Bumrungrad International
โรงพยาบาลบำรุงราษฎร์

NIPT Report

Patient Details:			
Patient ID		**Clinician Name**	
Patient Forename		**Hospital/Clinic Name**	
Patient Surname		**Date of Blood Draw**	02 Dec 2018
Patient Date of Birth	03 Nov 1973	**Maternal Age (at test)**	45 years
Pregnancy Status:	Singleton	**Gestation Age (at test)**	10

Test Results:

Trisomy	Background Risk	Risk Score	Clinical Summary
Trisomy 21 (Down's Syndrome)	1 : 16	> 95%	**High Risk** **Invasive Test Recommended**
Trisomy 18 (Edwards' Syndrome)	1 : 34	< 1 : 1,000,000 (<0.0001%)	Low Risk
Trisomy 13 (Patau's Syndrome)	1 : 107	< 1 : 1,000,000 (<0.0001%)	Low Risk

Fetal Fraction: 6%

Fetal Sex: Female

Supplementary Information:

- The detection rate of the IONA® test for trisomies 21, 18 and 13 is >99%.
- If fetal sex determination is requested, the accuracy is 99%. A "Sex Determination Failure" result may be reported if there is insufficient data to support the sex determination analysis. This is separate from the trisomy analysis and does not reflect on the quality of any other result generated by the IONA® test.
- The IONA® test estimates the risk of trisomies by determining the relative amounts of chromosomes 13, 18 and 21 in placentally-derived cell-free DNA extracted from the mother's plasma. The adjusted risk accounts for the background risk of the mother at the time of sampling (default). Additionally, the test may use the results of the First Trimester Combined Test as the background risk. If this has been done, a superscript ^{CT} will appear by the background risk next to any, or all, of the trisomy results.
- The IONA® test is a screening test and a high risk result should be discussed with the healthcare professional and confirmed by an appropriate diagnostic test (e.g. amniocentesis).
- The maternal age-adjusted risk score is capped. The cap is derived from an estimate of the prevalence of biological factors such as placental mosaicism. The result caps are: T21 >95%, T18 >75% and T13 >60%. These are the maximum risk estimates displayed on the report.
- In dichorionic twins, scientific publications suggest that the detection rate is reduced from greater than 99% to about 95%.
- A result with an IONA® test risk score greater than or equal to 1:150 (~0.67%) is considered high risk.

This test is indicated for screening NOT diagnosis — (results should be reviewed and discussed with your healthcare provider)

Originating sample ID: **N/A**
Sequencing run and sample validity checks passed: **Yes**
IONA® Software version: **TOA**: 1.7.0.7833.746; **DAA**: 1.7.0.7833.572

Sample notes (if entered):

Reported by

(E-signature)

(Mallika Chaowanathikhom, M.Sc.)

Approved by

(E-signature)

(Wipa Panmontha, Ph.D.)

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NIPT Report

Patient Details:			
Patient ID		**Clinician Name**	
Patient Forename		**Hospital/Clinic Name**	
Patient Surname		**Date of Blood Draw**	10 Dec 2018
Patient Date of Birth	19 Nov 1986	**Maternal Age (at test)**	32 years
Pregnancy Status:	Singleton	**Gestation Age (at test)**	12

Test Results:

Trisomy	Background Risk	Risk Score	Clinical Summary
Trisomy 21 (Down's Syndrome)	1 : 420	1 : 783,624 (0.0001%)	Low Risk
Trisomy 18 (Edwards' Syndrome)	1 : 976	< 1 : 1,000,000 (<0.0001%)	Low Risk
Trisomy 13 (Patau's Syndrome)	1 : 3074	< 1 : 1,000,000 (<0.0001%)	Low Risk

Fetal Fraction: 8%

Fetal Sex: Female

Supplementary Information:

