harmony
PRENATAL TEST

Harmony PRENATAL TEST Sample Reports:

Ariosa Diagnostics Clinical Lab Report

Clear **ANSWERS**
to Questions that Matter

CLIA

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Patient and Provider Information

Ariosa Diagnostics, Inc.
5945 Optical Court
San Jose, CA 95138

Questions:

sjc.clientservices@roche.com

US: (855) 927-4627

Intl: +1 (925) 854-6246

PATIENT NAME:	Jane Doe
DATE OF BIRTH: (MM/DD/YYYY)	01/01/1980
MRN:	1234567890123456789
LABORATORY ID:	AD12345678-PAT
OTHER ID:	
GESTATIONAL AGE:	10 wks 5 days
# OF FETUSES:	1
IVF STATUS:	non-IVF pregnancy
COLLECTION DATE (MM/DD/YYYY) :	07/20/2015
RECEIVED DATE (MM/DD/YYYY) :	07/21/2015
ACCOUNT #:	7654321
CLINIC NAME:	The Clinic Offering Test
REFERRING/ORDERING CLINICIAN:	Ordering Physician MD
REFERRING/ORDERING CLINICIAN FAX #:	123-456-7890
OTHER CLINICIAN:	Genetic Counselor MA, CGC
OTHER CLINICIAN FAX #:	987-654-3210
REPORT DATE: (MM/DD/YYYY)	07/28/2015

Test Results

Fetal cfDNA Percentage: 10.5%

CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 18 (T18)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient

TEST DESCRIPTION

The Harmony Prenatal Test® measures the relative proportion of chromosomes to aid in the assessment of fetal trisomies 21, 18, and 13. Harmony® 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.

CLINICAL DATA

	Detection Rate	False Positive Rate
T21	> 99% (95% CI: 97.9-99.8%)	< 0.1% (95% CI: 0.02-0.08%)
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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: http://www.harmonytest.com/PPV

REFERENCES: Stokowski R et al. Prenatal Diagnosis 2015; 35, 1-4; Data on file.

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HIGH PROBABILITY RESULT

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Contact Line 1^
Contact Line 2^
Contact Line 3^
Contact Line 4^
Contact Line 5^
Contact Line 6^
Contact Line 7^
Contact Line 8^

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Test Results

Fetal cfDNA Percentage: 10.5%

CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	**High Probability**	**Greater than 99/100 (99%)**	**Genetic counseling and additional testing**
Trisomy 18 (T18)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient

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Fetal Sex **Male**

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Fetal Sex test quantifies the Y chromosome. A "female" result indicates absence of Y chromosome and a "male" 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.

CLINICAL DATA

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Fetal Sex: > 99% accuracy for male or female sex (95% CI: 99.2-100%)

REFERENCES: Stokowski R et al. Prenatal Diagnosis 2015; 35, 1-4; Data on file.

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Monosomy X

Monosomy X greater than 99% probability - Recommend genetic counseling

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Monosomy X test quantifies the X chromosome. Monosomy X is reported at a probability of 1% or greater. It does not exclude other sex chromosome aneuploidies. Monosomy X has only been validated in singleton pregnancies.

CLINICAL DATA

	Detection Rate	False Positive Rate
T21	> 99% (95% CI: 97.9-99.8%)	< 0.1% (95% CI: 0.02-0.08%)
T18	97.4% (95% CI: 93.4-99.0%)	< 0.1% (95% CI: 0.01-0.05%)
T13	93.8% (95% CI: 79.9-98.3%)	< 0.1% (95% CI: 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: http://www.harmonytest.com/PPV

Monosomy X: Monosomy X provides probability for non-mosaic Monosomy X. Limited numbers of Monosomy X cases have been evaluated to date.

REFERENCES: Stokowski R et al. Prenatal Diagnosis 2015; 35, 1-4; Data on file.

