



## Recurrent Pregnancy Loss<sup>©2005</sup>

Pregnancy loss can result from genetic causes, non-genetic causes, or a combination of both. Detailed pregnancy, personal, and family medical histories assist in determining and prioritizing evaluations which will be the most informative. A thorough assessment may involve specialties in addition to genetics, such as maternal fetal medicine, reproductive endocrinology, gynecology, urology, and psychology. In the event of pregnancy loss in the second or third trimester, it is recommended that a detailed external physical examination be conducted by a geneticist. An autopsy by a pathologist experienced in fetal evaluation should be performed if possible. Further genetic studies may also be indicated based on these evaluations.

Non-genetic causes may include environmental factors such as chemical exposure, smoking, or alcohol use during pregnancy. Uterine abnormalities such as a bicornate uterus or fibroids increase the risk of pregnancy loss, as can an incompetent cervix. Uncontrolled diabetes and maternal immunologic disorders are associated with recurrent pregnancy loss. Other maternal health problems, such as infections, are suspected but have not been proven to cause recurrent losses.

Genetic causes of pregnancy loss can include extra or missing chromosomes in the fetus, or structural rearrangements of the chromosomes. Each of these problems can create too much or too little genetic material in the fetus, and increase the risk of pregnancy loss. Harmful changes in the genes, located on the chromosomes themselves, can also cause pregnancy loss.

Roughly 60% of first trimester miscarriages are due to chromosomal abnormalities in the fetus, the majority of them happening by chance. Extra chromosome conditions, such as Down syndrome, generally happen at random, and the recurrence risk is usually equal to the maternal age risk or just slightly higher. However, for 5% of couples with recurrent pregnancy loss, one parent carries a balanced rearrangement of his/her own chromosome material. That parent has no health problems, but he/she has an increased chance of a conception with an unbalanced rearrangement. This can cause recurrent pregnancy loss or a baby being born with physical and developmental problems.

We discussed the importance of studying the chromosomes of both members of the couple. If their studies are normal, their risk of having a child with a chromosome abnormality would be the mother's age risk. If a chromosome rearrangement is found in either parent, the risk of a child with a chromosome abnormality is usually increased, although a healthy pregnancy outcome may still be possible. Another genetic counseling session would be scheduled to discuss any specific chromosome rearrangement found. Your doctor will receive the chromosome results approximately 10-14 days after the blood sample is received at Emory Genetics Laboratory.