According to the ISPD, The following protocol options are currently considered appropriate for a screening program:

1. cfDNA screening as a primary test offered to all pregnant women.
2. cfDNA secondary to a high-risk assessment based on serum and ultrasound screening protocols (options 4-9 below).
3. cfDNA contingently offered to a broader group of women ascertained as having high or intermediate risks by conventional screening. Contingent provision of cfDNA could also include a protocol in which women with very high risks are offered invasive prenatal diagnosis, while those with intermediate risk are offered cfDNA.
4. Ultrasound nuchal translucency at 11-13 completed weeks combined with serum markers at 9-13 weeks' gestation.
5. Extending option (4) to include other first trimester serum or sonographic markers. Ultrasound performance needs to be prospectively validated by the center where the screening is performed.
6. A contingent test whereby women with borderline risks from option (4) have option (5) at a specialist center and risk is subsequently modified.
7. Four maternal serum markers (quadruple test) at 15-19 weeks, for women who first attend after 13 weeks 6 days gestation.
8. Combining options (4) and (7) in either a stepwise or contingent protocol – provided that all screening test data are included in the final risk assessment. Integrated screening can be offered as a serum integrated test when NT measurement is unavailable.
9. Contingent second trimester ultrasound to modify risks for aneuploidy for women having options (4), (7) or (8). Ultrasound performance must be prospectively validated by the center where the screening is performed.

ISPD suggests that combining biochemical screening with first trimester ultrasound nuchal translucency is optimal for patients desiring aneuploidy screening.

The ISPD states that cfDNA can be considered for women who are:

- determined to be high risk through other screening
- who did not receive other screening AND who are considered to be high risk based on
- family history,
- maternal age or
- ultrasound makers suggestive of T21, T13 or T18.

Women interested in cfDNA should receive detailed counseling that explains the benefits

and limitations of the tests, and patients should be made aware that these tests are still under clinical development.

Unless exceptional circumstances exist, the ISPD recommends against:

- The use of maternal age as a sole criterion for aneuploidy risk assessment.
- First trimester measurement of NT with no additional tests
- Conventional screening tests for chromosome abnormalities following successful and unambiguous cfDNA screening.

*Of note-many members of the committees in the author groups of these ISPD documents are employed as staff or consultants for companies offering cfDNA.

Related Links:

The International Society of Prenatal Diagnosis (ISPD) Position Statement from the Chromosome Abnormality Screening Committee on Behalf of the Board of the International Society for Prenatal Diagnosis (2015)

Position Statement from the Aneuploidy Screening Committee on Behalf of the Board of International Society for Prenatal Diagnosis – April 2013