Analysis Report : PrenatalSafe® 5 - Non-Invasive Prenatal Test (NIPT)

Report date:08/07/2016 ***Time:***19:49

Referring Centre details

Referring Centre:

City:

Patient's details

Surname: ***Name:***

Date of birth: ***Place of birth:***

Ethnicity: N.A. ***Sex:*** F

Physician: ***Sample's ID:***

Indication:

Clinical details:

Sample's details

Sample Type: blood ***Our Sample's ID:*** E22746

Acceptance Date: 05/07/2016 ***Acceptance Time:*** 16:49 ***Collection Date:*** 04/07/2016

Analysis details

Analysis performed: PrenatalSafe® 5 - Non-Invasive Prenatal Test (NIPT)

Code OMIM: ***Mode of Inheritance:***

Gene investigated: ***OMIM:*** ***Reference Sequence:***

Method of Analysis: Massive Parallel Sequencing (MPS) mediante tecnica Next Generation Sequencing (NGS)

Diagnostic strategy:

Sample Processing Date: 05/07/2016 ***Analysis completed:*** 08/07/2016

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Analysis Restults

Result:

POSITIVE Test Result.
Results consistent with **TRISOMY for chromosome 18**.
No aneuploidy detected for chromosomes 21, 13 and sex chromosomes.
Fetal sex: **Male**
Fetal fraction: 2.4%

Interpretation:

This specimen showed an **increased** representation of chromosome 18 (**Trisomy 18**), suggestive of **Edwards syndrome**. Genetic counseling and clinical correlation are recommended. Confirmatory testing is required if fetal confirmation and clinical interpretation of the suspected event are desired. Please refer to the "Performance" and "Limitations of the Test" sections of the enclosed technical report for additional information.

Technical notes:

PrenatalSafe® test is designed to detect chromosome aneuploidies and is validated for chromosomes 21, 18, and 13, X and Y. The test is performed by a directed analysis of cell-free fetal DNA (cffDNA) in maternal blood by Massive Parallel Sequencing (MPS) of the whole fetal genome, using Next Generation Sequencing (NGS) technology (HiSeq 2500 NGS sequencer, Illumina). Bioinformatic analysis has been performed by SAFeR™ algorithm (Verinata Health inc., Illumina). The test is validated for singleton and twin pregnancies with gestational age of at least 10 weeks. The test is neither intended nor validated for diagnosis or for use in pregnancies with more than two fetuses, mosaicism, partial chromosomal aneuploidy, translocations, or maternal aneuploidy. These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal abnormalities, birth defects, or other complications. A negative test result does not preclude the presence of trisomy 21, trisomy 18, or trisomy 13, Monosomy X, XXX, XXY, and XYY. When an aneuploidy detected result is reported in a twin pregnancy, the status of each individual fetus cannot be determined. The presence or absence of Y chromosome material can be reported in a twin pregnancy, however, the occurrence of sex chromosome aneuploidies, such as Monosomy X, XXX, XXY, and XYY, cannot be evaluated in twin pregnancies. There is a small possibility that the test results might not reflect the chromosomes of the fetus, but may reflect the chromosomal changes of the placenta (confined placental mosaicism), or of the mother (chromosomal mosaicism). Limit of Detection (LOD) of the method: fetal fraction greater than or equal at 2% (Fiorentino et. al., 2016 Prenatal Diagnosis).

Comments:

Further action:

Results verified by: Francesca Pizzuti ***Verification date:*** 08/07/2016

Results validated by: Francesco Fiorentino ***Validation date:*** 08/07/2016

This report represents a true copy to the primary document, that is detained in the archives of Genoma Group Srl.

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Dr.ssa Marina Baldi

Genoma Group Srl

Lab Director
Dr. Francesco Fiorentino

Genoma Group Srl

Rome, 08 July 2016

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About the PrenatalSafe® prenatal test

The **PrenatalSafe®** Non-Invasive Prenatal Test (NIPT) looks at the DNA (genetic material) in your blood.

The **PrenatalSafe® 5** test can tell if there are too many or too few copies (also called an "aneuploidy") of certain chromosomes—21, 18, and 13—present in your fetus. The test can also look at sex chromosomes (X and Y), and can determine if there are too many or too few copies of the sex chromosomes. This test is available for singleton and twin pregnancies.

The **PrenatalSafe® Plus** test can also test for trisomies (too many copies) of chromosomes 9 and 16, as well as microdeletions of certain chromosomes, which are listed below.

