# Informed Consent for NIPS + Carrier Screening

FM173-2

This form describes the benefits, risks, and limitations of these screening tests. By signing below, I attest that I have read this form carefully—and asked any questions I may have of my healthcare provider—before making my decision about testing.

## Non-invasive prenatal screening

If my healthcare provider has ordered Invitae Non-Invasive Prenatal Screening (Invitae NIPS), my signature below attests to the following:

1. **PURPOSE:** The purpose of Invitae NIPS is to screen my pregnancy for certain chromosomal abnormalities, also known as "aneuploidies." This test gives information about whether there may be extra copies (trisomy) of chromosomes 21, 18, and 13, and the option to know if there is an extra copy of a sex chromosome (X or Y), and/or a missing copy of a sex chromosome (MX). Fetal sex may also be reported. In addition, the option to screen for the following microdeletion (small, missing parts of chromosomes) syndromes: 1p36 deletion, 4p- (Wolf-Hirschhorn syndrome), 5p- (cri-du-chat syndrome), 15q11.2 (Prader-Willi syndrome/Angelman syndrome), 22q11.2 deletion (DiGeorge syndrome or velocardiofacial syndrome) is also available. For chromosomes 21, 18, and 13, Invitae NIPS is validated in singleton and twin pregnancies. In twin pregnancies, sex chromosome testing can only screen for the presence or absence of the Y chromosome, and not for extra or missing sex chromosomes. Invitae NIPS can be performed as early as 10 weeks 0 days gestational age. I will consult my healthcare provider if I would like more information about this screening test, including risks, limitations, performance data, error rates, descriptions of the conditions being screened, and what these results may mean to my pregnancy.
2. **METHODS:** Invitae NIPS screens for specific chromosomal abnormalities by looking at the DNA (genetic material) in my blood. The sample of blood includes a combination of both my DNA and the DNA from the pregnancy. A technology called next-generation sequencing (NGS) is used to count the amount of DNA from each tested chromosome and/or from specific regions of chromosomes. The laboratory then uses an analysis method to determine if each of the conditions I have elected to test for is likely to be present or absent.

   Up to two tubes of my blood will be drawn and sent to Invitae Corporation. Invitae will process and track my blood sample and send it to Verinata Health, Inc., a wholly owned subsidiary of Illumina, Inc., which will then perform the screen.
3. **SEX OF PREGNANCY:** Depending upon the option I and my healthcare provider elect, the test results may include the sex of the pregnancy. If I do not wish to know the sex, I will tell my healthcare provider not to disclose this information to me. Depending upon the test ordered, I may not be able to prevent learning the sex of my pregnancy. In rare instances, incorrect sex results can occur.
4. **RISKS AND LIMITATIONS:** This screening test looks only for specific chromosomal abnormalities. This means that other chromosomal abnormalities may be present and could affect my pregnancy. A "No Aneuploidy Detected" result does not guarantee a healthy pregnancy or baby and does not eliminate the possibility that my pregnancy may have birth defects, genetic conditions, or other conditions, such as open neural tube defects or autism.

   There is a small possibility that the test results might not reflect the chromosomes of the fetus, but may reflect chromosomal changes of the placenta (confined placental mosaicism, CPM) or of my DNA (maternal chromosomal abnormalities). While these tests are not designed to assess my health, in some cases, information about my health may be revealed directly or indirectly (e.g., when combined with other information). Examples include maternal XXX, sex chromosome status or benign or malignant maternal neoplasms. In a twin pregnancy, the status of each individual fetus cannot be determined.

   This test, like many tests, has limitations, including false negative and false positive results. This means that the chromosomal abnormality being tested for may be present even if I receive a negative result (this is called a "false negative"), or that I may receive a positive result for the chromosomal abnormality being tested for, even though the abnormality is not actually present (this is called a "false positive").

   In the case of a twin pregnancy, the presence or absence of Y chromosome material can be reported. The occurrence of sex chromosome aneuploidies cannot be evaluated in twin pregnancies. In the case of a vanishing twin, the test result may reflect the DNA of the vanishing twin, leading to a higher probability of false positive or false negative results.

   No irreversible clinical decisions should be made based on these screening results alone. If definitive diagnosis is desired, chorionic villus sampling or amniocentesis would be necessary. In some cases, other testing may also be necessary. Some rare chromosomal aneuploidies may occur only in mosaic form. Clinical consequences depend on the chromosome involved and can not be predicted prenatally.

   I will consult my healthcare provider for more information about my results and what they may mean for my pregnancy, what options I will have for further testing, and whether additional testing is recommended for me based on my clinical history.

5. **SECONDARY FINDINGS:** In the course of performing the analysis for the indicated tests, information regarding other chromosomal alterations, also known as "secondary findings" may become evident. Invitae and Illumina's policy is to NOT REPORT on any secondary findings that may be noted in the course of analyzing the test data.

