

informaSeq Prenatal Test

Simple, safe, and accurate for you and your pregnancy.

The informaSeqSM Prenatal Test is an early and accurate test for assessing the risk of Down syndrome and other chromosome conditions. The test also offers an optional analysis for fetal sex and sex chromosome (X,Y) conditions.

What is a trisomy?

Humans have 23 pairs of chromosomes, which are strands of DNA and proteins that carry genetic information. A trisomy is a chromosomal condition that occurs when there are three copies of a particular chromosome instead of the expected two.

Trisomy 21 is due to an extra chromosome 21 and is the most common trisomy at the time of birth.

Trisomy 21, also called Down syndrome, is associated with mild to moderate intellectual disabilities and may also lead to digestive disease and congenital heart defects. It is estimated that trisomy 21 is present in 1 out of every 830 newborns.¹

Trisomy 18 is due to an extra chromosome 18. Trisomy 18, also called Edwards syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 18 often have congenital heart defects as well as various other medical conditions, shortening their lifespan. It is estimated that trisomy 18 is present in approximately 1 out of every 5,000 newborns.²

Trisomy 13 is due to an extra chromosome 13. Trisomy 13, also called Patau syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 13 usually have severe congenital heart defects and other medical conditions. Survival beyond the first year is rare. It is estimated that trisomy 13 is present in approximately 1 out of every 16,000 newborns.³

informaSeq assesses risk for trisomies of chromosomes 21, 18, and 13 in the fetus, but does not rule out all fetal abnormalities.

What are sex chromosome conditions?

The sex chromosomes (X and Y) are the ones that make us either male or female. X and Y chromosome conditions occur when there is a missing, extra, or incomplete copy of one of the sex chromosomes. You and your physician may choose the informaSeq Test options with Y or X,Y evaluation. Including Y or X,Y evaluation can assess risk for XXX, XYY, XXY (Klinefelter syndrome), and a missing X chromosome (Monosomy X) in a girl (Turner syndrome). There is significant variability in the severity of these conditions, but most individuals have mild, if any, physical or behavioral features or learning challenges.⁴

What will the informaSeq Prenatal Test tell me?

The informaSeq Prenatal Test evaluates the risk for trisomies 21, 18, and 13. Additional test options called informaSeq with Y Analysis and informaSeq with X,Y Analysis are also available. These provide information on fetal sex as well as on X and Y sex chromosome conditions. The informaSeq Prenatal Test, informaSeq with Y Analysis, and informaSeq with X,Y Analysis do not assess risk for mosaicism, partial trisomies, or translocations.



How well does the informaSeq Prenatal Test perform?

The informaSeq Prenatal Test is based on the newest advances in non-invasive prenatal testing. It is a simple and safe blood test that has been shown in clinical studies to assess the risk of fetal trisomies with high accuracy.^{5,6}

The informaSeq Test has been shown to have detection rates of up to 99% and false positive rates as low as 0.1% for trisomy 21, 18, and 13.^{5,6}

Who can get the informaSeq Prenatal Test?

The informaSeq Prenatal Test can be ordered by healthcare professionals for at-risk women with pregnancies of at least 10 weeks' gestational age. The informaSeq Prenatal Test is an early and accurate test for assessing the risk of Down syndrome and other chromosome conditions. The informaSeq Prenatal Test and informaSeq with Y analysis can be ordered for all *in vitro* fertilization (IVF) singleton and twin pregnancies. informaSeq with X,Y Analysis is not available for twin pregnancies.

The test is not intended for use in women with multiple gestations other than twins.

What do my informaSeq Prenatal Test results mean?

Decreased Risk result

If the informaSeq Prenatal Test results show **No Aneuploidy Detected**, the chance of having a baby with trisomy 21, trisomy 18, or trisomy 13 is low. As with any test, a Decreased Risk result reduces, but does not eliminate, the chance of having an affected pregnancy. A Decreased Risk result should be considered in view of other prenatal screening and/or diagnostic tests in determining the appropriate course of prenatal care.

Increased Risk result

If the informaSeq Prenatal Test results show **Aneuploidy Detected or Aneuploidy Suspected**, there is an increased chance of having a baby with trisomy 21, trisomy 18, or trisomy 13. If your result is **Increased Risk**, your healthcare provider may offer genetic counseling and/or diagnostic testing to determine if your baby is affected with one of these conditions.

