CEP41; CERKL; CFTR; CHAT; CHM; CHRNA1; CHRND; CHRNE; CHRNG; CHST3; CIITA; CIT; CLCN5; CLN3; CLN5; CLN6; CLN8; CLRN1; CNGA3; CNGB3; CNTNAP1; CNTNAP2; COL17A1; COL1A2; COL27A1; COL4A3; COL4A4; COL4A5; COL6A1; COL6A2; COL6A3; COL7A1; COLQ; COX10; CPLANE1; CPS1; CPT1A; CPT2; CRB1; CRPPA; CRTAP; CSPP1; CTNS; CTSD; CTSK; CUL4B; CYBA; CYBB; CYP11B1; CYP11B2; CYP17A1; CYP19A1; CYP1B1; CYP27A1; CYP27B1; DAG1; DBT; DCLRE1C; DHCR7; DHDDS; DHH; DKC1; DLD; DLG3; DLL3; DMD; DMPK; DMXL2; DNAH5; DNAI1; DNAI2; DOK7; DPYD; DUOX2; DYM; DYNC2H1; DYNC2I2; DYSF; EBP; ECEL1; EDA; EDAR; EDN3; EDNRB; EGR2; EIF2AK3; EIF2B5; ELP1; EMD; EPM2A; ERCC2; ERCC3; ERCC4; ERCC5; ERCC6; ERCC8; ESCO2; ETTA; ETFB; ETFDH; ETHE1; EVC; EVC2; EXOSC3; EYS; F11; F2; F5; F8; F9; FAH; FAM161A; FANCA; FANCB; FANCC; FANCD2; FANCG; FGD1; FGD4; FH; FIG4; FKBP10; FKRP; FKTN; FMR1; FOXP3; FOXRED1; FXN; G6PC1; G6PD; GAA; GALT; GALE; GALK1; GALNS; GALNT3; GALT; GAMT; GBA; GBE1; GCDH; GCH1; GDAP1; GDF5; GFM1; GFPT1; GH1; GJA1; GJB1; GJB2; GJB3; GJB6; GK; GLA; GLB1; GLDC; GLE1; GMPPB; GNAT2; GNE; GNPAT; GNPTAB; GNPTG; GNS; GORAB; GP1BA; GP9; GPC3; GRHRP; GRIA3; GRIP1; GUCY2D; GUSB; HADH; HADHA; HADHB; HAX1; HBB; HCFC1; HEPACAM; HESX1; HEXA; HEXB; HGD; HGSNAT; HJV; HK1; HLCS; HMGCL; HOGA1; HPD; HPRT1; HPS1; HPS3; HPS4; HPS5; HPS6; HSD17B3; HSD17B4; HSD3B2; HSPG2; HYL5; IDS; IDUA; IFT172; IGF1R; IGHMBP2; IL1RAPL1; IL2RG; INPP5E; INPPL1; INSR; INVS; ITGA7; ITGB3; ITGB4; IVD; JAK3; KATNB1; KCNJ1; KCNJ11; KCNQ1; KCTD7; KDM5C; KIAA0586; KIF7; KLHL40; L1CAM; LAMA2; LAMA3; LAMB1; LAMB3; LAMC2; LARGE1; LCA5; LDLR; LDLRAP1; LGI4; LHCGR; LHX3; LIFR; LIPA; LMBRD1; LMNA; LMOD3; LOXHD1; LPL; LRPPRC; LSS; LYRM7; LYST; MAN2B1; MAA; MAT1A; MCCC1; MCCC2; MCOLN1; MECP2; MED12; MED17; MEFV; MESP2; MFN2; MFS2; MID1; MKKS; MKS1; MLC1; MLYCD; MMAA; MMAB; MMACHC; MMADHC; MMUT; MOCS1; MPI; MPL; MPV17; MPZ; MRE11; MTFMT; MTHFR; MTM1; MTR; MTRR; MTPP; MUSK; MYK; MYO15A; MYO7A; NAGLU; NAGS; NBN; NCF1; NDE1; NDRG1; NDUFA10; NDUFAF1; NDUFAF2; NDUFAF3; NDUFAF5; NDUFAF6; NDUFS1; NDUFS2; NDUFS4; NDUFS6; NDUFS7; NDUFS8; NDUFV1; NDUFV2; NEB; NEK1; NEU1; NHLRC1; NPC1; NPC2; NPHP1; NPHP3; NPHP4; NPHS1; NPHS2; NR0B1; NR2E3; NRXN1; NTRK1; OAT; OCA2; OCRL; OFD1; OPA3; OPHN1; OTC; P3H1; PAH; PANK2; PAX3; PC; PCBD1; PCCA; PCCB; PCDH15; PDE6C; PDHA1; PDHB; PEPD; PEX1; PEX10; PEX12; PEX13; PEX16; PEX2; PEX26; PEX3; PEX5; PEX6; PEX7; PFKM; PHF6; PHGDH; PIGN; PKHD1; PKLR; PLA2G6; PLEC; PLEKHG5; PLOD1; PLOD2; PLP1; PMM2; PMP22; PNPO; POLG; POLR1C; POLR1D; POMGNT1; POMGNT2; POMT1; POMT2; POP1; POR; POU1F1; PP1B; PPT1; PQBP1; PREPL; PROP1; PRPS1; PRX; PSAP; PSAT1; PTS; PUS1; PYGM; QDPR; RAB23; RAG1; RAG2; RAPSN; RARS2; RBM10; RDH12; RIPK4; RMRP; RNASEH2A; RNASEH2B; RNASEH2C; ROR2; RP2; RPE65; RPGRIP1L; RPL10; RPS6KA3; RS1; RTEL1; RXYLT1; RYR1; SACS; SAMHD1; SBDS; SBF2; SCN5A; SCNN1A; SCNN1B; SCNN1G; SDHA; SDHAF1; SELENON; SEPSECS; SERAC1; SERPINA1; SGCA; SGCB; SGCG; SGSH; SH2D1A; SH3TC2; SIL1; SIX6; SLC12A1; SLC12A3; SLC12A6; SLC16A2; SLC17A5; SLC19A2; SLC19A3; SLC22A5; SLC25A12; SLC25A13; SLC25A15; SLC25A20; SLC25A4; SLC26A2; SLC26A3; SLC26A4; SLC2A1; SLC35A3; SLC35D1; SLC37A4; SLC39A4; SLC3A1; SLC45A2; SLC4A11; SLC5A5; SLC6A8; SLC7A7; SLC7A9; SLC9A6; SMARCA1; SMN1; SMPD1; SMS; SOST; SRD5A2; ST3GAL5; STAR; STS; SUCLA2; SUFU; SUMF1; SUOX; SURF1; TAT; TAZ; TBCE; TCAP; TCIRG1; TCN2; TCTN1; TCTN2; TCTN3; TECPR2; TERT; TG; TGM1; TH; TIMM8A; TMEM138; TMEM216; TMEM231; TMEM237; TMEM67; TMEM70; TNNT3; TNNT1; TNXB; TP53; TPM3; TPO; TPP1; TREX1; TRIM32; TRIM37; TRIP11; TRMU; TSEN54; TSFM; TSHB; TSHR; TTC19; TTC21B; TTC37; TTC8; TTN; TTPA; TYMP; TYR; TYRP1; UBR1; UGT1A1; UNC13D; UPF3B; UROS; USH1C; USH1G; USH2A; VAPB; VHL; VLDLR; VPS13A; VPS13B; VRK1; VSX2; VWF; WAS; WDR35; WDR62; WNT10A; WNT7A; WRN; XIAP; XPA; XPC; ZDHHC9; ZFYVE26; ZIC3; ZMPSTE24