## Carrier screening

If my healthcare provider has ordered Invitae Carrier Screening, my signature below attests to the following:

1. **PURPOSE:** I understand that my healthcare provider has ordered one or more tests offered by Invitae Corporation ("Invitae") to determine if I am a carrier for one or more genetic disorders. A carrier has a change in a specific gene or genes that increases his or her chance of having a child with a genetic disorder or birth defect. The test(s) ordered may include any one or more of the comprehensive list of tests that can be found at www.invitae.com.
2. **METHODS:** Testing is performed on a small sample of blood, saliva, or isolated DNA. Once collected, the sample will be sent to Invitae for testing.
3. **RESULTS:** I understand that if the test results are positive, I may be a carrier of the disorder tested. I may also learn that I have an undiagnosed disorder, that I carry pathogenic variants in multiple genes, or that I am at increased risk for a different disorder caused by pathogenic variants in one of the genes tested. Invitae does not report all DNA changes that could be disease causing. If test results are negative, my risk to be a carrier of, or to have the disorder tested, is reduced but not eliminated.
4. **INCIDENTAL FINDINGS:** In the course of processing the ordered tests, Invitae may find changes in genes that are unrelated to the clinical concern that prompted my having the test but are clearly associated with a significant risk for disease and could therefore negatively impact my own health and/or the health of my close relatives. These are known as "incidental findings." Invitae will report such incidental findings when (and only when) there are accepted medical interventions available that can prevent or treat the health consequences of these genetic changes.
5. **RISKS AND LIMITATIONS:** My healthcare provider has explained the effectiveness and limitations of the test(s), and I understand that the test results may not provide definitive conclusions regarding reproductive risk. While this testing is highly accurate, rare testing errors may occur. Further testing may be warranted for myself or my partner and this additional testing may or may not be covered by insurance. Accurate results may not be obtained for reasons including but not limited to sample mix- up, bone marrow transplant, recent blood transfusion, or technical problems. Sometimes for technical reasons, results cannot be generated. Additional samples may be needed if results are not generated.
6. **TESTING OF ADDITIONAL FAMILY MEMBERS:** may be requested, which could discover previously unknown information about family relationships, such as nonpaternity (someone who is not the biological father), or adoption. I have discussed with my healthcare provider if and/or how such results will be shared with me.

## Additional information

Whether my healthcare provider ordered Invitae Carrier Screening and/or Invitae Non-Invasive Prenatal Screening, my signature below attests to the following:

1. **NONDISCRIMINATION:** There are state and federal laws that prohibit discrimination against individuals for the purpose of employment or obtaining health insurance and that prohibit insurers and employers from seeking an individual's genetic information without consent. In accordance with such laws, Invitae will not disclose or interpret my genetic information for use by employers or insurers. However, it is my responsibility to consider the possible impact of my test results as they relate to insurance rates, obtaining disability or life insurance, and employment. The Genetic Information Nondiscrimination Act (GINA), a US Federal law, provides some protections against genetic discrimination. For more information on GINA, visit www.genome.gov/10002328.
2. **PREGNANCY OUTCOME INFORMATION:** Collecting information on my pregnancy after testing is part of a laboratory's standard practice for quality purposes and is required in several states. As such, Illumina, Invitae or their respective designees may contact my healthcare provider to obtain this information. By executing this informed consent, I agree to allow my healthcare provider to provide this information to Illumina, Invitae or their respective designees.
3. **PRIVACY:** I understand that my data and personal information will be stored and protected in compliance with applicable regulatory requirements (e.g., HIPAA and equivalent protections), and I acknowledge that I have read and understand Invitae's Privacy Policy and Notice of Privacy Practices (available at www.invitae.com/privacy).

