Laboratoire Médical

Prenatal Screen Sage™

Safe and Accurate foetal genetic test

Sage™ prenatal screen

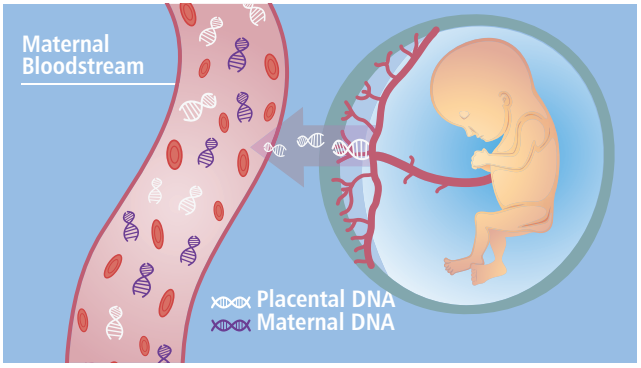
The Sage™ prenatal screen is a non-invasive prenatal test (NIPT) for pregnant women. It estimates the risk of a fetus having Down's syndrome and other genetic diseases.

Carried out on a small maternal blood sample, the test enables pregnant women to get fast, safe and reliable results, reducing the need for other invasive tests and the associated risks, stress and anxiety.

How does the Sage™ test work?

During pregnancy, the placenta leaks foetal cell-free DNA (cfDNA) which circulates in the maternal bloodstream. As a result, a maternal blood sample contains a mixture of foetal and maternal circulating DNA.

The Sage™ prenatal screen directly measures the amount of this cfDNA and can detect small changes in the DNA ratio between the maternal and foetal cfDNA to estimate the risk of a foetus having Down's syndrome and other genetic disorders.



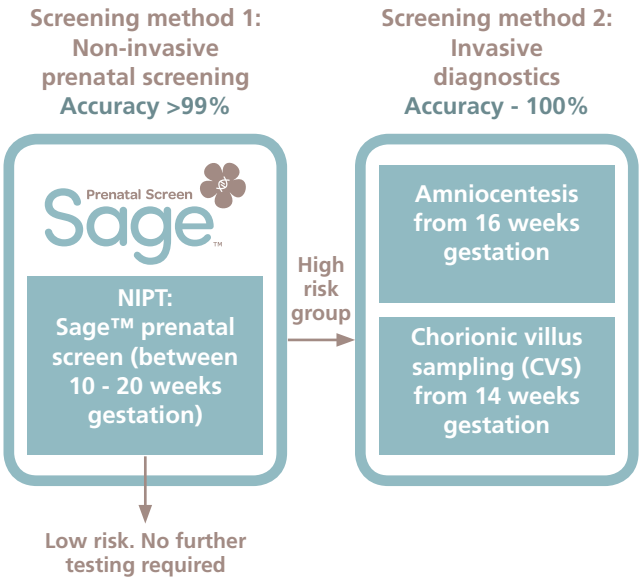
Who can have the Sage™ test?

- Mother must be between 10-20 weeks pregnant
- Both single or twin pregnancies
- The Sage™ prenatal screen is unsuitable if the mother has:
  - Cancer
  - Trisomy
  - Blood transfusion in the past 12 months
  - Undergone Stem Cell Therapy or Immunotherapy
  - Received an organ transplant

Advantages of the Sage™ test

- **Safe:** Non-invasive with no risk of miscarriage
- **Accurate:** >99% detection of autosomal trisomy conditions
- **Simple:** Uses just one maternal blood sample of 10ml.
- **In depth:** In addition to the common trisomies 21, 18 and 13, a whole chromosome analysis is available upon request, along with sex chromosome aneuploidies and microdeletions.

Sage™ prenatal screening pathway



What does Sage™ screen for?

1. Autosomal aneuploidies

- Trisomies occur when three, instead of the usual two, copies of a chromosome are present in each cell. When the number of chromosomes differs from the usual two this is referred to as an aneuploidy.
- The Sage™ prenatal screen test estimates the risk of a fetus having Down's syndrome (Trisomy 21), Edwards' syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13).
- Edwards' and Patau's syndrome are much rarer than Down's but are very serious and many affected babies are not born alive or do not survive for long after birth.

Accurate with >99% detection of trisomy conditions.

2. Sex chromosome aneuploidies

- In addition to the autosomal trisomies, the Sage™ prenatal screen may also report on sex chromosomal aneuploidies.
- Women have two 'X' chromosomes (XX) while men have one 'X' and one 'Y' chromosome (XY).
- The impact of sex chromosome aneuploidy is generally much milder than aneuploidy of chromosomes 13, 18, and 21. The following aneuploidies can be screened for:
  - Monosomy X – Turner syndrome
  - XXX – Triple X syndrome
  - XXY – Kleinfelter syndrome
  - XYY – Jacob's syndrome

3. Microdeletions

- A microdeletion syndrome is caused by the absence of a small portion of genetic material in the chromosome.
- They vary greatly in severity, with the symptoms of microdeletions ranging from minimal developmental delays to severe anomalies e.g. cardiac defects, neurological malformations, etc.

4. Fetal sex determination

Upon request, and in regions where fetal sex determination is permitted, the fetal sex can be determined with 99.9% accuracy.

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