Investigation Questions Accuracy of Prenatal Genetic Tests

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General Information

Source: NBC Nightly News  
Creator: Lester Holt/Kate Snow  
Resource Type: Video News Report  
Copyright: NBCUniversal Media, LLC.  
Event Date: 05/21/2015  
Air/Publish Date: 05/21/2015  
Clip Length: 00:03:15

Description

Prenatal screening tests reveal the risk of serious genetic abnormalities before a baby is born. A 2015 investigation by the New England Center for Investigative Reporting casts doubt on the reliability of these tests and doctors warn that they are not a replacement for diagnostic tests.

Keywords

Genetics, Genetic Testing, Prenatal, Prenatal DNA Test, Fetus, Pregnant, Pregnancy, Mothers, Blood Tests, Accuracy, Results, Stacie Chapman, Lincoln Chapman, Edwards Syndrome, Amniocentesis, CVS, Chorionic Villus Sampling, Error, Investigation, New England Center for Investigative Reporting, Reliability, Michael F Greene, Massachusetts General Hospital, OB-GYN, Obstetrician, Gynecologist, Risk, Disease, Diagnosis, Counseling, Genetic Counseling, Ethics, Medical Ethics, Health, Wellness, Debate

Citation

MLA

**APA**


**CHICAGO MANUAL OF STYLE**


**Transcript**

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LESTER HOLT, anchor:

We are back with an eye opening report in our special series this week, Would You Want To Know. It's about the new medical frontier in DNA testing. Tonight, a closer look at the new kind of genetic tests given to pregnant women to check the health of their unborn babies, a simple blood test promises more accurate results earlier than ever, but there are growing concerns these tests have been oversold and misunderstood. Our National Correspondent Kate Snow reports.

KATE SNOW, reporting:

When Stacie and Lincoln Chapman were expecting their first child, her doctors' office recommended a new kind of blood test to check her unborn baby's DNA, but then, Stacie's doctor called with terrible news. The test was positive for something called Edwards Syndrome.

STACIE CHAPMAN: She explained to me that if the baby did survive to full term, would last-- would survive maybe hours, maybe days.

SNOW: You must have been devastated.

CHAPMAN: Uh-huh, totally devastated.

SNOW: They made an appointment to terminate the pregnancy the very next day.

CHAPMAN: We didn't want the baby to suffer.

SNOW: But that night, the doctor called back and suggested they should wait after eight long weeks Stacie had amniocentesis and then an even bigger shock, her unborn baby was fine.

CHAPMAN: I still didn't believe it. I just didn't know what to believe anymore. Like I knew but I was afraid to believe it.

SNOW: How long did it take you to believe?

CHAPMAN: Until they put him in my arms and they lifted him and swarm above the screen and he drooled on me. I just couldn't believe he was ours.
SNOW: These prenatal DNA tests given as early as ten weeks are marketed for their accuracy. The maker of the test Stacie used says in high risk women, a positive result for Edwards Syndrome is right 97.6% of the time, but several recent medical studies and an investigation by the New England Center for Investigative Reporting cast doubt on the reliability of prenatal DNA blood tests when they have a positive result for a disease.

CHAPMAN: Everyday gets better.

SNOW: A study last year found that a positive result on one popular test, not the one Stacie used, could be wrong more than half of the time.

DR. MICHAEL F. GREENE (Massachusetts General Hospital): When is your due date actually?

SNOW: Leading OB-GYN Doctor Michael Green says tests like the one Stacie took are looking for the risk of disease, not actually diagnosing that a child will be born with it.

DR. GREENE: Before a woman makes an irrevocable decision, she shouldn't make it based only on a screening test. It must be based on the results of a diagnostic test.

SNOW: The maker of the test Stacie took agrees, saying the test is not a replacement for a diagnostic test such as CVS or amniocentesis later in pregnancy. And like some other manufacturers recommends women have genetic counseling and the results be confirmed. Stacie and Lincoln's boy, Sam, is a happy, healthy, eighteen-month-old. Do you think about what might have happened?

LINCOLN CHAPMAN: From time to time, you know, when he is playing or he does something funny, we'll say, can you believe it? He's here and, you know, and almost wasn't.

SNOW: The sweet face of a little boy showing the limits of the brave new world of genetic testing. Kate Snow, NBC News, Providence, Rhode Island.