1527258, Centro de Medicina Laboratorial

GERMANO DE SOUSA
CENTRO DE MEDICINA LABORATORIAL
O LABORATÓRIO DE PORTUGAL

harmony
PRENATAL TEST

Noninvasive prenatal test for trisomies 21, 18 e 13
Chrom. X and Y aneuploidies and Microdeletion 22q11.2

Patients Name (first and last name)

Date of Birth (MM/DD/YY)

Mobile number / e-mail address

Clinical information at blood collection date:

Blood collection date: ☐☐.☐☐.☐☐ Time: ☐☐:☐☐

Gestational age (weeks/days): ☐☐+☐ (acording to ultrasound; minimum 10w+0d)

☐ singleton ☐ twins max. 2 fetus [1]

[1] *In case of vanishing twin the Harmony® test cannot be performed.*

☐ If in vitro fertilization: (If spontaneos conception do not fill up)

☐ Self /Own eggs ☐ Egg donation

Patients age (in case of own eggs) or donnor age at the time of egg collection ☐☐ years

Weigth: ☐☐☐,☐ kg Hight: ☐☐☐ cm

LMP or EDD: ☐☐.☐☐.☐☐

Anomalies identified by US:

Harmony® test type requested

☐ Trisomies 21, 18 e 13

☐ Trisomies 21, 18 e 13 + Sex Chromosome Aneuploidy Panel [2]

☐ Trisomies 21, 18 e 13 + Monosomy X [2]

☐ Trisomies 21, 18 e 13 + Microdeletion 22q11.2 [2]

☐ + Fetal sex determination

[2] only singletons

Sample Barcode label:

☐ redraw

Ordering clinicians statement :

I confirm that my patient was totaly informed of the capabilities and limitations and potential risks of the test. The patient gave total consent to perform the test.

Ordering clinicians label:

Ordering clinicians name in simple text:

Place, date

X

Ordering clinicians signature

Autorization to perform Harmony® test

My signature on this form indicates that I read or was read to me, the informed consent on the back of this form. I understand and give my autorization to Grupo Germano de Sousa to perform the test requested. I had the opportunity to ask questions and discuss the capabilities and limitations of the test. I also know that I can obtain genetic counsealling before and after the test.I understand the limitations of the Harmony test indicated overleaf and take responsibility for carrying out a more comprehensive test than that indicated by my doctor.
I agree that my personal data, included in this form, and my sample can be send to a certified laboratory, so the Harmony test can be performed.
I authorize the sending of the results to the doctor indicated by me

I agree with the storage and utilization anonimyzed of my blood to quality control and investigations use.
☐ Yes ☐ No

Place, date

X

Patient signature

Results phisician mobile number or e-mail address

Informações sobre o teste pré-natal Harmony®

The Harmony test is a screening test performed to for Trisomie 21, 18 and 13, in fetuses with 10 weeks of gestation or more.

The Harmony test is a screening test and should not be considered or used as diagnostic test. Clinical studies show high performance rates for the Harmony test, however, not all fetuses afected with trisomies will be detected.

Some fetuses with trisomies could receive a "LOW RISK". Some euploide fetuses can receive a "HIGH RISK RESULT".

The results should be presented and interpreted in the correct clinical context for adequated counseling.

Exceptionally, the harmony test may not provide any results.

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Patient Informed Consent

The Harmony Prenatal Test is a laboratory-developed screening test that analyzes cell-free DNA (cfDNA) in maternal blood. The test provides a probability assessment, not a diagnosis, of fetal chromosomal or genetic conditions, and fetal sex determination. Consider Harmony results in the context of other clinical criteria. Follow up confirmatory testing based on Harmony results for Trisomy 21, 18, 13, sex chromosome aneuploidy, or 22q11.2 could reveal maternal chromosomal or genetic conditions in some cases. Results from the Harmony Prenatal Test should be communicated in a setting designated by your healthcare provider that includes the availability of appropriate genetic counseling.

Who is eligible for the Harmony Prenatal Test?

Women who are at least ten weeks pregnant are eligible for the Harmony Prenatal Test offerings. Patients with a twin pregnancy are not eligible for sex chromosome aneuploidy or 22q11.2 options. The Harmony Prenatal Test is not for patients with:

- a history of or active malignancy
- a pregnancy with fetal demise
- a pregnancy with more than two fetuses
- a history of bone marrow or organ transplants

What are the limitations of the Harmony Prenatal Test for Trisomies 21, 18, and 13, sex chromosome aneuploidy, and fetal sex determination?

The Harmony Prenatal Test is not validated for use in pregnancies with more than two fetuses, fetal demise, mosaicism, partial chromosome aneuploidy, translocations, maternal aneuploidy, transplant, malignancy, or in women under the age of 18. Harmony does not detect neural tube defects. Certain rare biological conditions may also affect the accuracy of the test. For twin pregnancies, HIGH PROBABILITY test results apply to at least one fetus; male test results apply to one or both fetuses; female test results apply to both fetuses. Due to the limitations of the test, inaccurate results are possible. A LOW PROBABILITY result does not guarantee that a fetus is unaffected by a chromosomal or genetic condition. Some non-aneuploid fetuses may have HIGH PROBABILITY results. In cases of HIGH PROBABILITY results and/or other clinical indications of a chromosomal condition, confirmatory testing is necessary for diagnosis.