The **PrenatalSafe® Karyo** test analyzes every chromosome in the genome, providing karyotype-level insight. Though not a fetal karyotype, it offers a level of information previously only available from a karyotype analysis. It provides information about gains or losses of chromosome material **e 10 Mb** across the genome.

The **PrenatalSafe® Karyo Plus** test can also test for 9 microdeletion syndromes, which are listed below. It provides information about gains or losses of chromosome material **e 7 Mb** across the genome.

The **PrenatalSafe®** prenatal test is performed on a maternal blood sample which contains DNA (genetic material) from both the mother and fetus. It is available for women who are at least 10 weeks pregnant. This screening test can detect over 99% of the abnormalities evaluated for chromosomes 21, 18 and 13 and about 95% of cases of Monosomy X (see list below).

The **PrenatalSafe®** prenatal test has been studied in patients who have an increased risk for having a baby with an incorrect change in the number of certain chromosomes.

Your health care provider will determine if this test is appropriate for you and can provide you with more details about the chromosome abnormalities being evaluated.

Chromosome abnormalities evaluated with PrenatalSafe®:

Trisomy 21	This is caused by an extra copy of chromosome 21 and is also called Down syndrome. This is the most common genetic cause of intellectual disability. Individuals with Down syndrome have an average IQ of 50 and all have some degree of intellectual disability. Some children with Down syndrome have defects of the heart or other organs that may require surgery or medical treatment. Some have other medical conditions including hearing or vision loss.
Trisomy 18	This is caused by an extra copy of chromosome 18 and is also called Edwards syndrome. This causes severe intellectual disability. Most babies with Trisomy 18 have multiple severe birth defects of the brain, heart and other organs. Poor growth during pregnancy is common and many babies are miscarried or stillborn. Of those babies born alive, most die before one year of age. Babies who survive have profound intellectual disabilities and growth and development problems.
Trisomy 13	This is caused by an extra copy of chromosome 13 and is also called Patau syndrome. This causes severe intellectual disability. Most babies with trisomy 13 have multiple severe birth defects of the brain and other organs. Many babies are miscarried or stillborn. Of those babies born alive, most die before one year of age.
Sex Chromosome Aneuploidies	The PrenatalSafe® prenatal test also gives your healthcare provider the option to test for changes in the number of sex chromosomes. Sex chromosome aneuploidies are conditions in which there is a change from the usual 2 copies of sex chromosomes in males (XY) or females (XX). About 1 in 400 babies that are born will have a sex chromosome aneuploidy. The most common sex chromosome aneuploidies are caused by a missing sex chromosome in girls (45,X or monosomy X, also called Turner syndrome) or an extra chromosome in boys or girls (47,XXY (Klinefelter syndrome), 47,XYY, or 47,XXX). Children with a sex chromosome aneuploidy can have difficulties with language skills, motor skills, and learning, but can lead healthy and productive lives.

(Arthur Robinson & Mary G Linden, 1993, Clinical Genetics Handbook, Second Edition. Cambridge, Mass, Blackwell Scientific Publications)

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Microdeletion syndromes and trisomies 9 and 16

All pregnancies have a risk for being affected with a chromosome disorder, whether a microdeletion or a trisomy. Collectively, microdeletion syndromes are common, affecting approximately 1 in 1,000 pregnancies, and have clinical features that can affect growth, intellectual ability, and development. Trisomy 9 or 16 often result in a first-trimester miscarriage. These microdeletion syndromes and trisomies usually occur spontaneously without any family history.

Trisomies 9 and 16

Trisomy 9: A rare chromosomal condition with the vast majority of instances resulting in miscarriage in the 1st trimester. While the majority of live births will not survive during early postnatal period, those that do will have serious health concerns, including intellectual disability and cardiac defects. It can also occur in mosaic form;

Trisomy 16: The most commonly occurring autosomal trisomy seen in first trimester miscarriages. Rare survivors with mosaic trisomy 16 are at increased risk for health concerns including intra-uterine growth restriction, intellectual disability, and cardiac defects. There is a small increased risk for a woman to have a pregnancy with a viable trisomy following a miscarriage with trisomies 9 or 16. The ability to identify these important chromosomal causes of miscarriage can help with risk assessment as well as monitoring and management of subsequent pregnancies.