Limitations for the above lists of genes:

The following genes are not fully covered: *GREM1, WT1, CACNA2D1, DCHS1, F9, KCNE1, MYH6, KCNE1L, AFF2, ATRX, CBS, CLCN5, CLN8, GP1BA, MLYCD, NEB, RIPK4, SLC6A8, TNXB*. Therefore there may be variants in certain regions of these genes that are not identified with this test.

The following genes have regions that are not unique: *ABCC9, DUOX2, GK, MYH7, SDHA, ABCC6, TMEM231, TTN, VWF, GBA, OPHN1, PMS2, SMN1*. Therefore there may be variants in these genes that cannot be assigned to the correct location and limit the interpretation.

Patient Confidentiality

To maintain confidentiality, test results will only be released to the ordering healthcare provider or ordering laboratory, and upon your request, to additional healthcare provider(s) indicated on this test requisition form. Test results will only be disclosed to others by your written consent and/or if demanded by a court of competent jurisdiction. It is your responsibility to consider the possible impact of test results on insurance rates, the ability to obtain disability, life or long-term care insurance and employment. The Genetic Information Non-discrimination Act (GINA), enacted by the US Federal Government, provides some protection against discrimination by health insurance companies and employers based on genetic test results, but does not cover life, disability, or long-term care insurance. Information about GINA is available at <https://www.genome.gov/10002328>.

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Anonymized information obtained from the test may be included in variant and allele frequency databases used to help healthcare providers and scientists understand human disease, as well as in scientific publications. Names and personal identifying information will not be revealed. Separate from the above, if there are opportunities to participate in research relevant to your condition, and you have consented for recontact, Variantyx may contact you or your healthcare provider for research purposes.

Turnaround Time

The turnaround time (TAT) of this test can be found on the Variantyx website, which begins at the time of sample receipt. Please note that the following scenarios will likely result in extension of the turnaround time (1) when the DNA sample fails QC and is determined to be insufficient for testing, requiring collection of a new sample; (2) when the test is sent for orthogonal confirmation at an external laboratory. In the second scenario, the turnaround time can be expected to be extended by the turn around time of the external laboratory plus 1 week.

Sample Retention

DNA extracted from submitted samples may be stored for at least 3 months following completion of testing and may be discarded thereafter. After completion of testing, anonymized DNA may be used for test development and improvement, internal validation, quality assurance and training purposes before being discarded. The raw data files containing the DNA sequences (BAM files) can be transferred to the tested individuals at no additional charge, upon request, for a minimum of two years.

New York state residents: No other test shall be performed on this sample except the test ordered by the clinician, unless waived by the patient or authorized individual. In addition, the patient's biological sample will be destroyed within 60 days or upon the completion of testing, unless waived by the patient or authorized individual. Orthogonal confirmation of results at a reference lab cannot be performed unless the patient or authorized individual provides permission to do so.

Before making the decision to consent to testing, there are some things to consider:

- Individuals without symptoms may find this information beneficial to know for family or life planning decisions.
- However you may have negative feelings upon learning of your risk to develop these conditions, such as severe depression and anxiety.
- In some cases, test results could impact your ability to obtain or retain life insurance and/or employment.
- If you have any concerns about this test we recommend discussing this with your healthcare provider.
- You may get the following results:
 - Pathogenic/likely pathogenic finding: A genetic change was found and individuals are likely to show symptoms of a specific condition within their natural lifespan.
 - Intermediate (gray zone) finding: A genetic change was found. An individual with an intermediate result may or may not show symptoms in their natural life. There is a higher risk to have a child with the condition.
 - Carrier status: A genetic change was found in an individual. That individual is not likely show symptoms of the condition in their natural lifespan, but is at a higher risk to have a child with the condition.
 - No pathogenic/likely pathogenic findings: You are not found to be at risk for the conditions that were tested. A negative result does not mean there is no risk to develop these or other neurological genetic conditions in the future. There may be other conditions that were not identified by the test.