Informed Consent/Decline for informaSeq Prenatal Testing

1. The purpose of the informaSeq Prenatal Test is to identify pregnancies that may be at increased risk for trisomy 21, trisomy 18, or trisomy 13.
2. The informaSeq Prenatal Test is a screening test to assess risk.
 - Not all affected fetuses can be detected: some will be missed by any testing.
 - Some women with normal fetuses will have abnormal screening results.
3. Abnormal screening results may indicate the need for further testing, such as ultrasound and/or CVS or amniocentesis.
4. The decision to consent to, or to refuse the ab testing is entirely mine.
5. No test(s) will be performed and reported on my sample other than those authorized by my doctor; and any unused portion of my original sample will be destroyed within 2 months of receipt of the sample by the laboratory.
6. My doctor may release my pregnancy outcome or ultrasound and amniocentesis results to Esoterix Genetic Laboratories, LLC to be used for statistical analysis of the laboratory's performance.
7. Esoterix Genetic Laboratories, LLC will disclose test results ONLY to the doctor named below, to his/her agent, unless otherwise authorized by the patient or required by law.

(Continued on other



Informed Consent/Decline for informaSeq Prenatal Testing

(Continued from other side)

My signature below indicates that I have read, or had read to me, the above information and I understand it. I have also read or had explained to me the specific disease(s) or conditions(s) tested for, and the specific test(s) I am having, including the test descriptions, principles, and limitations. I have had the opportunity to discuss the purposes and possible risks and limitations of this testing with my doctor or someone my doctor has designated. I know that genetic counseling is available to me before and after the testing. I have all the information I want and all my questions have been answered.

I have decided that:

I want the informaSeq Prenatal Test.*

I do not want the informaSeq Prenatal Test.

Patient Signature

Date

Obtained by

*informaSeq with Y Analysis or informaSeq with X,Y Analysis are additional test options and may be ordered for fetal sex or sex chromosome conditions.

California, Georgia, and New York have statutes requiring laboratories to send confidential results of certain genetic tests to state or federal health agencies for monitoring the detection of birth defects. It is a standard of care for physicians to obtain informed consent for genetic testing. This model consent form is designed to address the requirements of New York State Civil Rights Law Section 79-1 and Massachusetts General Law Chapter 111, Section 70G. Integrated Genetics requires that all reproductive genetic testing sent to any of our laboratories be accompanied by the signed attestation on the front of this Test Requisition Form. Relevant patient educational materials are also available through Integrated Genetics. This model informed consent form is provided by Integrated Genetics as a courtesy to physicians and their patients.

Integrated Genetics is a business unit of Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America® Holdings.

If my result suggests Increased Risk, what additional testing is available?

If an informaSeq Prenatal Test shows you are at Increased Risk, it does not necessarily mean that the pregnancy has one of these fetal abnormalities. Your healthcare provider may offer you one of the following procedures:

- **Chorionic villi sampling (CVS)** is a procedure that takes a small amount of tissue from the developing placenta. The tissue is then sent to a laboratory to test the chromosomes. CVS is typically performed between 10 and 12 weeks of pregnancy. CVS is associated with a small risk of miscarriage.
- **Amniocentesis** is a procedure that withdraws a small amount of fluid that surrounds the fetus. The fluid is then sent to the laboratory to test the chromosomes. An amniocentesis is usually performed around or after the 16th week of pregnancy. Amniocentesis is associated with a small risk of miscarriage.

informaSeq Prenatal Test does not test for open neural tube defects. Open neural tube defects occur when the baby's neural tube does not close completely and an opening remains along part of the baby's spine or head. Open neural tube defects occur in about 1 out of every 1,500 live births.⁷ A second trimester blood test called MSAFP, or an ultrasound, is required to detect open neural tube defects.

Does insurance cover the informaSeq test?

Integrated Genetics has contracts with over 400 health insurance plans. If you are a member of a plan, the informaSeq test may be covered. The best way to confirm if the informaSeq prenatal test is covered by your particular insurance is to contact your provider.

For your convenience, our billing team will file claims with your health insurance company, and we offer a variety of simple and convenient ways for you to pay any balance you may owe.

