

285th OMICS International Conference

Global Healthcare & Fitness Summit

July 20-22, 2015 San Francisco, USA

Molecular pathogenesis in granulosa cell tumor is not only due to somatic FOXL2 mutation

Yen-Chein Lai

Chung Shan Medical University, Taiwan

Granulosa cell tumors are rare ovarian malignancies. Their characteristics include unpredictable late recurrent and malignant behavior. Recent molecular studies have characterized the FOXL2 402C>G mutation in adult-type granulosa cell tumor. In this study, we report an 80-year-old woman with a granulosa cell tumor arising from ovary. She presented with a huge pelvic mass with postmenopausal bleeding. No obvious intraperitoneal tumor implants were observed during operation. Final diagnosis was granulosa-theca cell tumor without capsule invasion. No recurrent disease was noted during 3-year post-operation follow-up period. Molecular studies showed a heterozygous FOXL2 402C>G mutation in the tumor by direct gene sequencing. In addition, DNA replication error on analysis of the lengths of CAG repeats in androgen receptor gene revealed defective DNA mismatch repair system in the granulosa cell tumor. We propose that the 402C>G mutation in FOXL2 is critical to the development of adult granulosa cell tumor. However, the malignant behavior of this tumor is driven by DNA mismatch repair deficiency. Unequal DNA copy numbers were noted on array comparative genomic hybridization. This implies that there is malignant potential even in the early stage of the granulosa cell tumor. Late malignant recurrence may be a late event of DNA repair function disability not directly related to pathognomonic FOXL2 mutation.

Biography

Yen-Chein Lai has completed her Ph.D. at the age of 29 years from National Taiwan University College of Medicine. Her PhD research focused on mechanism analysis of HDV ribozyme activity. She has served as an Assistant Professor in School of Medical Laboratory and Biotechnology of Chung Shan Medical University since 1998. She has had much experience in the teaching of Molecular Diagnostics and Clinical Chemistry courses. Her research aims are the molecular-genetic analysis of patients with certain cancer syndromes. She has published more than 10 papers in reputed journals and has been serving as an editorial board member of repute.

yenchein@csmu.edu.tw

Notes: