

## Why perform chromosomal analysis in abortion material?

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Chromosome anomalies are the major cause of recognized early gestational loss. In up to 60% of early pregnancy loss, chromosomal anomalies are detected, and up to 30% are found in late abortions. Of all chromosomal changes, 95% are of numerical and 3-5% are of structural nature.

The frequencies of anomalies are distributed as follows: Trisomies represent about 50% of all abnormalities, followed by triploidies and monosomies. Tetraploidies are reported in around 5% and structural changes account for about 3-5%.

Chromosomal analysis in abortion material should be performed to assess the cause for the abortion and to assess a potential risk for further pregnancies. If the chromosomal analysis reveals for example an unbalanced chromosomal anomaly, such as a translocation, a balanced parental chromosomal anomaly should be verified, as this could increase the risk for further abortions.

Unfortunately, karyotyping does not always lead to a result. Maternal cell contamination and/or no further growth of cells are the main reasons. DNA-based technologies such as array CGH and multiplex ligation probe amplification (MLPA) are useful alternatives in these cases.

This lecture will give an overview on anomalies in abortion material. Indications for investigations and the potential of newer techniques such as DNA amplification based methods (e.g. array CGH) are discussed.

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