DNA testing fetus leads moms to their own cancer diagnoses

Last year 800,000 woman used non-invasive prenatal testing to find out if their fetuses had genetic disorders like Down Syndrome. And 26 of them also discovered they had cancer.

Non-invasive prenatal testing (NIPT) analyzes DNA from a blood sample taken from a mom-to-be. Up to 10 percent of the genetic material circulating in a pregnant woman's blood stream comes from her fetus. Fetal cells pass through the placenta and into a mother's bloodstream. These genetic tests can identify the non-maternal DNA and check it to make sure it has the right number and quality of chromosomes meaning the baby will be free of genetic disorders.

Tumors also shed their DNA into the bloodstream, although less than a fetus. And cancer cells often have major chromosomal abnormalities that can be identified, just like Downs Syndrome, by a NIPT test.

Eunice Lee took an NIPT test at 10 weeks pregnant Virginia Hughes at BuzzFeed reports. Her results indicated she might have cancer:

> My OB walked into my room and was holding the sheet of paper in her hand, Lee, an anesthesiologist in Santa Barbara, California, told BuzzFeed News. The doctor had just hung up the phone with Sequenom Laboratories, the San Diego company that performs the test. "The director of the laboratory called my obstetrician and told her I needed to be worked up for cancer," Lee said, "which was just alarming, to her and also to myself, because I had no idea I had cancer."

The Sequenom test that Lee used and others like it can't definitively tell if a person has cancer. She must undergo further testing to discover if there is a tumor or other pathology. Lee, for example, opted for a full body MRI scan that identified a tumor in her colon. She had surgery and subsequently delivered a healthy baby boy.

But Sequenom and other companies are working on similar blood tests that can identify the genetic signatures of cancers left in the bloodstream and, in some cases, even identify the type and severity of the disease. These tests could replace biopsies, the traditional tissues tests used to track cancers Ed Yong reported in Nature:

> By developing and refining techniques for measuring and sequencing tumour DNA in the bloodstream, scientists are turning vials of blood into 'liquid biopsies' — portraits of a cancer that are much more comprehensive than the keyhole peeps that conventional biopsies provide. Taken over time, such blood samples would show clinicians whether treatments are working and whether tumours are evolving resistance.

Because circulating tumor DNA changes every 2 hours instead of weeks like traditional protein-based cancer biomarkers, a liquid biopsy approach can offer almost real time data about how the cancer is

responding to treatment or if it has developing further mutations that require different drugs. In Lee's case, a second NIPT test showed now evidence of tumor DNA in her blood after surgery so she decided against chemotherapy.

Getting a cancer diagnosis from a pregnancy test might be the ultimate 'incidental finding.' Reporting these unexpected results to patients is a hot topic in the ethical practice of genomic medicine. The FDA has not yet solidified guidelines on what should be reported to patients or how and if patients can chose what they want to know. Beyond diagnosing a cancer, incidental findings can include unexpected paternity results or high risk for other cancers or neurological diseases, Tabitha Powledge writes at the GLP.

In these cases of cancers caught through NIPT, the incidental findings relate to a completely different person (mom) than the test is targeting (baby), Hughes writes:

> "The FDA is struggling to figure out how to regulate these things. Everybody's screaming and yelling about what it's going to look like," Laura Hercher, a genetic counselor at Sarah Lawrence College said. On the other hand, if they didn't release the results, that could cause the women harm. "It's really hard, ethically, to make the case that you should sit on this information," Hercher said.

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Additional Resources:

- What should be done with unsettling 'incidental findings' in gene screens? Genetic Literacy Project
- Cancer genomes to enter private practice, Genetic Literacy Project
- It's time for a cancer genomics revolution, Genetic Literacy Project