**Informed consent**

# Myriad Genetics® FirstGene® Multiple Prenatal 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 seek genetic counseling prior to signing this form.

## Purpose

- The FirstGene screen is a prenatal cell-free DNA screen that uses a blood sample from a pregnant person to concurrently assess:
  - The pregnant person's carrier status
  - Fetal status regarding single-gene conditions (such as cystic fibrosis and spinal muscular atrophy)
  - Fetal status regarding common chromosomal abnormalities (such as Down syndrome, trisomy 18, and trisomy 13)
  - Fetal RhD status (when the pregnant person is known to be RhD negative)
- The conditions assessed in the FirstGene screen can lead to health issues including birth defects, intellectual disabilities, and pregnancy complications. The severity of these can vary.
- The FirstGene screen is intended for pregnant persons who are at least 8 weeks pregnant with a single fetus and who are genetically related to the fetus.
- The FirstGene screen is designed to determine whether you, the pregnant person, carry genetic changes called mutations in specific genes that could cause serious genetic conditions in your children. For most of the recessive ("single gene") conditions on the FirstGene screen, typically both reproductive partners must carry a mutation in the same gene for their children to be at risk of being affected and developing symptoms of that condition.
- The FirstGene screen uses cell-free DNA (cfDNA) to provide a fetal risk assessment for these recessive conditions. When the fetus is predicted to have two mutations in the same gene, they are likely to be affected with the condition indicated, and at a higher risk of developing the associated symptoms. When the fetus is predicted to have one mutation in a gene, it is likely that they are a carrier of that condition, which is generally not associated with a risk of developing symptoms.
  - In the case of fragile X, only the mother needs to carry a mutation for their children to be at risk of developing symptoms.
- The FirstGene screen also uses cfDNA to assess the risk of a current pregnancy having a chromosome condition, such as Down syndrome, trisomy 18, and trisomy 13. These conditions can lead to health issues including birth defects and intellectual disabilities, and the severity of these symptoms can vary.
- The FirstGene screen assesses the RhD status of the fetus when the pregnant person is identified as being RhD negative, and therefore at risk for maternofetal rhesus incompatibility if the fetus is RhD positive. Maternofetal rhesus incompatibility can be managed by using a medication that prevents the pregnant person's immune system from attacking fetal cells. The FirstGene screen can help assess whether your pregnancy has a risk of maternofetal rhesus incompatibility due to fetal RhD status. You should speak with your healthcare provider to determine the most appropriate treatment options when there is a risk of maternofetal rhesus incompatibility
- A screening test indicates whether there is an increased or decreased risk for a condition, while a diagnostic test indicates whether the condition is present or not. Fetal and maternofetal risk assessments performed with the FirstGene screen are screening tests.

## Benefits

- Fetal recessive disease and chromosomal risk assessments can help you make more informed pregnancy management decisions. 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 providers about how to best plan and prepare for birth.
- Carrier screening results from the FirstGene screen can help you make more informed decisions regarding your current and future pregnancies. Carrier screening results may also benefit your other family members. If you are identified as a carrier of a recessive condition, your biological relatives are more likely to test positive for the same mutation(s), thereby allowing them to discover previously unknown conditions and risks.
- Fetal RhD status can help assess the risk for complications in your pregnancy that can occur as a result of maternofetal rhesus incompatibility. You should speak with your healthcare provider to determine the most appropriate treatment options when there is a risk of maternofetal rhesus incompatibility.

## What you might learn

With the FirstGene screen, fetal risk assessments use DNA from the pregnant person as well as cfDNA to assess the risk of fetal chromosomal abnormalities (Down syndrome, trisomy 18, trisomy 13, 22q11.2 deletion syndrome) and recessive genetic conditions (cystic fibrosis, spinal muscular atrophy, Hb Bart syndrome, beta globin-related

*Instructions for healthcare providers:*
This document is provided for your convenience and can be used at your discretion and some states may have specific documentation requirements for informed consent.

hemoglobinopathy (including beta thalassemia and sickle cell disease), hexosaminidase A deficiency (including Tay-Sachs disease), congenital disorder of glycosylation, PMM2-related, medium-chain acyl-CoA dehydrogenase deficiency, Canavan disease, Smith-Lemli-Opitz syndrome, phenylalanine hydroxylase deficiency, familial dysautonomia, Pompe disease, carnitine palmitoyltransferase II deficiency, autosomal recessive polycystic kidney disease, PKHD1-related, methylmalonic acidemia, MMUT-related, galactosemia, methylmalonic aciduria and homocystinuria, cblC type, GNPTAB-related disorders, homocystinuria, CBS-related). The pregnant person will also receive insight on carrier risk for fragile X.

