Magee-Womens
Research Institute

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Genetics of Recurrent Pregnancy Loss

Goal: The goal of this research study is to investigate possible genetic causes of recurrent pregnancy loss (RPL)

Study overview: The potential RPL etiologies include endocrine and autoimmune factors; fetal and placental infections; anatomical defects; environmental, occupational, personal habits, and genetic factors [1]. Up to 50% of patients with RPL have no clearly defined etiology. This study focuses on investigation of genetic causes of RPL that includes recurrent chromosomal abnormalities, unbalanced chromosomal aberrations resulting from parental balanced rearrangements, parental genetic mutations affecting fetal viability or ability to carry pregnancy.

Genetic evaluation of miscarriage tissue obtained at the time of the second and subsequent pregnancy losses is recommended as a part of clinical evaluation and should be offered to all couples with two or more consecutive pregnancy losses. The combination of a genetic evaluation on miscarriage tissue with an evidence-based assessment for RPL will identify a probable or definitive cause in over 90% of miscarriages. Chromosome microarray analysis adds significantly to RPL assessment [2]. This research study is being conducted by the University of Pittsburgh.

Qualifications for the Study:

- Family with RPL of unexplained cause
- Experienced 2 or more losses that have no discernible cause
- Women of reproductive age up to 37
- Currently not pregnant
- Participation of both partners are highly encouraged to achieve informative results (Please contact study coordinator for eligibility if only one partner is available)
- Cytogenetic evaluation on miscarriage tissue and its availability for a further genetic testing is desired. (Please request fresh tissue storage upon submission for cytogenetic testing)

What will take place in the study?

Once a woman and her partner have consented to participate, we will collect a blood sample from them. This blood sample will only be about 10cc (1 purple top tube). This will be used to obtain DNA from the parents. The parents will also give consent for previous products of conception to be included in the study. These materials have previously been collected for clinical purposes and we will only use them once clinical testing has been completed. We will also collect a family history and family pedigree from the parents.

Outcomes:

We may not find a definitive cause for your losses. The goal of the study is to improve the overall diagnostic tools that are used in recurrent pregnancy loss cases. By studying patients with unknown causes we may discover new ways to identify a cause.

References:

1. https://www.ncbi.nlm.nih.gov/books/NBK554460/
2. Popescu F, Jaslow CR, Kutteh WH. Recurrent pregnancy loss evaluation combined with 24-chromosome microarray of miscarriage tissue provides a probable or definite cause of pregnancy loss in over 90% of patients. Hum Reprod. 2018;33(4):579-587. PMID: 29538673.

Study Coordinator Contact info: Jessica Settino; email: settinoj@upmc.edu or phone: 412-641-7457