- The detection rate of the IONA® test for trisomies 21, 18 and 13 is >99%.
- If fetal sex determination is requested, the accuracy is 99%. A "Sex Determination Failure" result may be reported if there is insufficient data to support the sex determination analysis. This is separate from the trisomy analysis and does not reflect on the quality of any other result generated by the IONA® test.
- The IONA® test estimates the risk of trisomies by determining the relative amounts of chromosomes 13, 18 and 21 in placentally-derived cell-free DNA extracted from the mother's plasma. The adjusted risk accounts for the background risk of the mother at the time of sampling (default). Additionally, the test may use the results of the First Trimester Combined Test as the background risk. If this has been done, a superscript ^{CT} will appear by the background risk next to any, or all, of the trisomy results.
- The IONA® test is a screening test and a high risk result should be discussed with the healthcare professional and confirmed by an appropriate diagnostic test (e.g. amniocentesis).
- The maternal age-adjusted risk score is capped. The cap is derived from an estimate of the prevalence of biological factors such as placental mosaicism. The result caps are: T21 >95%, T18 >75% and T13 >60%. These are the maximum risk estimates displayed on the report.
- In dichorionic twins, scientific publications suggest that the detection rate is reduced from greater than 99% to about 95%.
- A result with an IONA® test risk score greater than or equal to 1:150 (~0.67%) is considered high risk.

This test is indicated for screening NOT diagnosis — (results should be reviewed and discussed with your healthcare provider)

Originating sample ID: **N/A**
Sequencing run and sample validity checks passed: **Yes**
IONA® Software version: **TOA**: 1.7.0.7833.746; **DAA**: 1.7.0.7833.572

Sample notes (if entered):

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Sex Chromosome Aneuploidy Report

Patient Details:			
Patient ID		**Clinician Name**	
Patient Name		**Hospital/Clinic Name**	
Patient Surname		**Date of Blood Draw**	17-Dec-18
Patient Date of Birth	23-Feb-85	**Maternal Age (at test)**	33
Pregnancy Status	Single	**Gestation Age (at test)**	10

Test Results:		
Sex Chromosomes		Low Risk

Sex Chromosome Aneuploidies:		
XO		Low Risk
XXY/XYY		Low Risk
XXX		Low Risk

Risk description: Low risk; High Risk - Further Investigation Recommended

Supplementary Information:

- The NIPT test screens a maternal blood sample for chromosome aneuploidy in fetal DNA using the following methodology:
- Extraction of cell-free placental DNA from the maternal blood sample
- High throughput sequencing of the extracted cell-free placental DNA
- Calculation of molecular mass of placental DNA in all chromosomes
- The method is intended for use in pregnant women who are at least 10+0 weeks pregnant. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly lower in twin pregnancies due to multiple sources of fetal DNA
- Based on the scope, the NIPT test can detect the following:
- Sex chromosomal aneuploidies: XO, XXX, and XXY/XYY

Result is indicated for screening, NOT diagnosis - (results should be reviewed and discussed with your healthcare provider)

Originating sample ID: N/A
Algorithm Version:
Pipeline version: sage_link_v2

Sample notes (if entered):

Reported by

(E-signature)

(Mallika Chaowanathikhom, M.Sc.)

Approved by

(E-signature)

(Wipa Panmontha, Ph.D.)

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Sex Chromosome Aneuploidy Report

Patient Details:

Patient ID		**Clinician Name**	
Patient Name		**Hospital/Clinic Name**	
Patient Surname		**Date of Blood Draw**	30-Oct-18
Patient Date of Birth	01-Jan-92	**Maternal Age (at test)**	26
Pregnancy Status:	Single	**Gestation Age (at test)**	12

Test Results:

Sex Chromosomes	🔴	**High Risk** **Further Investigation Recommended**

Sex Chromosome Aneuploidies:

XO	🔴	**High Risk** **Further Investigation Recommended**
XXY/XYY	🟢	Low Risk
XXX	🟢	Low Risk

※ Risk description: 🟢 Low risk; 🔴 High Risk – Further Investigation Recommended

Supplementary Information:

- The NIPT test screens a maternal blood sample for chromosome aneuploidy in fetal DNA using the following methodology:
- Extraction of cell-free placental DNA from the maternal blood sample
- High throughput sequencing of the extracted cell-free placental DNA
- Calculation of molecular mass of placental DNA in all chromosomes
- The method is intended for use in pregnant women who are at least 10+0 weeks pregnant. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly lower in twin pregnancies due to multiple sources of fetal DNA.
- Based on the scope, the NIPT test can detect the following:
- Sex chromosomal aneuploidies: XO, XXX, and XXY/XYY

Result is indicated for screening, NOT diagnosis – (results should be reviewed and discussed with your healthcare provider)

Originating sample ID: N/A
Algorithm Version:
Pipeline version: sage_link_v2

Sample notes (if entered):

Reported by	Approved by
(E-signature)	(E-signature)
(Mallika Chaowanathikhom, M.Sc.)	(Wipa Panmontha, Ph.D.)