- The following describes the possible positive results for fetal risk assessments with the FirstGene screen:
  - Positive: Fetal Aneuploidy - an “Aneuploidy Detected” result indicates that the analysis of cfDNA in the pregnant person's blood is consistent with an increased risk of the pregnancy being affected by a specific chromosome condition (e.g. Down syndrome).
  - Positive: Recessive/"Single Gene" Condition
    - A Positive “Fetus Predicted Affected" result indicates that the analysis of the cfDNA in the pregnant person's blood is consistent with the fetus having two mutations in the same gene and is at high risk of being affected with the condition indicated.
    - A Positive "Fetus at Increased Risk” result indicates that the analysis of the cfDNA in the pregnant person’s blood is consistent with the fetus having at least one mutation in the gene indicated, and is at increased risk of being affected with the condition indicated.
- The following describes positive results for patient carrier screening analysis performed in the FirstGene screen:
  - A "Positive Carrier" result indicates that the analysis of the DNA in the pregnant person's blood has found a mutation in the pregnant person's blood, and that they are a carrier for the condition noted. You may be identified as a carrier of one or more conditions.
  - A Positive “High Reproductive Risk” result indicates that the analysis of the DNA in the pregnant person's blood is consistent with the pregnant person being a carrier of fragile X syndrome, which is inherited in an X-linked pattern. Note that fragile X analysis in not performed on the cfDNA.
- The following describes positive results for RhD maternofetal genotype incompatibility:
  - A “Predicted Fetal Status: RhD Positive” result indicates that the analysis of the cfDNA in the pregnant person's blood is consistent with the fetal RhD status being incompatible with your RhD status.
- You may receive a positive result for more than one condition. All "positive" results should be discussed with your healthcare provider to determine next steps, including pursuing diagnostic testing (such as chorionic villus sampling (CVS) and/or amniocentesis) and additional evaluation, as well as genetic counseling. While follow-up testing is recommended according to professional guidelines, the decision of whether to pursue additional testing is entirely yours and should be made in consultation with your healthcare provider.
- Negative results
  - A Negative “Low Risk” fetal chromosome screen result indicates a reduced risk of your pregnancy to be affected by the chromosomal conditions screened. This result indicates a significantly reduced likelihood but does not entirely eliminate the possibility of your pregnancy to be affected by one of these conditions.
  - A Negative "Low Risk" fetal single gene result indicates that no mutations were found in the analysis of the fetus's DNA. This reduces but does not eliminate the risk for your fetus to be a carrier of a condition on the panel
  - A Negative Patient Carrier Screen: Negative results indicate that no mutations were found in the analysis of your DNA. This reduces but does not eliminate the risk for you to be a carrier of a condition on the panel.
  - A “Predicted Fetal Status: RhD Negative” result indicates that the analysis of the cfDNA in the pregnant person’s blood is consistent with the fetal RhD status being compatible with your RhD status.
  - Negative results significantly reduce, but cannot eliminate, the risk for either the pregnant patient to be a carrier for the recessive conditions screened or the possibility of the pregnancy to be affected by one of the chromosomal or recessive conditions screened and do not guarantee a healthy pregnancy/child.

## Procedure

- The FirstGene screen can be done 8 weeks into pregnancy or later.
- A blood sample is taken from your arm and sent to Myriad for screening.
- Except in rare cases, your specimen will be kept for a maximum of 180 days.*

## Risks

- Genetic testing may reveal sensitive information about your health and your family members, as well as your reproductive partner and their family members.
- Your results may lead to your healthcare provider recommending additional testing and evaluation of your pregnancy, or otherwise impact medical decisions.
- Your results may provide information that can have an impact on your medical decisions.

## Limitations of The FirstGene Multiple Prenatal Screen

- The FirstGene fetal chromosome screen is designed to detect most pregnancies in which the fetus has one of the chromosome conditions listed above but does not detect or look for all known genetic diseases, syndromes, or birth defects.
- The FirstGene carrier screen and fetal single gene screen is designed to detect deleterious mutations associated with recessive conditions. It cannot detect every mutation, nor does it look for all known genetic diseases or syndromes.

