New research discovers blood test to prenatally identify dangerous fetal heart defects

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Researchers at Beaumont Hospital in Royal Oak, Michigan have discovered a blood test that uses artificial intelligence and genetic-

related biomarkers in pregnant women to accurately detect fetal congenital heart defects well before birth.

"We know that when congenital heart defects are diagnosed early—ideally before birth—outcomes can improve significantly and mortality and morbidity [can be] reduced," said Dr. Ray Bahado-Singh, system chief of Obstetrics and Gynecology, Beaumont Health, and lead author of "Accurate Prenatal Detection of Fetal Congenital Heart Defects," which appears in a recent issue of *American Journal of Obstetrics and Gynecology*.

Bahado-Singh, his team from Beaumont and researchers from three other institutions harnessed the power of artificial intelligence to identify and evaluate fetal DNA that circulates in the mother's bloodstream.

Dr. Bahado-Singh cautions that larger prospective studies are needed to validate these findings. However, he said this minimally invasive detection method is an important first step in establishing an effective postnatal action plan that quickly puts at-risk infants on track to receive the intensive medical and surgical attention they need.

"Once confirmed, these results could lead to exciting new protocols, and most importantly, improved outcomes for newborns and their families," Dr. Bahado-Singh said. "The next steps after a positive test would include performing an echocardiogram prenatally and repeated[ly] after birth to confirm the presence and nature of a cardiac defect."

Currently, ultrasound that images the fetal heart is the only available screening tool for early (prenatal) detection of congenital heart defects, Dr. Bahado-Singh said. Unfortunately, in the United States, only about half of fetal congenital heart defects are identified on prenatal ultrasound. Because of this limitation, it is now the standard of care to screen and monitor newborn oxygen levels via pulse oximetry.

Unfortunately, pulse oximetry still misses about 10% of critical newborn heart defects, leading to increased deaths and severe complications.

Birth defects, particularly those that stem from heart disease, are a leading cause of infant mortality. These include holes in the heart, the most common, and other potentially deadly cardiac birth defects that might affect oxygen levels and blood flow at birth; valve abnormalities and others. Smoking, alcohol use, environmental toxins and vitamin deficiency are all known contributors to the development of congenital heart defects. These agents cause chemical (epigenetic) changes in DNA, which can affect the function of genes critical to the development of the heart, Dr. Bahado-Singh explained, by turning genes on or off.

He continued, "You need a certain group of genes turned on and others turned off, for example, to make sure the chambers of the heart are fully developed. Switching the wrong genes on or off in this normally perfectly orchestrated symphony can cause maldevelopment, leading to major heart defects."

Artificial intelligence analysis of the circulating (including fetal) DNA extracted from the mother's blood, Dr. Bahado-Singh explained, "enables us to efficiently review potentially billions of pieces of information in the genome. This includes swiftly identifying specific predictors of a possible fetal heart defect and separating those that need continued monitoring."

Ultimately, Dr. Bahado-Singh would like to see all pregnant women achieve ready access to screening through a blood test, which requires no advance appointments or significant time off from work. Those who test positive could then be referred for detailed fetal and newborn cardiac ultrasounds and appropriate early intervention, as needed.

"We are still a ways away from that," he said. "But it's exciting to

contemplate the possibilities."

More information: Ray Bahado-Singh et al, Cell-free DNA in maternal blood and artificial intelligence: accurate prenatal detection of fetal congenital heart defects, *American Journal of Obstetrics and Gynecology* (2022). DOI: 10.1016/j.ajog.2022.07.062

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