Magee-Womens
Research Institute

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Determinants of Early Embryonic Development

Goal: The goal of this research study is to investigate possible genetic determinants of early embryo development that may be contributing to infertility in couples with a diagnosis of unexplained infertility.

Study overview:

Almost 30% of couples with infertility carry the diagnosis of unexplained infertility. Rather than unexplained, it might be due to problems during early embryo development. In vitro fertilization (IVF) provides a unique opportunity to identify cases where the possible problem may be with development of the embryo. We know that early on, maternal genes play a large role in embryo development and then later on, the embryo's own genes take over. A handful number of maternal effect genes are known[1], while many more genes remain to be determined. We hope to learn more about the maternal and paternal genes that may be involved in this complex process.

This study will look at couples who do not have a known cause of infertility and subsequently may or may not go through the process of in vitro fertilization (IVF). Couples who have problems with the progression of multiple embryos to the blastocyst stage and those whose treatment did not result in clinical pregnancy are eligible. This research is being conducted by the University of Pittsburgh.

Qualifications for the Study:

- Diagnosis of infertility, inability to conceive
- No known maternal or paternal cause of infertility
- Maternal age < 40
- History of unsuccessful infertility treatment

What will take place in the study?

- Enrollment of women and their male partners
- Medical, family history and pedigree collection
- One-time blood sample collection which will only be about 10cc (in a purple top tube)
- Genetic analysis will be performed on DNA samples from both partners

Outcomes:

There is no guarantee relevant information will be found. We will likely not be able to use any information that is found for treatment purposes. It will be used to advance scientific knowledge for possible future use.

References:

1. Yatsenko, SA, and Rajkovic A. "Genetics of human female infertility." *Biology of reproduction* 101.3 (2019): 549-566.

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