- The FirstGene fetal RhD screen is designed to detect fetal RhD deletions. It cannot detect all genotypes associated with RhD negative status, nor does it look for all causes of maternofetal rhesus incompatibility.
- As with all medical screening tests, there is a risk of error, including a false positive or false negative result.
  - A "False positive" refers to identifying a mutation or aneuploidy that is not present in the sample.
  - A "False negative" is the failure to detect a mutation or aneuploidy 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 the FirstGene screen. 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.
- It is important for any relevant personal medical history, family history, and pregnancy history to be communicated to your healthcare provider and Myriad for accurate interpretation of your results.
- The FirstGene screen can only be performed on singleton pregnancies.
- The FirstGene screen is not available for pregnancies achieved with a donor egg.
- Additionally, pregnancies that have experienced the demise of one or more fetuses may not receive reliable results. Please check with your healthcare provider if this impacts your pregnancy and you desire screening.
- If either the patient or the pregnancy has cells with different chromosome contents (called "mosaicism"), the results of the FirstGene screen may not be accurate.
- If the patient and partner are related, the risk of a fetal false negative for single gene testing is much higher. Patients should inform their providers of known consanguinity. FirstGene testing is not appropriate for these individuals.

### Alternatives

- There are various options available that provide insight into the genetic health of the pregnancy, the carrier status of a pregnant person, and maternofetal rhesus incompatibility. These should be discussed with your healthcare provider.
- Diagnostic testing, such as chorionic villus sampling (CVS) or amniocentesis, is capable of providing definitive information about the genetic health of the pregnancy.
- As all screening for genetic conditions and chromosome differences is completely voluntary, a patient has the right to decline all such tests.

### Privacy

- Genetic information is protected under the federal Genetic Information Nondiscrimination Act (GINA), which generally prohibits health insurers and employers from discriminating against you based on your genetic information. However, you should be aware that current federal laws do not specifically prohibit genetic discrimination for life insurance, long-term care insurance and disability insurance. More information about GINA and its limitations is available at ginahelp.org
- Your FirstGene screen results will be reported to your healthcare provider or his/her/their agent.
- By agreeing to testing and signing this consent, you hereby authorize Myriad to share your FirstGene screen results with other authorized representatives that you've identified to Myriad or your healthcare provider, or as otherwise allowed by law.
- Myriad may find information that is not included on the original screen requested by your healthcare provider and may report these additional results, if clinically relevant. You authorize Myriad to share these results with you and your healthcare provider.
- You authorize Myriad to contact you about your screen, test or sample, as well as additional products or services offered as part of your FirstGene screen (e.g., cost estimates), and additional products and/or offers that may be relevant and/or interesting to you.
- When receiving notifications from Myriad and its affiliates, email, SMS and text messages may be sent to you to inform you that Myriad has prepared an estimate of your testing costs and to provide you with status updates about your test and results. These communications may be unsecure and carry typical risks associated with such transmissions. By agreeing to testing and signing this consent, you agree that (a) you understand the foregoing and understand the typical risk of email, SMS and text message transmissions, and (b) you give Myriad permission to communicate with you by email, SMS, or text message.
- Please refer to Myriad's Notice of Privacy Practices, available on the Myriad website, for additional information about Myriad's privacy practices, including how your protected health information may be shared with third-party vendors and service providers that we partner with to provide testing services to you, and your rights related to your protected health information.

### Quality Assurance and Research*

- Unless you contact us to request otherwise, by agreeing to testing, you authorize Myriad and its partners to use your sample and any information derived from your sample or otherwise collected about you for educational and/or research purposes. You will not be compensated for this use.
- Myriad may collect additional information regarding the outcome of your pregnancy from your healthcare provider for purposes of quality assurance and research.
- De-identified information may additionally be submitted to external research databases.
- You authorize Myriad to contact you about potential educational and/or research opportunities.
- Please contact us at **prenatalsupport@myriad.com** or at **+1-888-268-6795** if you wish to opt out of such research or future contact.

*Samples from residents of New York state will not be retained for more than 60 days after collection and will not be included in research studies. Specimens from all other states will be retained and maintained in accordance with applicable laws and regulations.

