

Abnormal Prenatal Blood Test Results Could Indicate Hidden Maternal Cancers

NIH study finds some abnormal results due to colorectal, breast, lung and pancreatic cancers.

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Researchers at the National Institutes of Health (NIH) found previously undetected cancers in 48.6% of pregnant people who had abnormal results for prenatal cell-free DNA ([cfDNA](#)) testing used to screen for chromosomal disorders in the fetus. Cancers included colorectal, breast, lung and pancreatic cancers, as well as lymphoma, cholangiocarcinoma and renal carcinoma.

The screening test analyzes placental DNA fragments circulating in the maternal bloodstream to identify an extra chromosome or to determine the baby's sex. The study appears in the [New England Journal of Medicine](#).

In addition to fetal DNA, cfDNA testing detects DNA released from the mother's red blood stem cells and, occasionally, abnormal DNA that may result from an undetected cancer in the asymptomatic pregnant person.

NIH's ongoing [IDENTIFY study](#) seeks to learn more about abnormal cfDNA test results that could indicate potential cancers. For the current analysis, researchers performed cancer screening of 107 IDENTIFY participants using whole body magnetic resonance imaging (MRI), standard medical diagnostic tests, and cfDNA sequencing.

A total of 52 participants were diagnosed with cancer. For this group, the authors found that whole body MRI was the most effective method for detecting cancer. Standard diagnostic techniques, such as taking a medical history, assessing symptoms, and physical examination, were of limited use in identifying a participant's cancer or its location.

Other abnormal cfDNA results were attributed to fibroids (benign uterine tumors), placental chromosomes that differed from fetal chromosomes, and clonal hematopoiesis in the mother (a precancerous state that can lead to blood cancers).

The researchers noted the need for additional studies to validate cfDNA sequencing patterns described by the investigators that could indicate cancer in this young, pregnant population

without obvious clinical symptoms.

The study was conducted by researchers in NIH's National Human Genome Research Institute, National Cancer Institute, and Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD).

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