GENOME-WIDE COPY NUMBER VARIANTS:

e 10 Mb At every chromosome in the genome, with **PrenatalSafe® Karyo** test.

e 7 Mb At every chromosome in the genome, with **PrenatalSafe® Karyo Plus** test.

Microdeletion syndromes

Microdeletions are chromosomal disorders caused by small missing pieces of chromosome material. They are usually not visible by standard methods of chromosome analysis. Microdeletions can occur on any of the 23 pairs of chromosomes. Some occur more commonly in a specific area of a particular chromosome and have been linked to known genetic syndromes. Most occur by chance, rather than being inherited from a parent, and can occur with no prior family history and without other risk factors, such as advanced parental age. Results from routine pregnancy screening are usually normal.

Many microdeletion syndromes can cause serious health issues including both physical and intellectual impairment—the severity of which can vary from individual to individual. These conditions usually cannot be detected by traditional serum screening and may or may not be associated with ultrasound abnormalities. Until now, an invasive procedure, such as chorionic villus sampling (CVS) or amniocentesis, was the primary way to detect such conditions prenatally.

Routine prenatal serum screens cannot assess microdeletion syndromes. Additionally, microdeletion syndromes may not have abnormal ultrasound findings. Early information would aid patients and physicians greatly in pregnancy and newborn care. These expansions to the PrenatalSafe® test, the microdeletion panel and the test for trisomies 9 and 16, provide patients and physicians with additional non-invasive prenatal testing (NIPT) options based on clinical context.

The microdeletion panel and trisomies 9 and 16 testing, offered as options to the **PrenatalSafe® Plus** and **PrenatalSafe® Karyo Plus** prenatal test, use the same proven whole-genome massively parallel sequencing

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technology as the original test. The microdeletion panel covers **6** (**PrenatalSafe® Plus**) and **9** (**PrenatalSafe® Karyo Plus**) of the more commonly seen and clinically relevant microdeletion regions:

Test		Microdeletion Syndrome	Incidence	Clinical Features (may include but not limited to)	Life Expectancy
PrenatalSafe® Karyo Plus	PrenatalSafe® Plus	**22q11.2 syndrome** (DiGeorge syndrome, Velocardialfacial syndrome)	1 in 4,000	Learning problems, congenital heart defects, palatal abnormalities	Usually normal, can be reduced for DiGeorge syndrome
		1p36 deletion syndrome	1 in 4,000 to 1 in 10,000	Characteristic craniofacial features, intellectual disability, seizures, brain and heart defects	Depends on the severity of features, but can be normal
		Angelman syndrome (15q11.2 deletion syndrome)	1 in 12,000	Intellectual disability, speech impairment, seizures	Normal
		Prader-Willi syndrome (15q11.2 deletion syndrome)	1 in 10,000 to 1 in 25,000	Hypotonia, morbid obesity, delayed motor and language skills, intellectual disability, hypogonadism	Normal, may be reduced depending on the severity of symptoms
		Cri du Chat syndrome (5p- deletion syndrome)	1 in 20,000 to 1 in 50,000	Intellectual disability, speech delay, cat-like cry	10% mortality in the first year; otherwise usually normal, but will depend on the severity of features
		Wolf-Hirschhorn syndrome (4p- deletion syndrome)	1 in 50,000	Growth deficiency, hypotonia, craniofacial features, intellectual disability, heart and brain abnormalities	Depends on severity of features
		Langer-Giedion syndrome (8q24-syndrome)	1/200.000	Benign bone tumors (exostoses), short stature, and distinctive facial features.	Normal
		Jacobsen syndrome (11q23-syndrome)	1/100.000	Developmental delay, distinctive facies, bleeding disorders and some behavior disorders.	20% of children die during the first two years of life
		Smith-Magenis syndrome (17p11.2-syndrome)	1/15.000 - 1/25.000	variable intellectual deficit, sleep disturbance, craniofacial and skeletal anomalies, psychiatric disorders, and speech and motor delay	Data on life expectancy are currently insufficient but patients have lived to more than 80 years of age

Individually, microdeletion syndromes are rare, with a low prevalence in the general population. False positive NIPT results may lead to unnecessary invasive testing. To this end, Genoma understands that not everyone is an appropriate candidate for additional microdeletion testing as part of their pregnancy care. For this reason Genoma provides this testing as an elective option. This test should be used in the context of the patient's history, including information about family history and pregnancy information such as an abnormal ultrasound.

