

## **Simple method of detection of the most common PALB2 and RECQL genes mutations.**

Andrzej Pławski

Instytut Genetyki Człowieka PAN w Poznaniu

Breast cancer is the most common cancer in women and in majority it is thought to be sporadic. 10-15% of breast cancer cases is considered to be hereditary. The most common genetic susceptibility is caused by mutation in BRCA1 and BRCA2 gene mutations which are responsible for 5-10 % of all familial cases. Cybulski et al and Erkkö et al. described the mutation in PALB2 and RECQL genes mutations increasing risk of breast cancer. In Europe the recurrent mutation of this gene are c.1667\_1667+3delAGTA in the RECQL gene and c.509\_510delGA and c.172\_175delTTGT in the PALB2 gene. In our region also frequent is deletion of exon 9 of the PALB2. We developed the method for fast and easy identification of those 4 mutations. The method is based on CHMR technique developed by our team few years ago which let to detect the point mutations and copy number variations in the same time. The Patent Office of the Republic of Poland granted the patent (official number P.436028) to the Institute of Human Genetics PAS for the invention entitled: "Method and diagnostic kit for detecting RECQL and PALB2 gene variants in Polish patients with breast cancer". Authors of the invention are: Andrzej Pławski, Jan Lubiński, Cezary Cybulski, Zdenek Kleibl, Emilia Lis-Tanaś, Paweł Boruń