

Polymorphisms and Premature Ovarian Failure (POF)

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Premature ovarian failure (POF) refers to the cessation of ovarian function before the age of 40 years with normal growth and development. POF occurs in approximately 1-2% of the female population. Etiology and pathophysiology of POF have not been identified. However, the known causes include genetic aberrations, autoimmune ovarian damage and iatrogenic intervention. Degenerative process in the ovary is controlled primarily by apoptosis, or programmed cell death.

In this study, we hypothesized that development of POF is associated with sequence variation, SNP (single nucleotide sequence) in hypervariable regions HV1 and HV2 of the mitochondrial DNA. Peripheral blood samples of genomic DNA were collected from 99 patients and from 80 healthy fertile women. We found that POF samples contained polymorphisms significantly differing from those of normal control. These results suggest that the identified SNPs in hypervariable regions may affect the development of POF caused by abnormality in mitochondria such as impaired respiratory function and accelerated apoptosis.