Form: UG-GS004-SCA-2018 v1

Revision: 09/Nov/18
TIID Ref:

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Microdeletion Screening Test Report

Patient Details:

Patient ID		**Clinician Name**	
Patient Name		**Hospital/Clinic Name**	
Patient Surname		**Date of Blood Draw**	17-Dec-18
Patient Date of Birth	23-Feb-85	**Maternal Age (at test)**	33
Pregnancy Status:	Single	**Gestation Age (at test)**	10

Test Results:

Microdeletion Syndromes	🟢	Low Risk

Microdeletion Syndrome:

DiGeorge syndrome	🟢	Low Risk
1p36 deletion syndrome	🟢	Low Risk
Angelman syndrome / Prader-Willi syndrome	🟢	Low Risk
Cri-du-Chat syndrome	🟢	Low Risk
Wolf-Hirschhorn syndrome	🟢	Low Risk

※ Risk description: 🟢 Low risk; 🔴 High Risk – Further Investigation Recommended

Supplementary Information:

- The NIPT test screens a maternal blood sample for microdeletions in fetal DNA using the following methodology:
  (1) Extraction of cell-free placental DNA from the maternal blood sample
  (2) High throughput sequencing of the extracted cell-free placental DNA
  (3) Calculation of molecular mass of placental DNA in all chromosomes
- The method is intended for use in pregnant women who are at least 10+0 weeks pregnant. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly lower in twin pregnancies due to multiple sources of fetal DNA.
- Based on the scope, the NIPT test can detect the following: Microdeletions - 5 specific disorders including:
  - DiGeorge syndrome
  - 1p36 deletion syndrome
  - Angelman syndrome / Prader-Willi syndrome
  - Cri-du-Chat syndrome
  - Wolf-Hirschhorn syndrome

Result is indicated for screening, NOT diagnosis – (results should be reviewed and discussed with your healthcare provider)

Originating sample ID: N/A
Algorithm Version:
Pipeline version:

Sample notes (if entered):

Reported by
(E-signature)
(Mallika Chaowanathikhom, M.Sc.)

Approved by
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(Wipa Panmontha, Ph.D.)

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Autosome Screening Test Report

Patient Details:			
Patient ID		**Clinician Name**	
Patient Name		**Hospital/Clinic Name**	
Patient Surname		**Date of Blood Draw**	17-Dec-18
Patient Date of Birth	23-Feb-85	**Maternal Age (at test)**	33
Pregnancy Status	Single	**Gestation Age (at test)**	10

Test Results:	
Autosomes	Low Risk

Supplementary Information:

- The NIPT test screens a maternal blood sample for chromosome aneuploidy in fetal DNA using the following methodology:
  - (1) Extraction of cell-free placental DNA from the maternal blood sample
  - (2) High throughput sequencing of the extracted cell-free placental DNA
  - (3) Calculation of molecular mass of placental DNA in all chromosomes
- The method is intended for use in pregnant women who are at least 10+0 weeks pregnant. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly lower in twin pregnancies due to multiple sources of fetal DNA.
- Based on the scope, the NIPT test can detect the following:
  - (1) Autosomes without chromosome 13, 18 and 21
- The test is capable of genome-wide aneuploidy detection over the whole fetal genome and gives the results for 19 pairs of chromosomes.
- In a study of over 2000 samples, 6 samples were determined to be at high-risk of having an autosomal aneuploidy other than 13, 18 and 21. This is a prevalence rate of 0.3%, which is consistent with prevalence in published studies.

