

9th Yazd International Congress and Student Award on Reproductive Medicine with 4th Congress of Reproductive Genetics

Key Lectures

K-86

The genetics of premature ovarian failure: Current perspectives

Shelling A.

Department of Obstetrics and Gynaecology, Faculty of Medical and Health Sciences, University of Auckland, Auckland, New Zealand.

Email: a.shelling@auckland.ac.nz

Premature ovarian failure (POF) is a common cause of infertility in women, characterised by amenorrhea, hypoestrogenism, and elevated gonadotropin levels in women under the age of 40. Many genes have been identified over the past few years that contribute to the development of POF. However, few genes have been identified that can explain a substantial proportion of cases of POF. The unbiased approaches of genome wide association studies and next generation sequencing

technologies have identified several novel genes implicated in POF. As only a small proportion of genes influencing idiopathic POF have been identified thus far, it remains to be determined how many genes and molecular pathways may influence idiopathic POF development.

However, due to POF's diverse etiology and genetic heterogeneity, we expect to see the contribution of several new and novel molecular pathways that will greatly enhance our understanding of the regulation of ovarian function. Future genetic studies in large cohorts of well defined, unrelated, idiopathic POF patients will provide a great opportunity to identify the missing heritability of idiopathic POF. The identification of several causative genes may allow for early detection and would provide a better opportunity for early intervention, and furthermore, the identification of specific gene defects will help direct potential targets for future treatment.