What are the limitations of the Harmony Prenatal Test for 22q11.2?

In addition to the limitations discussed above, the 22q11.2 option is not validated for use in pregnancies with more than one fetus or for women with a 22q11.2 duplication or deletion. A 22q11.2 deletion may not be detected in all fetuses. Due to the limitations of the test, a NO EVIDENCE OF A DELETION OBSERVED result does not guarantee result that a fetus is unaffected by a chromosomal or genetic condition . Some fetuses with a 22q11.2 deletion may receive a test result of NO EVIDENCE OF A DELETION OBSERVED. Some fetuses without the 22q11.2 deletion may receive a test result of HIGH PROBABILITY OF A DELETION. In cases of HIGH PROBABILITY results and/or other clinical indications of a chromosomal condition, confirmatory testing is necessary for diagnosis (amniocentesis and chorionic villus biopsy).

Data protection and confidenciality and contact details

Only authorized personal of Centro de Medicina Laboratorial Germano de Sousa, or other partners laboratories authorized, will be able to access the personal information or results in a extrictly confidential way, acordinly with the law regulations in practice. The Centro de Medicina Laboratorial Germano de Sousa garantee that the information transfer will occurs as specified in the Regulament (UE) 2016/679. The pacient have the rigth to retify, suppress or limit the access. Please use the following contacts: Centro de Medicina Laboratorial Germano de Sousa, Lda, Polo tecnológico de Lisboa, Rua Cupertino de Miranda, 9 - lote 8, 1600-513, Lisboa, or by e-mail prenatal@germanodesousa.com.

Test description

The Harmony Prenatal TestÈ measures the relative proportion of chromosomes to aid in the assessment of fetal trisomies 21, 18, and 13. HarmonyÈ performs a directed analysis of cell-free DNA (cfDNA) in maternal blood and incorporates the fetal fraction of cfDNA in test results. Test results also incorporate maternal
age (or egg donor age) and gestational age related probability based on information provided on the test requisition form. Probability of less than 1% is defined as low probability and 1% or greater is defined as high probability. Harmony has been validated in singleton and twin pregnancies of at least 10 weeks gestational age. Harmony is not validated for use in pregnancies with more than two fetuses, demised twin, mosaicism, partial chromosome aneuploidy, translocations, maternal aneuploidy, transplant, malignancy, or in women under the age of 18. Harmony does not detect neural tube defects. Twin results reflect the probability that the pregnancy involves at least one affected fetus. Analysis of cfDNA does not always correlate with fetal genotype. Not all aneuploid fetuses will have a high probability result and some euploid fetuses will have a high probability result. The Harmony Prenatal Test is not a diagnostic test and results should be considered with other clinical criteria and communicated in a setting that includes appropriate counseling.
Fetal Sex test quantifies the Y chromosome. A ñfemaleò result indicates absence of Y chromosome and a ñmaleò result indicates presence of Y chromosome. It does not exclude sex chromosome aneuploidy. For twin pregnancies, a male result indicates one or two male fetuses. Sex Chromosome Aneuploidy (SCA) Panel measures proportions of the X and Y chromosomes. Sex chromosome conditions (Monosomy X, XXY, XYY, XXX, XXYY) are reported at probabilities of 1% or greater. An XYY or XXYY result indicates two or more fetal Y chromosomes. Sex Chromosome Aneuploidy Panel has only been validated in singleton pregnancies. 22q11.2 test uses targeted analysis of chromosome cfDNA fragments from within a 3Mb region of 22q11.21 to determine the probability of a deletion. ñHigh probability of a deletionò indicates that the analysis detected a decrease of cfDNA fragments consistent with a deletion in the 22q11.21 region, which may be fetal, maternal or both. ñNo evidence of a deletion observedò indicates the analysis does not find an increased probability for a deletion in the 22q11.21 region. Not all fetuses with 22q11.2 deletions will be classified as high probability. This test does not rule out the possibility of other clinically significant aneuploidy, single gene conditions, microdeletions or microduplications being present in the fetus. Women with a known 22q11.2 deletion are not eligible for this test. 22q11.2 test has only been validated in singleton pregnancies.

Clinical Data

	Bcrcargm1 P_rc	D_jqc Nmqgrgtc Rate
T21	>99% (IC de 95%: 97,9-99,8%)	(IC de 95%: 0,02-0,08%)
T18	97,4% (IC de 95%: 93,4-99,0%)	(IC de 95%: 0,01-0,05%)
T13	93,8% (IC de 95%: 79,9-98,3%)	(IC de 95%: 0,01-0,06%)

Detection and false positive (discordant result) rates based on probability cut-off of 1/100 (1%). Because these conditions are rare, limited numbers of aneuploidy twin and egg donor pregnancies have been evaluated. The negative predictive value for trisomy 21, 18, and 13 is greater than 99%. Positive predictive value (PPV) varies by prevalence. The probability result reported is not equivalent to the PPV. For more information regarding PPV refer to: www.harmonytest.com/PPV

Fetal Sex >99% accuracy for male or female sex (95% CI: 99.2-100%)
SCA Panel SCA Panel provides probability for non-mosaic fetal sex chromosome aneuploidies. Test performance varies by condition. Limited numbers of sex chromosome aneuploidy cases have been evaluated to date.
22q11.2 Limited numbers of 22q11.2 cases have been evaluated to date.