

## PRENATAL DIAGNOSIS AND SCREENING COMPARISON

|                                                          | TESTING OPTIONS                                                                                                                                                                                                                                                                                                                                                                                                                                             | WHEN PERFORMED IN PREGNANCY                                                                                    | RESULTS AVAILABLE                                                                    | TYPE OF TEST & DETECTION RATE/ACCURACY                                                                                                                                                                                                       | CHANCE OF MISCARRIAGE FROM PROCEDURE        |
|----------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------|
| <b>D<br/>I<br/>A<br/>G<br/>N<br/>O<br/>S<br/>I<br/>S</b> | <b>CHORIONIC VILLUS SAMPLING (CVS)<sup>1,2</sup></b><br>Analysis of placental tissue for extra, missing or rearranged chromosomes or other genetic diseases known in the family or suspected in this pregnancy. Does not screen for open spina bifida (OSB) <sup>3</sup> .                                                                                                                                                                                  | 10+0 to 12+6 wks                                                                                               | 8 to 11 days<br><br>~ 11 to 15 weeks in pregnancy                                    | Fetal chromosome analysis only<br><br>~ <b>99% accurate</b> in identifying structural chromosome problems, ≤2% risk for mosaicism                                                                                                            | "Not significantly different from amnio"    |
|                                                          | <b>AMNIOCENTESIS<sup>1</sup></b><br>Analysis of fetal cells in amniotic fluid for extra, missing or rearranged chromosomes or genetic diseases known in the family or suspected in pregnancy. Screens for OSB.                                                                                                                                                                                                                                              | 15+5 to 23+6 wks recommended                                                                                   | 6 to 8 days<br><br>~ 16 to 23 weeks in pregnancy                                     | Fetal chromosome analysis & Screening of amniotic fluid AFP<br><br><b>99.9% accurate</b> in identifying structural chromosome problems, ≥98% open spina bifida detected                                                                      | Less than 1 in 300 to 1 in 500 (< 0.2-0.3%) |
|                                                          | <b>NON-INVASIVE PRENATAL TESTING (NIPT)</b><br>(InformaSeq testing by Integrated Genetics/LabCorp)<br>DNA-based maternal blood test to identify if the baby may have additional amounts of chromosomes 13, 18, and 21. This testing can also predict fetal sex and sex chromosome variations.                                                                                                                                                               | 10+0 wks or later                                                                                              | 7 to 10 days                                                                         | <b>Condition</b> – Detection / FPR <sup>4</sup><br><b>Down syndrome</b> – 99.9% / 0.2%<br><b>Trisomy 18</b> – 97.4% / 0.4%<br><b>Trisomy 13</b> – 87.5% / <0.1%<br>Monosomy X – 95.0% / 1.0%<br>Female – 97.6% / 0.8%<br>Male – 99.1% / 1.1% | None                                        |
| <b>S<br/>C<br/>R<br/>E<br/>E<br/>N<br/>I<br/>N<br/>G</b> | <b>SERUM INTEGRATED SCREEN<sup>5</sup></b><br>Screening for Down syndrome, trisomy 18 and open spina bifida that involves two blood draws. Combines measurement of maternal serum PAPP-A, and hHCG in 1 <sup>st</sup> trimester and HCG, AFP, UE3, DIA in 2 <sup>nd</sup> trimester. A result will not be given until after the second blood. Indicates high risk if >1:270 for Down syndrome, >1:100 for trisomy 18, or OSB if AFP ≥2.5MoM for singletons. | 1 <sup>st</sup> blood draw<br>10+3 to 13+6 wks<br><i>AND</i><br>2 <sup>nd</sup> blood draw<br>15+0 to 21+6 wks | 3 to 4 days after<br>2 <sup>nd</sup> blood draw<br><br>~ 16 to 18 weeks in pregnancy | <b>Condition</b> – Detection / FPR <sup>4</sup><br><b>Down syndrome</b> – 92.0% / 5.0%<br><b>Trisomy 18</b> – 90.0% / <0.2%<br><br><b>80%</b> Open spina bifida,<br><b>95%</b> Anencephaly detection<br>Screen positive rate is 1 to 3%      | None                                        |
|                                                          | <b>LEVEL II ULTRASOUND</b><br>Examines the baby and placenta using sound waves to detect many physical changes in development (birth defects) and assess overall health of pregnancy.                                                                                                                                                                                                                                                                       | 18 to 22 weeks                                                                                                 | Same day as ultrasound                                                               | Expected to identify<br>~35 to 50% of birth defects<br>~97% of OSB                                                                                                                                                                           | None                                        |

<sup>1</sup> Indications include maternal age of 35 or above, abnormal screening results, ultrasound findings, or family history of a known or suspected genetic condition.

<sup>2</sup> Following CVS or NIPT, an additional MSAFP only blood draw at or after 15.0 weeks is available to test for open spina bifida.

<sup>3</sup> Examples of open spina bifida include anencephaly or any opening along the baby's head, neck or spine.

<sup>4</sup> FPR is the False Positive Rate.

<sup>5</sup> If testing is not initiated by 14 weeks, patients still have option of Quad screen (Detection: 79% Down syndrome, 73% T18, and OSB is same as sequential).