The Testing Process

PrenatalSafe® test analyzes the relative amount of chromosomal material across the genome in circulating cell-free DNA from a maternal blood sample.

Circulating cell-free DNA is purified from the plasma component of anti-coagulated maternal whole blood. A genomic DNA library is prepared to determine chromosomal representation using a technology called "massively parallel sequencing or MPS". A specific algorithm counts the number of copies of these chromosomes, and then uses a calculation method to determine if there are too many or too few copies of these chromosomes present in your fetus.

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Gain or loss of chromosomal material e7 Mb and e10 Mb are evaluated across the entire genome for **PrenatalSafe® Karyo** and **PrenatalSafe® Karyo Plus** test, respectively. Select chromosomal regions (1p, 4p, 5p, 8q, 11q, 15q, and 22q, for **PrenatalSafe® Plus** test, and 1p, 4p, 5p, 8q, 11q, 15q, 22q, 8q, 17p and 11q, for **PrenatalSafe® Karyo Plus** test) associated with known microdeletion syndromes are also evaluated. Fetal sex is assessed by Y chromosome representation.
The test is performed after 10 weeks, 0 days of pregnancy. Adequate DNA in the blood sample is required to complete the test. Additional samples may be needed if the sample is damaged in shipment or incorrectly submitted, or if a test repetition is needed.

Obtaining and Interpreting Test Results
Your results will tell your healthcare provider whether too few or too many copies of the chromosomes being tested for are present. If a genetic disorder is detected, follow up testing (such as amniocentesis or chorionic villus sampling) is recommended to confirm the result.

Your test report will include one of three possible results for chromosomes 21, 18, and 13: No Aneuploidy Detected, Aneuploidy Detected, or Aneuploidy Suspected (Borderline Value). Sex chromosomes will be reported as No Aneuploidy Detected, or Aneuploidy Detected, or XX or XY, as appropriate. In the case of a twin pregnancy, Y chromosome presence will be reported as Detected or Not Detected.

A **No Aneuploidy Detected** test result means that this test identified the expected number of copies of chromosomes reported.

An **Aneuploidy Detected** test result means that this test identified too many or too few copies of one of the chromosomes as seen on the report. This can indicate either a trisomy or a sex chromosome aneuploidy

An **Aneuploidy Suspected** test result means that this test identified more copies than expected of the chromosomes reported. This means that your provider should follow up on this result to obtain more information.

In the case of microdeletions testing, negative results will be classified as "**No abnormality detected**" and positive results classified as "**abnormality detected**" with additional comment indicating that interpretation is consistent with a loss in the genomic region that is associated with a particular syndrome.
There is also a chance that the sample submitted will not return any results; in this case a second sample may be requested to repeat the test.

Genetic counseling before and after testing is recommended. Results of "Aneuploidy Detected" or "Aneuploidy Suspected" are considered positive and patients should be offered invasive prenatal procedures for confirmation. Pregnancy management decisions, including termination of the pregnancy, should not be based on the results of this test alone.

A negative test does not ensure an unaffected pregnancy. Chorionic villus sampling and amniocentesis provide definitive diagnostic information, but may pose harm to the fetus.

Test Performance
The PrenatalSafe® prenatal test was tested in a multi-center clinical study, in a population of high risk patients, and the test performance is indicated in the table below.
***Performance* PrenatalSAFE® (Standard Protocol): follow-up March 2016**

	Trisomy 21	Trisomy 18	Trisomy 13	Monosomy X	SCA

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	(n=31.800)	(n=31.800)	(n=31.800)	(n=31.800)	(n=31.800)
True Positive	257	47	32	77	160
False Positive	6	6	7	48	58
True Negative	31536	31746	31746	31675	31582
False Negative	1	1	0	0	0
Sensitivity (95% CI)	99,61% (97.86% - 99.99%)	97,92% (88.93% - 99.95%)	100,00% (89.11% - 100.00%)	100,00% (95.32% - 100.00%)	100,00% (99.72% - 100.00%)
☐ % False Negative	0,39%	2,08%	0,00%	0,00%	0,00%
Specificity (95% CI)	99,98% (99.96% - 99.99%)	99,98% (99.96% - 99.99%)	99,98% (99.95% - 99.99%)	99,85% (99.80% - 99.89%)	99,82% (99.76% - 99.86%)
☐ % False Positive	0,02%	0,02%	0,02%	0,15%	0,18%
PPV (95% CI)	97,72% (95.10% - 99.16%)	88,68% (76.97% - 95.73%)	82,05% (66.47% - 92.46%)	61,60% (52.48% - 70.16%)	73,39% (67.01% - 79.13%)
NPV (95% CI)	100,00% (99.98% - 100.00%)	100,00% (99.98% - 100.00%)	100,00% (99.99% - 100.00%)	100,00% (99.99% - 100.00%)	100,00% (99.99% - 100.00%)