4. **GENETIC COUNSELING:** I understand that Invitae recommends that I consult with a genetic counselor before consenting to this test and a genetic counselor or my healthcare provider about my results. For a list of medical geneticists and counselors who may be available in my area, I may visit the National Society of Genetic Counselors website at www.nsgc.org. Further testing or additional physician consults may be warranted.
5. **NON-COVERED SERVICE:** In the event my insurer denies coverage for the Invitae NIPS and/or carrier test for any reason, including deeming the test to be a non-covered service or not medically necessary, I agree that Invitae may bill me directly for this service and I will remit payment directly to Invitae.
6. **CROSS-BORDER DATA TRANSFER:** If I am a resident of a country other than the United States, my specimen and associated health information will be sent to the United States in order for the testing to be completed. My data and personal information, including my results, will be stored in the United States. As part of the testing, additional health information about me will be created and maintained. My country may consider the legal privacy protections in the United States to be inadequate.
7. **DISCLOSURE OF TEST RESULTS:** The clinical reports are released only to the certified healthcare professional(s) listed on the test requisition from. Clinical reports are confidential and will only be released to other medical professionals with my explicit written consent. It has been explained to me that my clinical report is available for me to download from within the Invitae patient portal (www.invitae.com/patients/signin) after it has been released by my healthcare professional(s) or upon request in accordance with applicable law.
8. **RECONTACT FOR CLINICAL UPDATES:** I understand that knowledge of genetic information will improve over time, that new information may become available in the future that could impact the interpretation of my results, and that Invitae may notify me of clinical updates related to my genetic profile (in consultation with my primary clinician as indicated). I may request additional notifications and resources relevant to my genetic profile by creating an Invitae patient portal account at www.invitae.com/patients/signin.
9. **RESEARCH:** Sharing de-identified genetic data can significantly accelerate medical research for both individual patients and society as a whole. Invitae encourages patients to choose to share their genetic variants with the medical and scientific community to help accelerate our understanding of genetic conditions, improve genetic testing, find new therapies, and eventually prevent disease. Invitae will share results after they are de-identified, meaning that Invitae removes any information that identifies or could be used to identify me personally.
    a. **De-identified genetic information:** I understand Invitae may store and retain indefinitely at its discretion, except as prohibited by law, and use and/or disclose to third parties, including public databases, my de-identified genetic information for quality assurance, test development and/or validation, research, and/or educational purposes.
    b. **De-identified samples:** I understand Invitae may store and retain indefinitely at its discretion, except as prohibited by law, and use and/or share with third parties my de-identified samples for quality assurance, test development and/or validation, research, and/or educational purposes.
    c. **Future contact regarding research:** I permit Invitae to contact me in the future about research opportunities that may be related to my condition or my test results.

**All patients (other than residents of those states specifically identified below):** I control how Invitae uses my data. I understand that I can log in to the Invitae patient portal (www.invitae.com/patients/signin) and click on Account Settings > Preferences if I would like to change my preferences with respect to how Invitae uses my de-identified data. If I choose to restrict the use of my de-identified genetic information or sample(s): I understand that to the extent that such information has already been used or shared, it cannot be retracted or destroyed, and I understand that my de-identified genetic information and/or sample(s) may still be used for quality assurance, test development and/or validation; shared with public databases; and/or (in connection with de-identified information) used or disclosed to third parties, not on an individual basis but as aggregated information for research or education purposes.

**Residents of New York or Alaska:** I understand that my sample(s) (or my minor child's sample(s)) shall be destroyed no more than 60 days after they were taken or at the end of the testing process, whichever occurs later, unless a longer period of retention is expressly authorized. I understand that I can authorize a longer period of sample retention if I log in to the Invitae patient portal (www.invitae.com/patients/signin) and click on Account Settings > Preferences (my "account settings"). I further understand that if I authorize my samples to be retained longer, I can change my account settings to allow the use of my de-identified samples for research and/or education. If I do not opt in, I understand that my de-identified samples will not be used for quality assurance, test development and/or validation; nor and/or used, disclosed to, or shared with third parties for research or education purposes. I further understand that I can change my account settings to allow the use of my de-identified genetic information for research and/or education, or to allow Invitae to contact me in the future regarding research opportunities. If I do not opt in, I understand that my de-identified genetic information may still be used for quality assurance, test development and/or validation; shared with public databases; and/or used, disclosed to, or shared with third parties, not on an individual basis but as aggregated information, for research or education purposes.

**Residents of Florida, Massachusetts, Minnesota, New Hampshire, Texas, Vermont and countries other than the United States:**
I understand that I can log in to the Invitae patient portal (www.invitae.com/patients/signin) and click on Account Settings > Preferences to opt in to allow the use of my de-identified genetic information or samples for research and/or education, or to allow Invitae to contact me in the future regarding research opportunities. If I do not opt in, I understand that my de-identified genetic information and/or samples may still be used for quality assurance, test development and/or validation; shared with public databases; and/or used, disclosed to, or shared with third parties, not on an individual basis but as aggregated information, for research or education purposes.

10. **VOLUNTARY CONSENT:** I understand that my consent to testing is voluntary, and I may choose not to have my sample tested.
11. **COPY OF THIS FORM:** I have a right to receive a copy of this form.

## BY SIGNING BELOW, I ATTEST TO THE FOLLOWING:

1. I have read (or had read to me), and that I understand, the information provided in this consent;
2. I have all the information I want, and all my questions have been satisfactorily answered; and
3. I hereby consent to carrier testing.

| Patient signature | Date |
|---|---|
| | |

| Patient name (please print) | Email address |
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## HEALTHCARE PROVIDER STATEMENT

By signing below, I attest that:

1. I am the referring physician or authorized healthcare provider;
2. I have explained the purpose of test described above;
3. The patient has had the opportunity to ask questions regarding this test and/or seek genetic counseling; and
4. The patient has voluntarily decided to have this test performed by Invitae.

| Healthcare provider signature | Date |
|---|---|
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