

Cell-free nucleic acids as non-invasive biomarkers of gynecological cancers, ovarian, endometrial and obstetric disorders and fetal aneuploidy

Abstract

BACKGROUND

Proper folliculogenesis is fundamental to obtain a competent oocyte that, once fertilized, can support the acquisition of embryo developmental competence and pregnancy. MicroRNAs (miRNAs) are crucial regulators of folliculogenesis, which are expressed in the cumulus–oocyte complex and in granulosa cells and some can also be found in the bloodstream. These circulating miRNAs are intensively studied and used as diagnostic/prognostic markers of many diseases, including gynecological and pregnancy disorders. In addition, serum contains small amounts of cell-free DNA (cfDNA), presumably resulting from the release of genetic material from apoptotic/necrotic cells. The quantification of nucleic acids in serum samples could be used as a diagnostic tool for female infertility.

METHODS

An overview of the published literature on miRNAs, and particularly on the use of circulating miRNAs and cfDNA as non-invasive biomarkers of gynecological diseases, was performed (up to January 2014).

RESULTS

In the past decade, cell-free nucleic acids have been studied for potential use as biomarkers in many diseases, particularly in gynecological cancers, ovarian and endometrial disorders, as well as in pregnancy-related pathologies and fetal aneuploidy. The data strongly suggest that the concentration of cell-free nucleic acids in serum from IVF patients or in embryo culture medium could be related to the ovarian hormone status and embryo

quality, respectively, and be used as a non-invasive biomarker of IVF outcome.

CONCLUSIONS

The profiling of circulating nucleic acids, such as miRNAs and cfDNA, opens new perspectives for the diagnosis/prognosis of ovarian disorders and for the prediction of IVF outcomes, namely (embryo quality and pregnancy).