



MINISTERO
DELL'INTERNO

RISK ASSESSMENT

For years now, researches have been underway to try and identify foetuses at risk of chromosomal pathologies without resorting to invasive methods (amniocentesis and chorionic villus sampling): while ensuring a diagnostic and thus certain answer, these types are not devoid of risks.

The tendency is thus to limit such investigations to specific situations and as an in-depth examination following other tests.

To better understand the available investigation options and orientate yourself through the various methods and timeframes, the Central Tuscany USL (Local Health Authority) has established a Consultation, called "ACCESS TO PRENATAL SCREENING", which you should take part in before any prenatal investigation is performed and is booked by the Family Clinic midwife at the time of delivering the Pregnancy Booklet. It is preferable to be accompanied by the partner to the Consultation and inform yourself beforehand on the presence in the respective families of problems, particularly of a congenital type.

To date, the Tuscany Region offers the COMBINED TEST as a free-of-charge basic investigation for all women: the "combination" of maternal age, dosage of some blood markers (hCG and PAPP-A) and ultrasound assessments (nuchal translucency), allows a calculation of the risk for that specific pregnancy that the foetus is affected by trisomy 13, 18, 21.

The EXPANDED TEST represents an extension of the Combined type, and allows calculation of the risk of a pregnancy-related pathology on the maternal side: preeclampsia. It is performed through an additional blood sample and an ultrasound that assesses other parameters as well (foetal flowmetry of the uterine arteries). As this examination is not included in the Regional Pregnancy Booklet, it must be paid for.

If the combined test highlights an "intermediate" or "high" risk, the NIPT (Non Invasive Prenatal Test) will be suggested: by means of a blood sample, it is possible to detect fragments of placental DNA in the maternal blood and analyse them in order to highlight the risk of chromosomal abnormalities. This investigation envisages a contribution to the cost – i.e. a co-payment of charges by the user. Alternatively, or by way of a more in-depth analysis following a positive NIPT, the invasive test can be proceeded with.