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Juneau Biosciences presents new genetic discoveries in endometriosis at annual ASRM meeting

Published on October 23, 2015 at 3:36 AM

This week, Juneau Biosciences, LLC presented new discoveries concerning the genetics of endometriosis at the annual American Society of Reproductive Medicine (ASRM) meeting in Baltimore, MD. The ASRM is an international and multi-disciplinary organization devoted to advancing knowledge and expertise in reproductive medicine and biology, with a particular focus on human infertility. In an oral presentation, Dr. Kenneth Ward, M.D., Juneau's founder and CEO, described several "major gene" effects associated with endometriosis discovered through DNA sequencing of affected women. The ASRM Scientific Program Committee recognized this work with an award: the "Endometriosis Special Interest Group Prize Paper".

Endometriosis is a common (but under-diagnosed) disease that affects up to ten percent of women. The condition, characterized by the deposition and growth of endometrial tissue outside the uterus, involves symptoms of reduced fertility and pain. Even mildly and moderately affected individuals often experience a reduced quality of life, and endometriosis commonly requires surgical treatment. U.S. health care costs related to endometriosis are estimated to exceed \$100 billion annually; Dozens of studies have shown that genetic factors contribute to both the occurrence and the severity the disease.

Juneau's study used the latest DNA sequencing techniques to study hundreds of affected women. Serious mutations were found in DNA samples from women who have endometriosis compared to women drawn from the general population. Most of the genes with endometriosis related mutations had not been studied before in this context; the function of several is completely unknown. Several of the implicated genes are known to function in the human immune system. Many scientists believe that a faulty immune system may cause endometriosis by failing to find and destroy endometrial tissue growing outside of the uterus. Women who have endometriosis are also more likely to develop autoimmune disorders in which a patient's immune system attacks her own body's tissues.

Juneau is studying the genetic mechanisms that contribute to endometriosis with the aim of developing DNA-based tests to predict and to diagnose endometriosis. It is difficult to determine whether a woman has endometriosis based on her symptoms, physical examination, or imaging studies such as ultrasound. Definitive diagnosis requires a surgical procedure such as laparoscopy, but the sensitivity and specificity of laparoscopy is not 100%. Due to the risk, invasiveness, and expense of diagnosing the condition, women with endometriosis often suffer for over a decade or more before being correctly diagnosed.

Source:

Juneau Biosciences, LLC