Performance **PrenatalSAFE® (FAST Protocol): follow-up March 2016**

	Trisomy 21 (n=15.258)	Trisomy 18 (n=15.258)	Trisomy 13 (n=15.258)	Monosomy X (n=15.258)	SCA (n=15.258)
True Positive	156	30	20	57	91
False Positive	2	1	1	18	22
True Negative	15100	15227	15279	15183	15145
False Negative	0	0	0	0	0
Sensitivity (95% CI)	100,00% (88.43% - 100.00%)	100,00% (88.43% - 100.00%)	100,00% (83.16% - 100.00%)	100,00% (93.73% - 100.00%)	100,00% (96.03% - 100.00%)
☐ % False Negative	0,0%	0,0%	0,0%	0,0%	0,0%
Specificity (95% CI)	99,99% (99.96% - 100.00%)	99,99% (99.96% - 100.00%)	99,99% (99.96% - 100.00%)	99,88% (99.81% - 99.93%)	99,85% (99.78% - 99.91%)
☐ % False Positive	0,01%	0,01%	0,01%	0,12%	0,15%
PPV (95% CI)	98,73% (83.30% - 99.92%)	96,77% (83.30% - 99.92%)	95,24% (76.18% - 99.88%)	76,00% (64.75% - 85.11%)	80,53% (72.02% - 87.38%)
NPV (95% CI)	100,00% (99.98% - 100.00%)	100,00% (99.98% - 100.00%)	100,00% (99.98% - 100.00%)	100,00% (99.98% - 100.00%)	100,00% (99.98% - 100.00%)

PPV: Positive Predictive Value; NPV: Negative Predictive Value; SCA: Sex Chromosomes Aneuploidy.

Performance **PrenatalSAFE® Karyo: pre-clinical validation data**

	Trisomy 21 (n=1419)	Trisomy 18 (n=1419)	Trisomy 13 (n=1419)	SCA (n=1419)	CNV (n=1419)
True Positive	100	31	14	36	37
False Positive	0	0	0	0	0
True Negative	1319	1388	1405	1383	1382
False Negative	0	0	0	0	0
Sensitivity (95% CI)	100,00% (96.38% - 100.00%)	100,00% (88.78% - 100.00%)	100,00% (76.84% - 100.00%)	100,00% (90.26% to 100.00%)	100,00% (90.51% to 100.00%)
Specificity (95% CI)	100,00% (99.72% - 100.00%)	100,00% (99.73% - 100.00%)	100,00% (99.74% - 100.00%)	100,00% (99.73% to 100.00%)	100,00% (99.73% to 100.00%)
PPV (95% CI)	100,00% (96.38% - 100.00%)	100,00% (88.78% - 100.00%)	100,00% (76.84% - 100.00%)	100,00% (90.26% to 100.00%)	100,00% (90.51% to 100.00%)
NPV (95% CI)	100,00% (99.72% - 100.00%)	100,00% (99.73% - 100.00%)	100,00% (99.74% - 100.00%)	100,00% (99.73% to 100.00%)	100,00% (99.73% to 100.00%)

PPV: Positive Predictive Value; NPV: Negative Predictive Value; SCA: Sex Chromosomes Aneuploidy. CNV: Copy Number Variation (Fiorentino et al., ESHG conference 2016; ISPD Conference 2016)

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Limitations of the test

While the results of the PrenatalSafe® test are highly accurate, discordant results, including inaccurate fetal sex prediction, may occur. Cell-free DNA (cfDNA) testing does not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis.

The **PrenatalSafe®** prenatal test does not test for all health problems. Normal results do not eliminate the possibility that your pregnancy may have other chromosomal/genetic conditions, birth defects, or other complications. A '**No Aneuploidy Detected**' result greatly reduces the chances that your fetus has an extra or missing copy of one of the tested chromosomes but it cannot guarantee normal chromosomes or a healthy baby. The result of this test does not eliminate the possibility of other abnormalities of the tested chromosomes, and it does not detect abnormalities of untested chromosomes, other genetic disorders, birth defects, or other complications in your fetus or pregnancy.