**Financial responsibility**

- By agreeing to testing, you authorize Myriad to submit to your insurance carrier any and all of the information, including test results, necessary for processing your insurance claim.
- By agreeing to testing you also authorize Myriad to obtain a consumer credit report on you from a consumer reporting agency selected by Myriad. You understand and agree that Myriad may use your consumer credit report to confirm whether your income qualifies you for financial assistance. You further understand that this is not a credit application and will not impact your credit score.
- If you qualify for financial assistance, you agree to provide Myriad with any additional information or documentation that may be needed to confirm your qualification for the financial assistance program.
- If your insurance carrier does not reimburse Myriad in full or in part because your insurance carrier determines the FirstGene Multiple Prenatal Screen is not a covered service, is not medically necessary, or for any other reason, you agree to be responsible for payment if you choose to proceed with the screen.

This test is available only to individuals who are at least 18 years old or the services are being performed during pregnancy. I represent and warrant that I have the right, authority and capacity to consent to testing and am at least 18 years old or am taking this test during pregnancy.

In addition, I represent and warrant that (1) all information that I have submitted or that is submitted on my behalf is complete, accurate and truthful, and (2) in the event that I have allowed a third party to assist me in providing any information, I have reviewed and confirmed that all such information is complete, accurate and truthful prior to its submission to Myriad.

**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.**

☐ **I HAVE DECIDED NOT TO PURSUE TESTING and will discuss next steps with my healthcare provider.**

| Patient Name | Date of Birth | Patient Signature | Date |
|---|---|---|---|

| Ordering Healthcare Provider Name | Ordering Healthcare Provider Signature | Date |
|---|---|---|

Myriad genetics®

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# Notice of Nondiscrimination and Notice of Availability of Language Assistance Services and Auxiliary Aids and Services

Myriad Genetics, Inc. (“Myriad”) complies with applicable federal civil rights laws and does not discriminate, exclude or treat individuals less favorably on the basis of race, color, national origin (including limited English proficiency and primary language), age, disability, or sex (including pregnancy, sex characteristics, sexual orientation, gender identity, and sex stereotypes) or any other basis prohibited by federal, state or local laws.

Myriad provides free aids and services to people with disabilities or limited English proficiency to communicate effectively with us, such as TTY/TDD calls or written information in suitable formats. Myriad provides free language services to people whose primary language is not English through qualified interpreters. These resources may be obtained by contacting the Civil Rights Coordinator or by calling Myriad’s customer service directly (or by first dialing 711 for hearing impaired individuals to be connected through a relay service operator) at 1-800-469-7423.

Myriad’s Discrimination Grievance Procedure and the process for filing a complaint can be accessed on the Company’s website at Myriad.com.

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ATTENTION: If you speak _____, free language assistance services are available to you. Appropriate auxiliary aids and services to provide information in accessible formats are also available free of charge. Call 1-800-469-7423 (or by first dialing 711 for relay services).

### Spanish

ATENCIÓN: Si habla español, tiene a su disposición servicios gratuitos de asistencia lingüística. También se encuentran disponibles de forma gratuita ayudas y servicios auxiliares adecuados para proporcionar información en formatos accesibles. Llame al 1-800-469-7423 (o marcando primero 711 para servicios de retransmisión).

### Chinese

**注意**：**如果您**讲中文，我们可以为您提供免费的语言协助服务。还免费提供适当的辅助工具和服务，以可访问的格式提供信息。致电 1-800-469-7423（**或先**拨打 711 获取转接电话服务）。

### Tagalog

PANSIN: Kung nagsasalita ka ng Tagalog, magagamit mo ang mga libreng serbisyo ng tulong sa wika. Ang naaangkop na mga pantulong na lunas at serbisyo upang magbigay ng impormasyon sa mga naa-access na format ay makukuha rin nang walang bayad. Tumawag sa 1-800-469-7423 (o sa pamamagitan ng unang pag-dial sa 711 para sa mga serbisyo ng relay).

### Vietnamese

CHÚ Ý: Nếu bạn nói tiếng Việt, chúng tôi có dịch vụ hỗ trợ ngôn ngữ miễn phí dành cho bạn. Các dịch vụ và h trợ bổ sung thích hợp để cung cấp thông tin ở các định dạng dễ tiếp cận cũng được cung cấp miễn phí. Gọi số 1-800-469-7423 (hoặc trước tiên quay số 711 để được dịch vụ chuyển tiếp).

