Abstract

Cell free DNA is short, fragmented DNA found in plasma and serum, into which it is released by various mechanisms. It was first identified in body fluids of cancer patients and soon became a biomarker for non-invasive diagnostics. Soon after, the presence of fetal cfDNA in maternal plasma was detected and became the subject of clinical utility. Non-invasive prenatal diagnosis is a screening method that helps to determine the likelihood of birth defects early in pregnancy, specifically using fetal cfDNA. The final results are determined by invasive methods such as aminocentesis and chorionic villus sampling. Invasive methods of prenatal diagnosis carry a risk of fetal loss of less than 1 %. Despite this low risk, scientists are constantly striving to improve non-invasive methods that can detect aneuploidies such as Down's or Edwards syndrome, as well as determine the sex of the fetus, monogenic diseases and the Rh factor. Polymerase chain reaction (PCR) or next-generation sequencing methods are used for detection.