



FACULTY OF MEDICINE



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MEDIA RELEASE

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British Columbian Researchers uncover new ovarian cancer gene: a connection between cancer and endometriosis

Vancouver, Canada. - Researchers with the Ovarian Cancer Research Program of BC (OvCaRe) reveal a major new cancer gene—ARID1A. Mutations are frequent in this gene and link two types of ovarian cancer to endometriosis.

The research paper, *ARID1A Mutations in Endometriosis-Associated Ovarian Carcinomas*, published today in the *New England Journal of Medicine*. Researchers looked at over 600 samples of ovarian cancer, leading to the conclusion that the ARID1A mutation and loss of function is believed to be an early event in the transformation of endometriosis into clear-cell and endometrioid cancer.

The ARID1A mutations were found in 46 per cent of ovarian clear-cell carcinomas and in 30 per cent of endometrioid carcinomas. After making the initial discovery the OvCaRe team engaged national and international collaborators to determine the frequency and relevance of these mutations. Clear-cell carcinoma and endometrioid carcinoma are the second and third most common forms of ovarian cancer; together they account for one quarter of all cases in North America and a greater proportion in Asia.

“Our discovery of the dominant mutation in clear-cell ovarian cancer raises hope for much needed treatments for this little understood cancer type. Connecting ARID1A gene mutations to endometriotic lesions accelerates us toward the development of tools to determine which women with endometriosis are at increased risk for ovarian cancer,” says Dr. David Huntsman, director of OvCaRe, a partnership program between the BC Cancer Agency and Vancouver Coastal Health Research Institute and professor, University of British Columbia, Faculty of Medicine.

The discovery was made through collaboration with Dr. Marco Marra and his team at the BC Cancer Agency’s Genome Sciences Centre, who fully decoded the RNA from 18 clear-cell carcinomas. Then through the use of novel bioinformatics tools the team took the billions of letters of genetic code and extracted a list of possible mutations.

“The finding that ARID1A is the most frequently mutated gene described thus far in endometrioid and clear cell ovarian cancers represents a major scientific breakthrough,” says Dr. Andrew Berchuck, director, Division of Gynecologic Oncology, Duke University Medical Center. “This discovery also sheds light on how endometriosis predisposes to the development of these cancers. The novel insights provided by this work have the exciting potential to facilitate advances in early diagnosis, treatment and prevention of endometrioid and clear cell cancers, which account for over 20 per cent of ovarian cancer cases.”

Huntsman explains, “ten years ago, ovarian cancer appeared to be an unsolvable problem—the liberating moment came when we established that ovarian cancer is actually a number of distinct diseases,” published in the journal *PLoS (Public Library of Science Medicine)* in 2008. “We tailor our research approach to each subtype with the hope of developing effective treatments specific to each disease.”

OvCaRe takes a collaborative and multidisciplinary approach, which has translated into major discoveries and leaps forward in ovarian cancer knowledge— made possible due to the strong support from the BC Cancer Foundation and VGH & UBC Hospital Foundation.

Paul Cacciatore, founder of the [Libby's H*O*P*E*™ website](#), lost his 26-year old cousin to

ovarian clear-cell cancer in July 2008. He created the website to honor Libby's memory, raise global ovarian cancer awareness through social media, and provide educational and emotional support for ovarian cancer survivors and their families. "This pioneering discovery by Dr. Huntsman and his dedicated ovarian cancer research team will allow the international research community to take the genomic 'high ground' in the battle against these formidable subtypes of epithelial ovarian cancer," says Cacciatore. He notes, "The Ovarian Cancer Research Program of BC's reported findings represent a critical first step towards development of one or more personalized targeted therapies to combat these lethal forms of ovarian cancer."

Historically, ovarian cancer has been a difficult disease to diagnose and treat. The symptoms in early stages are often vague, leading to late stage diagnosis and poor outcomes. In B.C., 310 women will be diagnosed with ovarian cancer in one year and approximately 220 will die from the disease.

The research initiative was generously supported by the [BC Cancer Foundation](#), [VGH & UBC Hospital Foundation](#), the Canadian Institute of Health Research and the Michael Smith Foundation for Health Research.

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Ovarian Cancer Research Program (OvCaRe) is a multidisciplinary research program involving clinicians and research scientists in gynaecology, pathology, and medical oncology at VGH and BC Cancer Agency. OvCaRe is a unique collaboration between the BC Cancer Agency, Vancouver Coastal Health Research Institute, and the University of British Columbia. Funding is provided through donations to VGH & UBC Hospital Foundation and the BC Cancer Foundation, who, in a joint partnership created a campaign to raise funds to make OvCaRe possible. The OvCaRe team is considered a leader in ovarian cancer research, breaking new ground in better identifying, understanding, and treating this disease. The team has published in *Nature*, *New England Journal of Medicine*, and works on the cutting edge of translational health research. For more information visit: www.ovcare.ca

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