

Predicting IVF success with genetic testing

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A researcher at Albert Einstein College of Medicine of Yeshiva University has helped to develop the first genetic blood test for predicting the chances that in vitro fertilization (IVF) will lead to a successful pregnancy. The test, reported in the online medical journal *PLoS One*, is based on the finding that different subtypes of the FMR1 gene (also known as the fragile X mental retardation gene) in potential mothers are associated with significantly different chances of conceiving with IVF.

“This is the first evidence that a specific gene appears to be directly associated with IVF outcomes,” said David Barad, M.D., associate clinical professor of epidemiology & population health and of obstetrics & gynecology and women’s health at Einstein and one of the study’s senior authors. Dr. Barad is also director of assisted reproduction at the Center for Human Reproduction (CHR) in New York City.

“Our research also suggests the FMR1 gene, some forms of which are known to predict premature ovarian failure, could be used to predict at what age a woman’s fertility is going to start decreasing,” he added.

The study also supports the belief that autoimmunity (immunity to one’s own cells or tissues) plays a role in infertility – a controversial topic among reproductive medicine specialists.

The study involved 339 female infertility patients who underwent a total of 455 IVF cycles at CHR. The researchers investigated the relationship between three different FMR1 genotypes and pregnancy outcomes and

autoimmunity levels. Women with the “normal” FMR1 genotype had a 38.6 percent pregnancy rate; those with the “heterozygous-normal/high” genotype had a 31.7 percent pregnancy rate; and women found to have the “heterozygous-normal/low” genotype had a 22.2 percent pregnancy rate.

The genotype associated with the lowest [pregnancy](#) rate (heterozygous-normal/low) was also associated with increased measures of autoimmunity. Women with this genotype also had a higher incidence of polycystic ovary syndrome (a common cause of infertility), which is thought to have an autoimmune component. “Previous studies have suggested that autoimmunity plays a role in infertility,” said Dr. Barad. “Now, for the first time, we have a potential genetic mechanism that underlies several different threats to infertility.”

The cost of the [blood test](#) for the FMR1 should be relatively low – comparable to screening tests for Tay-Sachs and other genetic diseases. While the [FMR1 gene](#) test is not yet clinically available, “It’s likely that the findings will lead to clinical applications in the future,” said Dr. Barad.

“Any test that is proven to have predictive value for a woman’s fertility would give her a heads up in terms of planning a family,” he added. “For instance, if a woman planning to go to law school or medical school learns she has a certain amount of risk of losing her ovarian function before she is 35, she may choose to bank her eggs or try having children at an earlier age, rather than delay.”

More information: Dr. Barad’s paper, “FMR1 Genotype with Autoimmunity-Associated Polycystic Ovary-Like Phenotype and Decreased Pregnancy Chance,” was published in the December 2010 issue of the online journal *PLoS One*.

Provided by Albert Einstein College of Medicine

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