Diana W. Bianchi [1]

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Diana W. Bianchi studied the medical treatment of premature and newborn infants in the US during the twentieth and twenty-first centuries. Bianchi helped develop non-invasive prenatal genetic tests that use cell-free fetal DNA found within maternal blood to diagnose genetic abnormalities of the fetus [3] during pregnancy [4]. The test provides a means to test fetuses for chromosomal and genetic abnormalities.

Bianchi earned her undergraduate degree in biology in 1976 from the University of Pennsylvania [5] in Philadelphia, Pennsylvania. She then attended Stanford University [6] School of Medicine in Stanford, California, where she graduated with her medical degree in 1980. While at Stanford, Bianchi researched different methods for detecting fetal cells in maternal blood. The primary method for cell detection is fluorescence-activated cell sorting, a technique that uses light to separate mixed biological samples, or samples of varied human cells, one cell at a time. She completed her medical residency at Children's Hospital Medical Center in Boston, Massachusetts, in 1986 while also researching fluorescence-activated cell sorting as a method for fetal cell detection. She began work as a medical geneticist at Boston Children's Hospital in 1986.

While at Boston Children's Hospital, Bianchi worked within the Genetics and Newborn Medicine Department. In 1990, she and collaborators isolated fetal DNA from fetal red blood cells found in the blood samples of pregnant women. They published their results in "Isolation of fetal DNA from nucleated erythrocytes in maternal blood". As the placenta [7] serves as a two-way membrane between the mother and fetus [3], several types of fetal cells cross the barrier and circulate in the mother's blood, including the precursors to placental cells (trophoblasts), red blood cells (erythrocytes), and white blood cells (leukocytes). Gordon Douglas and teams of researchers at Bellevue and New York Universities in New York, New York, isolated trophoblasts created by the fetus [3] in maternal blood as early as 1959, but fetal red blood cells were not isolated or genetically analyzed until 1990, when Bianchi and her team isolated and analyzed them.

To isolate fetal red blood cells from maternal blood, Bianchi and her team used blood samples from nineteen pregnant women 12.5 to 17 weeks after fertilization [8]. From each 20 milliliter sample, Bianchi extracted 0.1 to 1 nanograms (ng), which is one billionth of a gram. She then used a method of DNA amplification called polymerase chain reaction (PCR) to increase a specific DNA sequence only found on the Y-chromosome in the fetal DNA.

Bianchi's research on fetal cells in maternal plasma enabled the development of non-invasive prenatal genetic testing. Bianchi's research built on previous prenatal testing techniques, which became available in 1967 with the introduction of amniocentesis, a method for removing a sample of the fluid surrounding the fetus [3] (amniotic fluid) for genetic testing. Another technique was chorionic villus sampling, a method for removing a sample of placenta [7] for genetic testing. Researchers label both of those methods as invasive prenatal genetic

testing, as they require the injection of a hollow needle into the fetal environment, which carries some risk to the fetus [3] and a smaller risk to the mother. Scientists found that both of those tests, amniocentesis and chorionic villus sampling, are associated with increased rates of spontaneous abortions in women who undergo them. Bianchi's technique, called maternal fetal cell sorting, recovered fetal DNA from maternal blood and tested it for genetic abnormalities from a blood sample. Researchers considered taking a blood sample as a generally safe procedure with little to no risk of spontaneous abortions due to prenatal testing.

Though Bianchi said that she hoped her technique would enable the development of more non-invasive prenatal tests, maternal fetal cell sorting was limited by the relatively small quantity of fetal cells found in maternal blood. She left her position at Boston Children's Hospital in 1993 and moved to the Tufts Medical Center in Boston.

In 1996, Bianchi and her team found that progenitor cells from male fetuses remain in their mothers' blood for decades after birth. Progenitor cells refer to hematopoietic stem cells [9], which are fetal cells that can develop or differentiate into multiple blood cell types. Bianchi used samples from women carrying male fetuses because it was easier to identify male DNA within a female blood sample due to the difference in sex chromosomes between males and females. The scientists included three categories of women in the study: women currently pregnant with male fetuses, women who previously carried male fetuses to term, and women who had previously terminated pregnancies of male fetuses. Bianchi found that the male fetal cells persisted in the women's bloodstreams for potentially decades after their pregnancies.

In 2004, Bianchi, along with Kiarash Khosrotehrani, a researcher at the University of Queensland in Brisbane, Australia, discovered that the male fetal cells left in maternal blood after pregnancy [4] could differentiate into multiple cells types under the influence of the maternal environment. The researchers found fetal cells in several different maternal tissues, including the lymph nodes, the liver, and the cervix [10]. All of those fetal cells were blood cells. However, depending on the tissue in which the scientists found the cells, they showed other cell type characteristics. For example, those fetal cells found in the liver tissue had liver cell characteristics. The scientists also found fetal cells in high concentrations surrounding the diseased tissues of women with severe diseases such as cancer. The researchers theorized that the high concentration of fetal cells surrounding diseased maternal tissue might be an immune response by the fetal cells, and they called for further research to test the theory.

After 2004, the NIH funded Bianchi for greater than a decade to study the transfer of fetal cells into pregnant women. Bianchi was one of four authors on the book *Fetology: Diagnosis and Management of the Fetal Patient* in 2000. In 2002, Bianchi became an honorary member of the Society for Maternal-Fetal Medicine, and in 2006, an honorary member of the American Pediatric Surgical Association.

In 2010, Bianchi became the executive director of the Mother Infant Research Institute (MIRI) at part of Tufts Medical Center, with the goal to develop non-invasive prenatal diagnosis [11] and treatment. In 2016, the US National Institutes of Health [12] named Bianchi the director of the National Institute of Child Health and Human Development, headquartered in Rockville, Maryland.

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