



Recurrent Pregnancy Loss

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Pregnancy loss can result from many causes, including 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 including genetics, 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 of recurrent pregnancy loss 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 as well. Other maternal health problems, such as infections, may also contribute to 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 50-60% of first trimester miscarriages are due to chromosomal abnormalities in the fetus, the majority of which happen by chance. Conditions in which there is an extra chromosome, such as Down syndrome, generally happen at random, and the recurrence risk is usually equal to or just slightly higher than the maternal age-related risk. However, for 4-6% 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. Blood chromosome analysis is available for couples experiencing recurrent pregnancy loss to rule out a balanced chromosomal rearrangement in them.