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SCAP, Low Probability

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MRN:	1234567890123456789
LABORATORY ID:	AD12345678-PAT
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Sex Chromosome Aneuploidy Panel — Low Probability

TEST DESCRIPTION

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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.

CLINICAL DATA

	Detection Rate	False Positive Rate
T21	> 99% (95% CI: 97.9-99.8%)	< 0.1% (95% CI: 0.02-0.08%)
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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.

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SCAP Low Probability, Fetal Sex

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Fetal Sex	**Female**
Sex Chromosome Aneuploidy Panel	**Low Probability**

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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.

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Fetal Sex: > 99% accuracy for male or female sex (95% CI: 99.2-100%)

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Fetal Sex	**Female**
Sex Chromosome Aneuploidy Panel	**Monosomy X greater than 99% probability - Recommend genetic counseling**

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T18	97.4% (95% CI: 93.4-99.0%)	< 0.1% (95% CI: 0.01-0.05%)
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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: http://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.

REFERENCES: Stokowski R et al. Prenatal Diagnosis 2015; 35, 1-4; Data on file.

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Patient and Provider Information

Ariosa Diagnostics, Inc.
5945 Optical Court
San Jose, CA 95138

Questions:

sjc.clientservices@roche.com

US: (855) 927-4627

Intl: +1 (925) 854-6246

PATIENT NAME:	Jane Doe
DATE OF BIRTH: (MM/DD/YYYY)	01/01/1980
MRN:	1234567890123456789
LABORATORY ID:	AD12345678-PAT
OTHER ID:	
GESTATIONAL AGE:	10 wks 5 days
# OF FETUSES:	1
IVF STATUS:	non-IVF pregnancy
COLLECTION DATE (MM/DD/YYYY) :	07/20/2015
RECEIVED DATE (MM/DD/YYYY) :	07/21/2015
ACCOUNT #:	7654321
CLINIC NAME:	The Clinic Offering Test
REFERRING/ORDERING CLINICIAN:	Ordering Physician MD
REFERRING/ORDERING CLINICIAN FAX #:	123-456-7890
OTHER CLINICIAN:	Genetic Counselor MA, CGC
OTHER CLINICIAN FAX #:	987-654-3210
REPORT DATE: (MM/DD/YYYY)	07/28/2015

Test Results

Fetal cfDNA Percentage: 10.5%

CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 18 (T18)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient

22q11.2 — **No evidence of a deletion observed**

TEST DESCRIPTION

The Harmony Prenatal Test® measures the relative proportion of chromosomes to aid in the assessment of fetal trisomies 21, 18, and 13. Harmony® 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.

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. "High probability of a deletion" 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. "No evidence of a deletion observed" 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

	Detection Rate	False Positive Rate
T21	> 99% (95% CI: 97.9-99.8%)	< 0.1% (95% CI: 0.02-0.08%)
T18	97.4% (95% CI: 93.4-99.0%)	< 0.1% (95% CI: 0.01-0.05%)
T13	93.8% (95% CI: 79.9-98.3%)	< 0.1% (95% CI: 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: http://www.harmonytest.com/PPV

22q11.2: Limited numbers of 22q11.2 cases have been evaluated to date.

REFERENCES: Stokowski R et al. Prenatal Diagnosis 2015; 35, 1-4; Data on file.

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OTHER CLINICIAN FAX #:	987-654-3210
REPORT DATE: (MM/DD/YYYY)	07/28/2015

Test Results

Fetal cfDNA Percentage: 10.5%

CHROMOSOME	RESULT	PROBABILITY	RECOMMENDATION
Trisomy 21 (T21)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 18 (T18)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient
Trisomy 13 (T13)	Low Probability	Less than 1/10,000 (0.01%)	Review results with patient

22q11.2 — **High probability of a deletion - Recommend genetic counseling**

TEST DESCRIPTION

The Harmony Prenatal Test® measures the relative proportion of chromosomes to aid in the assessment of fetal trisomies 21, 18, and 13. Harmony® 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.

