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Prenatal Screening
for Birth Defects

Prenatal screening is an important part of routine care for pregnant women. It's often the first step in the detection of Down syndrome, trisomy 18, and open neural tube defects.

Over the years, screening tests have improved. More cases are picked up by current screens, and there are fewer false-positive results. This leads to fewer unnecessary, invasive follow-up tests. But the ideal test—a noninvasive diagnostic test—still eludes us.

Getting closer to the ideal test

For many years, scientists have been trying to test cell-free, fetal DNA found in the mother's blood. In the past year, tests that can do that have been commercialized. Unlike amniocentesis, these tests are a relatively noninvasive way of testing fetal cells for extra or missing chromosomes. That's why they are called noninvasive prenatal tests (NIPTs). NIPTs can detect trisomies 21, 18, and 13. Some can also detect 45,X (Turner syndrome), XXY (Klinefelter syndrome), and triple X syndrome.

Of course, NIPTs cannot detect neural tube defects.

Just how good are the NIPTs?

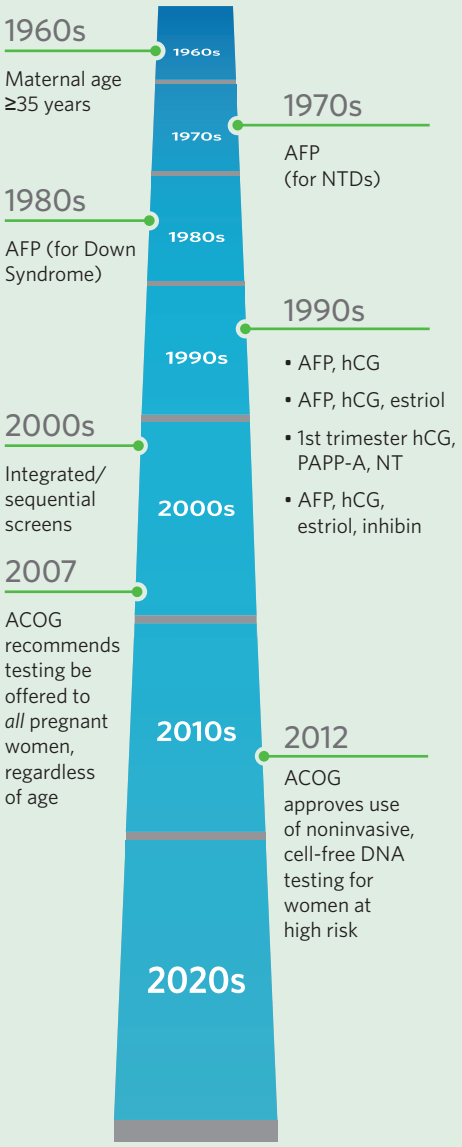
Data so far show that NIPTs are more accurate than all the maternal serum screening tests. But NIPTs have only been studied in pregnant women whose unborn child is more likely to have a birth defect. They have not been studied in women at low risk.

The table below gives a rough idea of how an NIPT compares to the best maternal serum screening test. But remember that data from the maternal serum screening test come from the general population. This includes women with low and high risk. Data from the NIPT, however, come primarily from women at high risk.

Birth Defect	Integrated Screen		NIPT[1-3]	
	% DR	% FPR	% DR	% FPR
Open neural tube defects	80	5	0	–
Down syndrome	92	5	>99	<0.1
Trisomy 18	90	5	>99	<0.1
Trisomy 13	0	–	>99	<0.1
Turner syndrome	0	–	92	<0.1

DR, detection rate; FPR, false-positive rate.

History of prenatal screening



Are NIPTs recommended?

Yes. These professional organizations have each issued statements in support of NIPTs:

- ACOG (American Congress of Obstetricians and Gynecologists)[4]
- ACMG (American College of Medical Genetics and Genomics)[5]
- ISPD (International Society for Prenatal Diagnosis)[6]
- NSGC (National Society of Genetic Counselors)[7]

When are NIPTs appropriate?

NIPTs are an option for screening women whose unborn children are more likely to have birth defects.[4-7] This includes women who:

- Are 35 years old or older
- Have had an ultrasound that indicated increased risk
- Have had a previous pregnancy with one of these birth defects
- Have had a maternal serum screening test that indicated increased risk

Testing can be performed as early as 9 weeks' gestation.

When are NIPTs not appropriate?

NIPTs are not for women who are at low risk of having a child with one of these birth defects.[4-7] It should not be offered as:

- A screening test for neural tube defects
- A diagnostic test; positive results should be followed up with CVS or amniocentesis

A second trimester AFP test should be ordered to screen for open neural tube defects.

Quest Diagnostics offers a complete menu

Quest Diagnostics offers maternal serum screening for open neural tube defects, Down syndrome, and trisomy 18. Screening can be performed in the first, second, or both first and second trimesters. Quest Diagnostics also offers an NIPT called the Panorama™ Prenatal Test.

For diagnosis of birth defects, Quest Diagnostics offers chromosome analysis using chorionic villus or amniotic fluid specimens. FISH and chromosomal microarray tests are offered as well. Amniotic fluid alpha-fetoprotein, acetylcholinesterase, and fetal hemoglobin are offered for open neural tube defect diagnosis.

References

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