In recent years, researchers have been trying to isolate traces of foetal DNA from maternal blood in order to identify a possible Down’s syndrome foetus and, trisomy of chromosomes 13 and 18.

At present, all International literature and the current Guidelines are against the utilization of such screening for clinical diagnostic purposes in general populations. In fact, the tests are extremely interesting but, for the moment, confined among the less proven prenatal Down screening. That’s why at moment there are too many uncertainties for to be used in diagnostics routine.

According to the position statement from:
- International Society for Prenatal Diagnosis (ISPD) (1);
- American College of Medical Genetics and Genomics (ACMG) (2);
- American College of Obstetricians and Gynecologists Committee on Genetics (ACOGG) (3);
- California Technology Assessment Forum (CTAF) (4);
- National Italian Guide Lines (5).

The Italian College of Fetal Maternal Medicine stated the present document:

In summary
- To date, the only accredited screening tests are the “combined”, being based on nuchal translucency and placental proteins. Regarding these tests, our scientific research centre, following current institutional guidelines, proposes the ones that have been experimented in the national population (SCA-TEST);
- The only diagnostic tests for foetal genetic and genomic anomalies are the chorionic villus sampling (CVS) and the amniocentesis which currently provoke the same levels of abortion risk as in the general population (6-9). These methods, in fact, since the introduction of modern and tested genomic techniques using CGH, have been increasingly used in diagnostics;
- The routine and diagnostic utilization of a test based on the research of foetal DNA in maternal blood, today, can no longer be proposed as a first-choice test. Even though the pregnant mother requests the test herself, it is necessary to provide detailed and explicit information regarding its limits (10-12).

Regarding the legal and financial aspects connected to the use of these new screening tests, some people have criticized the high costs and excessive cost/benefit ratio, commercial interests and, finally, the legal complications that have already arisen from the introduction in the commerce of these techniques. These aspects represent further elements to be taken into consideration so they can be better developed and possibly lead to future clinical use (13-15).

Recommendations
- Screening for Down’s syndrome through the search for fetal DNA in maternal blood must not be encouraged as a first choice, given there are many combined screening tests that have been widely experimented and which, today, can be considered to be more reliable;
- Pregnant women that opt for the prenatal screening of aneuploidy through the analysis of foetal DNA in maternal blood must be informed that such testing does not provide clinically validated results;
- The recent widespread use of such testing in clinical practice has not been supported by the International scientific community and their commercial sale by promoting companies and laboratories creates risks for those who use them for diagnostic purposes from a legal point of view. Therefore, the clinic, in order to operate correctly, and to be sufficiently safeguarded against disputes regarding professional responsibility, must state that:
  • The clinical use of such screening is not valid and cannot yet substitute combined tests;
  • There is a huge difference between a screening test and a diagnostic test. If a pregnant woman is looking for certainty in testing his own baby, they must be informed that the only diagnostic tests available are those carried out on foetal material extracted by CVS or amniocentesis.

Italian College of Fetal Maternal Medicine, May 2013
References


5. www.ifeto.it.