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. "High probability of a deletion" 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. "No evidence of a deletion observed" 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

	Detection Rate	False Positive Rate
T21	> 99% (95% CI: 97.9-99.8%)	< 0.1% (95% CI: 0.02-0.08%)
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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: http://www.harmonytest.com/PPV

22q11.2: Limited numbers of 22q11.2 cases have been evaluated to date.

REFERENCES: Stokowski R et al. Prenatal Diagnosis 2015; 35, 1-4; Data on file.

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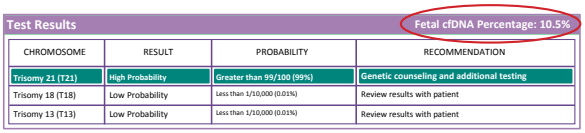
Harmony Prenatal Test Options

The Harmony Prenatal Test is validated for use in singleton, twin, and IVF pregnancies, including self and non-self egg donor pregnancies.[1]

		AVAILABLE FOR		
		SINGLETON	EGG DONOR & IVF	TWINS
Harmony Prenatal Test	Evaluates the probability of fetal trisomy 21, trisomy 18 and trisomy 13.	✓	✓	✓
The following test options are also available from the same blood draw:				
☐ **Fetal Sex**	Provides information regarding fetal sex. Assessment of fetal sex does not include assessment of sex chromosome aneuploidy. In twin pregnancies, a female result applies to both fetuses; a male result applies to one or both fetuses.	✓	✓	✓
☐ **Monosomy X**	Evaluates the probability of monosomy X, but no information regarding other sex chromosome aneuploidies.	✓	✓	
☐ **Sex Chromosome Aneuploidy Panel**	Evaluates the probability of X and Y chromosome aneuploidies, including monosomy X, XXX, XXY, XYY and XXYY.	✓	✓	
☐ **22q11.2**	Evaluates the probability of 22q11.2 deletion	✓	✓	

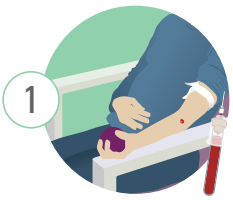
For both Monosomy X and the Sex Chromosome Aneuploidy Panel, fetal sex will only be reported if the Fetal Sex box is checked separately. However if the result indicates a high risk for sex chromosome aneuploidy, then this risk assessment will indirectly provide information regarding fetal sex. The Harmony Prenatal test is not available for more than 2 fetuses.

The Harmony Prenatal Test provides clear answers to questions that matter



- Clear Answers Early
- Personalized results that incorporate chromosome quantification fetal DNA fraction, gestational age, and maternal age [2-4]

Three Steps to Clarity



Draw a maternal blood sample at 10 weeks or later in pregnancy.

Submit sample and test requisition form per collection instructions

Lab results in as soon as little as 3 days from receipt, most in 5 days or less.

www.harmonytest.com

For assistance email **sjc.clientservices@roche.com** or call **1-855-927-4672** Outside the USA, call **+1 925-854-6246**

1. Stokowski et al. Prenatal Diagnosis 2015, 35, 1–4
2. Sparks et al. Prenat Diagn. 2012 Jan;32(1):3-9.
3. Sparks et al. Am J Obstet Gynecol. 2012 Apr;206(4):319.e1-9.
4. Juneau et al. Fetal Diagn Ther. 2014;36(4):282-6.

The Harmony Prenatal Test was developed, and its performance characteristics determined by Ariosa Diagnostics, a CLIA and CAP accredited clinical laboratory in San Jose, CA USA.

This testing service has not been cleared or approved by the US FDA.

Harmony is a non-invasive prenatal test (NIPT) based on cell-free DNA analysis and is considered a prenatal screening test, not a diagnostic test. Harmony does not screen for potential chromosomal or genetic conditions other than those expressly identified in this document. Before making any treatment decisions, all women should discuss their results with their healthcare provider, who can recommend confirmatory, diagnostic testing where appropriate.

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