Ultrasound

Over the course of your pregnancy, you will undergo several routine examinations designed to closely monitor not only your health but your baby's as well. Included among these procedures are a series of ultrasound exams as well as prenatal screening for fetal abnormalities.
Except in specific cases such as a multiple pregnancy or a family history of twins, you will undergo **three ultrasounds**.
These exams are performed using a manual probe that delivers high frequency sound waves to obtain an image of the fetus on a screen. As long as they are used for medical purposes, these ultrasound waves pose no threat to you or your baby[1]. Your first ultrasound, also referred to as a **dating scan**, will be performed around the **12th week** of pregnancy (calculated based on your last menstrual period). As its name indicates, this ultrasound is used to evaluate the gestational age of the pregnancy by measuring the size of the baby's head, abdomen and thigh bone as well as spine length. It is also used to estimate your expected date of delivery and to assess any potential risk of genetic disorder.
Two other **ultrasounds**, called **anatomy scans**, are performed around **22** and **32** weeks and consist in a detailed assessment of your baby's development. In particular, your baby's physical characteristics and organs are analyzed to make sure there are no abnormalities or malformations. Nevertheless, even when skillfully executed, ultrasound is not infallible and has its limits. The 32 week ultrasound is used to locate the placenta and screen for any delays in intra-uterine growth[1].
During the ultrasound, you will be lying down comfortably on your back on an examination table, in a room that has been darkened to facilitate viewing of the images displayed on the screen. Before beginning the exam, the sonographer will apply a gel to your abdomen which allows the sound waves to be transmitted. The ultrasound lets you see your baby for the first time and can be a very moving experience!

[1] https://www.has-sante.fr/portail/upload/docs/application/pdf/2012-06/rapport_echographies_foetales_vde.pdf

Prenatal screening

Prenatal screening for fetal abnormalities begins with the first ultrasound. Between the 11th and 14th weeks of pregnancy, a collection of fluid, called nuchal translucency, becomes visible at the back of the fetus's neck. Its thickness, the mother's age, the size of the baby and the levels of certain pregnancy hormones are **genetic disorder risk indicators**. This data is especially useful for assessing the risk of **trisomy 21** (Down Syndrome) and other less common disorders such as trisomy 13 (Patau syndrome), trisomy 18 (Edwards syndrome) and abnormalities affecting sex chromosomes X and Y)[2]. These parameters can be used to calculate a **probability threshold** for abnormalities, but cannot be used to validate a diagnosis. Beyond a certain threshold, **diagnostic exams** can be performed. These tests consist in analyzing chromosomes inside the cells of the fetus (fetal karyotype) and require a tissue sample obtained through amniocentesis or chorionic villus sampling (CVS). These invasive procedures carry a low risk of miscarriage, although the rate has decreased (from 1 percent to 0.1 percent) according to recent data[3].
It was recently recommended that **Non-Invasive Prenatal Testing (NIPT)**, which can take place starting from week 10[2], be performed before a karyotype analysis[3,4]. NIPT is based on the presence of fetal DNA in the maternal blood and is performed through a **simple blood draw**. The blood sample undergoes laboratory analysis to determine if it contains more than the usual amount of chromosomes 21, 13, 18, X or Y[2]. By using NIPT, the number of unnecessary invasive exams can be decreased, thereby reducing the associated risks. However, if the result of NIPT is positive, karyotyping is the only way to establish a diagnosis[2,3].

[2] http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Cell-free-DNA-Screening-for-Fetal-Aneuploidy
[3] https://www.has-sante.fr/portail/upload/docs/application/pdf/2017-06/dir71/presse_depistage_trisomie_21.pdf
[4] https://www.has-sante.fr/portail/jcms/c_2768535/fr/trisomie-21-la-has-actualise-ses-recommandations-concernant-le-depistage-prenatal-de-la-trisomie-21