Result is indicated for screening, NOT diagnosis - (results should be reviewed and discussed with your healthcare provider)

Originating sample ID: N/A
Algorithm Version:
Pipeline version: sage_link_v2

Sample notes (if entered):

Reported by

(E-signature)

(Mallika Chaowanathikhorn, M.Sc.)

Approved by

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(Wipa Panmontha, Ph.D.)

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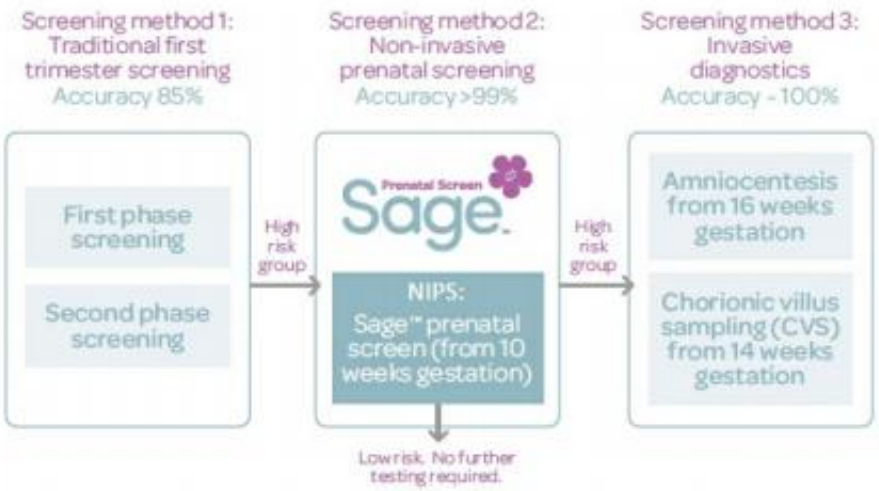
Autosome Screening Test Report

Chromosome	Risk	Test Results
Chromosome 1		Low Risk
Chromosome 2		Low Risk
Chromosome 3		Low Risk
Chromosome 4		Low Risk
Chromosome 5		Low Risk
Chromosome 6		Low Risk
Chromosome 7		Low Risk
Chromosome 8		Low Risk
Chromosome 9		Low Risk
Chromosome 10		Low Risk
Chromosome 11		Low Risk
Chromosome 12		Low Risk
Chromosome 14		Low Risk
Chromosome 15		Low Risk
Chromosome 16		Low Risk
Chromosome 17		Low Risk
Chromosome 19		Low Risk
Chromosome 20		Low Risk
Chromosome 22		Low Risk

※ Risk description: Low risk; High Risk - Further Investigation Recommended

Sage™ prenatal screening pathway



About Sage™ prenatal screen

The Sage™ prenatal screen is a new advanced non-invasive prenatal screening solution using the latest developments in DNA technology to detect placental DNA in maternal blood. Sage™ offers a menu-based chromosome analysis to estimate the risk of a fetus having Down's syndrome and other genetic disorders. Enabling pregnant women and their families fast, safe and reliable results and reducing the need for invasive tests and the associated risks, stress and anxiety. Sage™ is indicated for use in pregnant women who are at least 10 weeks pregnant. Chromosomal aneuploidy can then be detected using bioinformatics analyses, where the detection rate and sensitivity are over 99%.

Limitations

Sage™ is a screening test and all high-risk results should be confirmed through further investigation which may include tests such as amniocentesis or Chorionic Villus Sampling (CVS). Pregnant women with a high-risk result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. Pregnant women with a negative test result do not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes (micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparental disomy). The test is not reportable for known multiple gestations, or if the gestational age is less than 10 weeks.

Test method

A simple maternal blood sample is taken from the pregnant mother from 10 weeks gestation without any risk to the fetus. Circulating cell-free placental DNA was purified from the plasma component of anti-coagulated 10mL of maternal whole blood. It was then converted into a genomic DNA library for Next Generation Sequencing and then determination of chromosomal aneuploidy.

References:

1. Obstet Gynecol 2012;119:890-901.
2. BMJ 2011;342:c7401.
3. Prenat Diagn 2012;32:c7401.
4. ACOG/SMFM Joint Committee Opinion No. 545, Dec 2012.