Examples of possible findings you can learn more about:

1.) Adult-onset neurological disorders: These are conditions that affect your nervous system later in adulthood (40s-60s or sometimes later). Each condition may present differently. All of these conditions are progressive, meaning they worsen over time and shorten one's life. Symptoms of these conditions may vary. Some have muscle weakness and may lose their ability to walk or perform daily activities without assistance. Some conditions can impact the way you move different parts of your body, including your eyes or ability to swallow your food. Someone's personality or ability to think clearly can also be altered. Currently, there are no cures and there may be limited treatment or prevention options for these conditions. Some of these conditions include amyotrophic lateral sclerosis (ALS or Lou Gerhig's disease), ataxias, dementias, and/or muscular dystrophies.

2.) Huntington Disease (HD) and Huntington disease-like 2 (HDL2): HD and HDL2 are also progressive diseases that affect one's nervous system typically later in adulthood (30s-50s or sometimes later), although childhood onset can happen in rare cases. Symptoms of HD and HDL2 include uncontrolled bodily movements, which can make it difficult to walk, speak, swallow, or perform daily activities without assistance. Those with HD and HDL2 also have difficulty with memory, focusing on tasks, and organization. Mood is also impacted, as affected individuals may become irritable, sad, or lack emotional connection all together. Currently, there are no cures and there may be limited treatment or prevention options for these conditions.

Alzheimer disease risk (APOE): Alzheimer disease is a progressive condition that affects a person's ability to remember new or old information, organize tasks and ideas, and identify people and/or places around them. Symptoms tend to present after the age of 65. Currently, there is no cure for Alzheimer disease. APOE is a gene (or genetic information) that comes in three different forms:

- a.) APOE (e2): may protect a person from developing Alzheimer disease
- b.) APOE (e3): doesn't appear to impact one's risk of developing Alzheimer disease





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c.) *APOE* (e4): may increase one's risk to develop Alzheimer disease

It is important to note that there are many people who have an e4 gene that do not develop Alzheimer disease. Alternatively, there are many people who develop Alzheimer disease without the e4 gene.

In summary, you have the option to learn of your risk to develop certain life-limiting conditions, for which there are no cures and limited treatments. For some this knowledge may help in making future planning decisions (family planning, household accommodations, career/education planning) and for others knowing this information may cause severe depression and/or anxiety. This is a deeply personal decision and it is strongly recommended that all implications, both positive and negative, be considered with your care team before ordering this test.



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Consent

I have discussed Genomic Inform[®] with my healthcare provider including the purpose and procedure, risks, benefits and alternatives. I have been given an opportunity to ask questions about the test, and any questions I had were answered to my satisfaction. I acknowledge that I have sufficient information and understanding to give this informed consent.

1. I give permission to Variantyx and their affiliates to extract and sequence my DNA and perform genetic testing as described.
2. I give permission for my anonymized DNA to be used by Variantyx and their affiliates for test development or improvement, internal validation, orthogonal variant confirmation at an outside referral laboratory and/or quality assurance and training purposes.
3. I give permission for my anonymized sample and clinical information to be included in variant and allele frequency databases and publications. My name or other personal identifying information will not be used in or linked to any databases or publications.
4. I give / do not give permission for Variantyx to contact me or my healthcare provider about research studies. If no option is selected, no contact will be made.
5. For NY state residents : By checking this box I give permission for Variantyx and their affiliates to retain any remaining sample longer than 60 days for testing completion, test development/improvement, internal validation, orthogonal variant confirmation at an outside referral laboratory and/or quality assurance and training purposes.

It is strongly recommended that you receive genetic counseling from a licensed healthcare provider who can answer your questions about genetic testing and provide information about alternatives. Information about genetic counselors in your area is available at <https://www.nsgc.org/>.

Patient /Guardian first name _____ Patient /Guardian last name _____

Patient /Guardian signature _____ Date _____