### Arabic

تنبيه: إذا كنت تتحدث اللغة العربية، فستُتاح لك خدمات مساعدة لغوية مجانية. تتوفر أيضًا وسائل المساعدة والخدمات الإضافية المناسبة لتقديم المعلومات بأشكال مجانية سهلة الاستخدام لذوي الاحتياجات الخاصة. يمكنك الاتصال هاتفيًا بالرقم 1-800-469-7423 (أو بالاتصال برقم 711 أولاً للحصول على خدمات ترحيل الاتصالات).

### French

ATTENTION : Si vous parlez français, des services d'assistance linguistique gratuits sont à votre disposition. Des aides et services auxiliaires permettant de fournir des informations dans des formats accessibles sont également disponibles gratuitement. Appelez le 1-800-469-7423 (ou composez d’abord le 711 pour les services de relais).

### Korean

주의: 한국어를 사용하시는 경우, 무료 언어 지원 서비스를 받으실 수 있습니다. 접근 가능한 형식으로 정보를 제공하기 위한 적절한 보조 도구와 서비스도 무료로 제공됩니다. 1-800-469-7423으로 전화하세요(또는 청각 또는 언어 장애인을 위한 중계 서비스를 이용하시려면 먼저 711로 전화하세요).

### Russian

ВНИМАНИЕ: Если вы говорите по-русски, вам доступны бесплатные услуги языковой помощи. Соответствующие вспомогательные средства и услуги по предоставлению информации в доступных форматах также предоставляются бесплатно. Позвоните по телефону 1-800-469-7423 (или сначала наберите 711 для получения услуг ретрансляции).

### Portuguese

ATENÇÃO: Se você fala português, serviços gratuitos de assistência linguística estão disponíveis para você. Recursos auxiliares e serviços adequados para fornecer informações em formatos acessíveis também estão disponíveis gratuitamente. Ligue para 1-800-469-7423 (ou disque primeiro 711 para serviços de retransmissão).

### Hindi

ध्यान दे: यदि आप हिन्दी बोलते हैं तो आपके लिए निःशुल्क भाषा सहायता सेवाएं उपलब्ध हैं। सुलभ प्रारूप में सूचना उपलब्ध कराने के लिए उपयुक्त सहायक साधन और सेवाएं भी निःशुल्क उपलब्ध हैं। 1-800-469-7423पर कॉल करें (या रिले सेवाओं के लिए पहले 711 डायल करें)।

### Haitian Creole

ATANSYON: Si w pale kreyòl ayisyen, asistans sèvis yo lang yo disponib pou ou gratis . Èd ak sèvis oksilyè apwopriye pou bay enfòmasyon nan fòma aksesib yo disponib gratis tou. Rele 1-800-469-7423 (oswa konpoze anvan 711 pou sèvis relè).

### German

ACHTUNG: Wenn Sie Deutsch sprechen, stehen Ihnen kostenlose Sprachassistenzdienste zur Verfügung. Entsprechende Hilfsmittel und Dienste zur Bereitstellung von Informationen in barrierefreien Formaten stehen ebenfalls kostenfrei zur Verfügung. Rufen Sie 1-800-469-7423 an (oder wählen Sie zuerst 711 für Relay-Dienste).

### Italian

ATTENZIONE: Se parli italiano, potrai usufruire di servizi di assistenza linguistica gratuiti. Sono inoltre disponibili gratuitamente adeguati supporti e servizi ausiliari per fornire informazioni in formati accessibili. Rivolgersi al numero 1-800-469-7423 (oppure comporre prima il 711 per i servizi per non udenti).

### Polish

UWAGA: Jeśli znasz język polski, możesz skorzystać z bezpłatnych usług pomocy językowej. Odpowiednie pomoce i usługi zapewniające dostęp do informacji w przystępnych formatach są również udostępniane bezpłatnie. Zadzwoń pod numer 1-800-469-7423 (lub najpierw wybierz 711, aby skorzystać z usługi transmisji tekstu).

### Urdu

توجہ دیں: اگر آپ اردو بولتے ہیں تو، لسانی تعاون کی مفت خدمات آپ کے لیے دستیاب ہیں۔ قابل رسائی فارمیٹس میں معلومات فراہم کرنے کے لیے مناسب ضمنی آلات اور خدمات بھی بلا معاوضہ دستیاب ہیں۔ 1-800-469-7423 پر کال کریں (یا ریلے سروسز کے لیے پہلے 711 ڈائل کر کے)۔