Prenatal Screen Sage™
Safe, accurate, genetic evaluation

NON-INVASIVE PRENATAL ANEUPLOIDIES AND MICRODELETION SCREENING TEST REPORT

Patient Details:			
Patient ID	patient_id	**Clinician Name**	physician
Patient Surname	surname	**Hospital/Clinic Name**	hospital_name
Patient Forename	forename	**Pregnancy Status: Singleton/Twin**	pregnancy_type
Date of Blood Draw	collection_date	**Report Generated:**	created_date
Patient Date of Birth	dob	**TIID**	specimen_id_2
Reporting facility	No.376-5, Fuxing Rd., Shulin Dist., New Taipei City 238, Taiwan (R.O.C.)		+886 2 26758068

I. Screening results

Chromosomes	Risk	Z score	Test Results	Reference interval
Chromosome 21 			Low Risk	-6<Z score<2.8
Chromosome 18 			Low Risk	-6<Z score<2.8
Chromosome 13 			Low Risk	-6<Z score<2.8
Chromosome Y 	-		Detected	NA
Sex Chromosomes		Part III	Low Risk	Part III
Other Chromosomes		Part IV	Low Risk	Part IV
Microdeletion Syndromes		Part V	Low Risk	Part V
Fetal fraction				

II. Supplementary information

- The NIPS test screens a maternal blood sample for chromosome aneuploidy in placental 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 NIPS test can detect the following:
  (1) Whole Genome - 23 pairs of human chromosomes
  (2) Sex chromosomal aneuploidies: XO, XXX, XXY/XYY
  (3) Microdeletions - 5 specific disorders including: DiGeorge syndrome, 1p36 deletion syndrome, Angelman syndrome/Prader-Willi syndrome, Cri-du-Chat syndrome and Wolf-Hirschhorn syndrome
- The test is capable of genome-wide aneuploidy detection over the whole fetal genome and gives the results for 23 pairs of chromosomes. This test confers an accuracy of up to **99%** on the detection of fetal aneuploidy for chromosomes 13, 18 and 21. 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.

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

Doctor | Laboratory Director | Bioinformatics Scientist

III. Sex Chromosomes

Sex Chromosome Aneuploidies	Risk	Z score	Test Results	Reference interval
XO			Low Risk	Male -3<Z score<3 Female -2.8<Z score<2.8
XXY/XYY			Low Risk	
XXX			Low Risk	

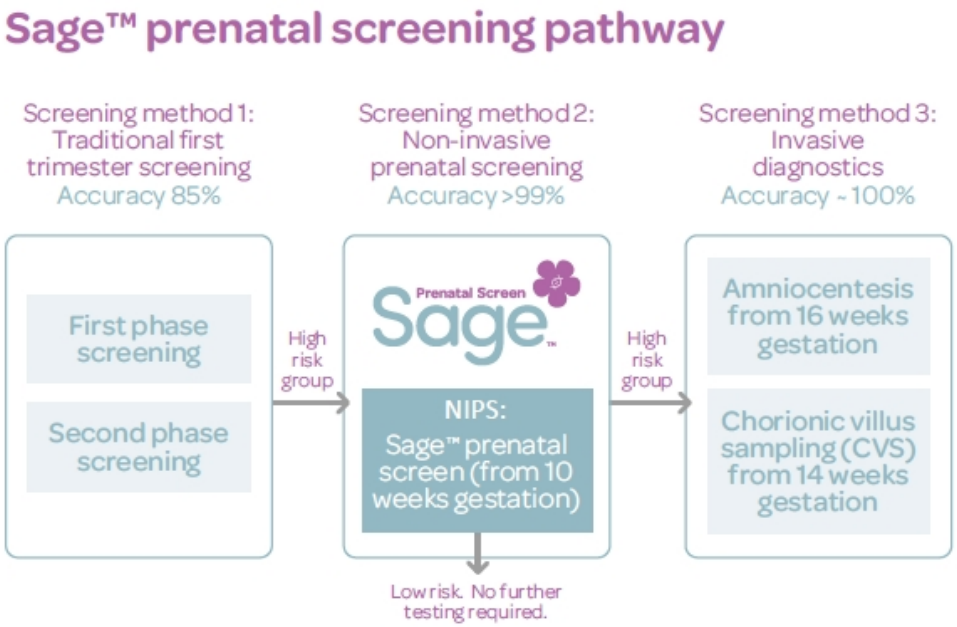
IV. Other Chromosomes

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

V. Microdeletion Syndromes

Microdeletion Syndrome	Risk	Z score	Test Results	Reference interval
DiGeorge syndrome			Low Risk	-4<Z score
1p36 deletion syndrome			Low Risk	-4<Z score
Angelman syndrome / Prader-Willi syndrome			Low Risk	-4<Z score
Cri-du-Chat syndrome			Low Risk	-4<Z score
Wolf-Hirschhorn syndrome			Low Risk	-4<Z score

Pipeline version: XXXXXXXX



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.