For more information about these payment plans please contact billing customer service at 800-845-6167. We are available between the hours of 8:00 AM and 5:00 PM, Monday through Friday.

www.mytestingoptions.com

informaSeqSM

non-invasive prenatal test

A simple non-invasive prenatal testing option for early risk assessment of Down syndrome and other conditions.

Ask your doctor about it today.

About Integrated Genetics

Integrated Genetics has been a leader in genetic testing and counseling services for over 25 years.

Learn more about our genetic testing and counseling services by calling our client services team at 800-848-4436 or visiting our web sites:

www.mytestingoptions.com
www.integratedgenetics.com

This brochure is provided by Integrated Genetics as an educational service for health care providers and their patients.



References

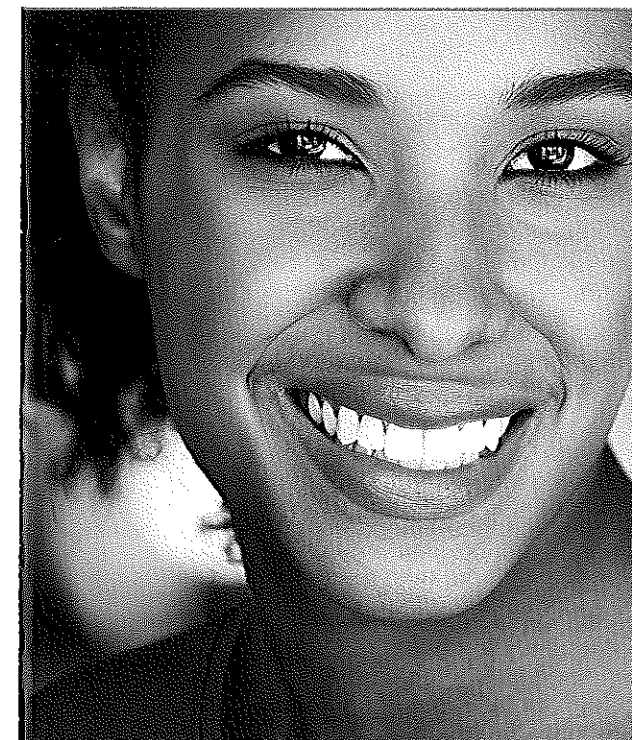
1. U.S. National Library of Medicine. Genetics Home Reference. Down Syndrome. <http://ghr.nlm.nih.gov/condition/downsyndrome>. Accessed July 1, 2014.
2. U.S. National Library of Medicine. Genetics Home Reference. Trisomy 18. <http://ghr.nlm.nih.gov/condition/trisomy-18>. Accessed July 1, 2014.
3. U.S. National Library of Medicine. Genetics Home Reference. Trisomy 13. <http://ghr.nlm.nih.gov/condition/trisomy-13>. Accessed July 1, 2014.
4. Nussbaum, Robert L. McInnes, Roderick, Huntington, Willard, Thompson & Thompson Genetics in Medicine, Sixth Edition. Saunders 2001.
5. Bianchi D, Parker, L, Wentworth, J, et al. DNA Sequencing versus Standard prenatal aneuploidy screening. *NEJM* 2014 Feb 27; 370 (9):799-808.
6. Futch T, Spinosa J, Bhatt S, de Feo E, Rava RP, Sehner AJ. Initial clinical laboratory experience in noninvasive prenatal testing for fetal aneuploidy from maternal plasma DNA samples. *Prenat Diagn*. 2013; 33:569-574.
7. U.S. Food and Drug Administration (FDA). <http://www.fda.gov/Drugs/DrugSafety/PostmarketDrugSafetyInformationforPatientsandProviders/DrugSafetyInformationforHealthcareProfessionals/ucm192649.htm>. Accessed July 1, 2014.

informaSeqSM is a service mark of Laboratory Corporation of America® Holdings. ©2014 Laboratory Corporation of America® Holdings. All rights reserved. rep-881-v1-0814 L13273-0814-1

www.integratedgenetics.com

informaSeqSM

non-invasive prenatal test



A test your patients can trust.

A simple, safe, and accurate non-invasive prenatal test for early risk assessment of Down syndrome and other conditions

