02 - MEDICINA DELLA RIPRODUZIONE

THE GENETICS OF NON-SYNDROMIC PRIMARY OVARIAN INSUFFICIENCY: A SYSTEMATIC REVIEW

Obiettivo: Several causes for primary ovarian insufficiency have been described, including iatrogenic and environmental factor, viral infections, chronic disease as well as genetic alterations. Given the large number of genes described in the literature so far, the aim of this review was to collect all the genetic mutations associated with non-syndromic primary ovarian insufficiency.

Metodi: All studies assessing genetic mutations associated with non-syndromic primary ovarian insufficiency were analyzed. For a study to be included in this review, it had to focus on candidate gene screening, or a genome-wide study. Syndromic primary ovarian insufficiency and chromosomal abnormalities were not evaluated.

Risultati: Single gene perturbations having positive correlation with non-syndromic primary ovarian insufficiency included genes on the X chromosome, such as BMP15, PGRMC1, and FMR1; and genes on autosomes, such as GDF9, FIGLA, NOBOX, ESR1, FSHR, and NANOS3. New strategies performed for identifying new genes associated with primary ovarian insufficiency development included linkage analysis in families with multiple affected members, CGH for copy number variations, genome-wide association studies, genome-wide sequencing of exomes, and in the future the next generation sequencing.

Conclusioni: This review, including nearly all the genetic abnormalities and genes associated with non-syndromic primary ovarian insufficiency detected so far, showed the variability of genetic factors in the origin of primary ovarian insufficiency, as ovarian function depends on the expression of multiple genes. These findings may help for future genetic screening studies on large cohort of women.