



## PRESS RELEASE

**Release Date:** FOR IMMEDIATE RELEASE  
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### IMMUNITY GENE IMPLICATED IN ENDOMETRIOSIS

#### First "Major Gene" Effect in This Common and Enigmatic Gynecologic Disease

SALT LAKE CITY, UT, October 13, 2015 – This week, Juneau Biosciences, LLC presented new discoveries concerning the genetics of endometriosis at the American Society of Human Genetics meeting in Baltimore, MD. Dr. Kenneth Ward, M.D., Juneau's founder and CEO, described the first "major gene" effect associated with endometriosis. Juneau scientists found damaging mutations in the *NLRP2* gene in one out of every eleven endometriosis patients studied. These mutations appear to increase the risk of developing endometriosis by as much as ten-fold.

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.

Juneau's study, based on the detailed genetic analysis of over 5,000 women, found an excess of *NLRP2* mutations in DNA samples from women who have endometriosis compared to women drawn from the general population. The *NLRP2* (an abbreviation for *NACHT*, *LRR* and *PYD* domains-containing protein 2) protein plays a role in our body's "innate" immune system. The mutations seen in the women with endometriosis are likely to interfere with a women's 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.

The *NLRP2* gene is expressed by white blood cells, by pre-implantation embryos, by female reproductive organs, and by endometriosis lesions. In most tissues, the function is unknown, and there have been no prior biologic studies in endometriosis. We know that the *NLRP2* gene is one of a few hundred "imprinted" genes in humans. The copy of the *NLRP2* gene we inherit from our fathers is "silent", and only the version we inherit from our mother is an "active" gene producing *NLRP2* proteins. Previously, mutations in the *NLRP2* gene have been associated with rare imprinting and reproductive disorders

Juneau is studying the genetic mechanisms that contribute to endometriosis with the aim of developing DNA-based tests to assess a woman's risk for endometriosis.

## ABOUT ENDOMETRIOSIS

Endometriosis affects more than 10 million women in the United States. It occurs when tissue similar to the lining of the uterus (womb) appears in other parts of the body, most commonly in the pelvis. Monthly bleeding and inflammation caused by these lesions may severely impact a woman's quality of life. Some affected women experience severe pain, others infertility, others problems with their menstrual periods, and some have no symptoms at all. Today, definitive diagnosis requires surgery. Due to the risk, invasiveness, and expense of diagnosing the condition, women with endometriosis often suffer for over a decade before even being correctly diagnosed. Treatment may involve hormonal suppression or surgical destruction of the abnormal tissue.

## ABOUT JUNEAU BIOSCIENCES, LLC

Juneau Biosciences is a privately-held medical genetics company. Launched in 2007, the Salt Lake City, Utah-based firm has focused on the development of novel molecular diagnostics and therapeutics for endometriosis, a condition that affects up to 10 percent of reproductive-age females. Juneau intends to improve the lives of millions of women through genetic research and innovative health care solutions. Juneau Biosciences' team of experts, state-of-the-art facilities, extensive knowledge-base and innovative research techniques place Juneau at the scientific forefront of developing diagnostics and treatments for women.