

Understanding Genetic Screening in Pregnancy: Tests, Choices, and Support



Highlights

Genetic screening (prenatal screening) tests estimate the chances that your baby could have a genetic or chromosomal condition - they are available to anyone who is pregnant.

You have the right to choose whether or not to undergo a prenatal screening test.

Screening tests include the combined first trimester screening test, maternal serum screening, and the non-invasive prenatal test (NIPT).

If your screening test shows that your baby has a high chance of having a genetic condition, a genetic counsellor can help you decide on further testing, which may include chorionic villus sampling (CVS) or amniocentesis.

In This Article

Overview of Genetic Screening in Pregnancy

Types of Genetic Screening Tests

Combined First Trimester Screening

Second Trimester Maternal Serum Screening

Non-Invasive Prenatal Test (NIPT)

Making the Right Choice

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Costs of Genetic Screening
Genetic vs. Carrier Screening
Tools And Assistance

Overview of Genetic Screening in Pregnancy

Types of Genetic Screening Tests

Combined First Trimester Screening

Down syndrome \ (trisomy 21\
Edwards syndrome \ (trisomy 18\
Patau syndrome \ (trisomy 13\

The levels of certain proteins in the mother's blood
The thickness of the fluid behind the baby's neck \ (nuchal translucency\
The presence or absence of the baby's nasal bone
The mother's age and weight
How far along the pregnancy is

Second Trimester Maternal Serum Screening

Down syndrome \ (trisomy 21\
Edwards syndrome \ (trisomy 18\
Neural tube defects \ (e.g., spina bifida\

Blood test results
The mother's age and weight
Pregnancy age

Non-Invasive Prenatal Test \ (NIPT

Down syndrome
Edwards syndrome
Patau syndrome
Sex chromosome conditions
Microdeletion syndromes

Making the Right Choice

What to Expect After Screening

Costs of Genetic Screening

Whether you are a public or private patient
The type of test being performed
The healthcare provider's recommendations

Genetic vs. Carrier Screening

Tools And Assistance

Your doctor or midwife can help explain your test results and guide you through possible next steps based on your individual situation.

A genetic counsellor can provide detailed information about what the results mean, outline the risks, explain diagnostic testing options, and support you in making decisions that align with your values and preferences.

Mental health professionals such as psychologists or counsellors can assist with emotional well-being, especially if you're feeling stressed or overwhelmed.

Peer support groups or parent communities can connect you with others who have gone through similar experiences, offering a sense of solidarity and shared understanding.

Family and friends can also be a valuable source of emotional and practical support during this time.