The **PrenatalSafe® 3** and **5** prenatal test are designed to look at full chromosome aneuploidies only, and has been validated for chromosomes 21, 18, 13 and chromosomes 21, 18, 13, X and Y only, respectively.

The **PrenatalSafe® Karyo** analyzes every chromosome in the genome, providing karyotype-level insight. It provides information about gains or losses of chromosome material e 10 Mb across the genome.

The **PrenatalSafe® Karyo Plus** test can also test for 9 microdeletion syndromes. It provides information about gains or losses of chromosome material **e 7 Mb** across the genome.

A patient with a **positive** PrenatalSafe® test result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results.

An **uninformative result** may be reported, the causes of which may include, but are not limited to, insufficient sequencing coverage, noise or artifacts in the region, amplification or sequencing bias, or insufficient fetal fraction.

There is a small possibility that the test results might not reflect the chromosomes of the baby, but instead might reflect chromosomal changes to the placenta (confined placental mosaicism), or in the mother (chromosomal mosaicism).

Inaccurate test results or a failure to obtain test results may occur due to one or more of the following rare occurrences: biological factors such as but not limited to too little DNA from the fetus in the maternal blood sample, placental, maternal, or fetal mosaicism (a mixture of cells with normal and abnormal chromosomes) or neoplasm; vanishing twin; prior maternal organ transplant; or an unrecognized twin pregnancy; other circumstances beyond our control; or unforeseen problems that may arise, or other causes.

The PrenatalSafe® test is not intended to identify pregnancies at risk for neural tube defects or ventral wall defects. cfDNA testing for whole chromosome abnormalities (including sex chromosomes) and for subchromosomal abnormalities could lead to the potential discovery of both fetal and maternal genomic

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abnormalities that could have minor, or no, clinical significance. Evaluating the significance of a positive or a non-reportable test result may involve both invasive prenatal testing and additional studies on the mother. Such investigations may lead to detection of maternal chromosomal or subchromosomal abnormalities, which on occasion may be associated with benign or malignant maternal neoplasms. cfDNA testing may not accurately identify fetal triploidy, balanced rearrangements, or the precise location of subchromosomal duplications or deletions; these may be detected by prenatal diagnosis with CVS or amniocentesis. The ability to report results may be impacted by maternal body mass index (BMI), maternal weight, and/or maternal systemic lupus erythematosus (SLE).

Microdeletions testing: limitations of the Test

This test is designed to detect subchromosomal deletions and is validated for common deletions in chromosomal regions 15q11.2, 5p15.2, 22q11.2, 1p36, 4p16.3, 8q24, 17p11.2 and 11q23. The test is validated for singleton pregnancies with gestational age of at least 10 weeks as estimated by last menstrual period. These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or subchromosomal abnormalities, birth defects, and other conditions. This test is not intended to identify pregnancies at risk for open neural tube defects. A negative test result does not eliminate the possibility of Angelman syndrome, Prader-Willi syndrome, 5p-/Cri-du-Chat syndrome, 22q11.2 deletion syndrome, 1p36 deletion syndrome, or 4p-/Wolf-Hirschhorn syndrome, 8q24/Langer-Giedion syndrome, 11q23/Jacobsen syndrome and 17p11.2/Smith-Magenis syndrome.
. In addition, conditions caused by other molecular mechanisms cannot be detected with this assay. There is a small possibility that the test results might not reflect the chromosome status of the fetus, but may reflect subchromosomal changes of the placenta (confined placental mosaicism), or of the mother.

Alternatives

This non-invasive prenatal screening test is only one option for detecting pregnancies at high risk for fetal chromosome abnormalities. There are multiple other screening options available during pregnancy and, if you want more details on your other options, you should ask your health care provider. You also have the option to decline all chromosome screening tests during your pregnancy. For women who want or need more conclusive information about the fetal chromosomes, commonly used invasive diagnostic tests such as CVS or amniocentesis are available and will detect >99% of all chromosome abnormalities, including rare abnormalities on chromosomes not evaluated with this or other screening tests.

Genetic Counseling:

If you have remaining questions about non-invasive prenatal testing after talking with your health care provider, we recommend that you make an appointment with a local genetic counselor who can give you more information about your testing options.

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