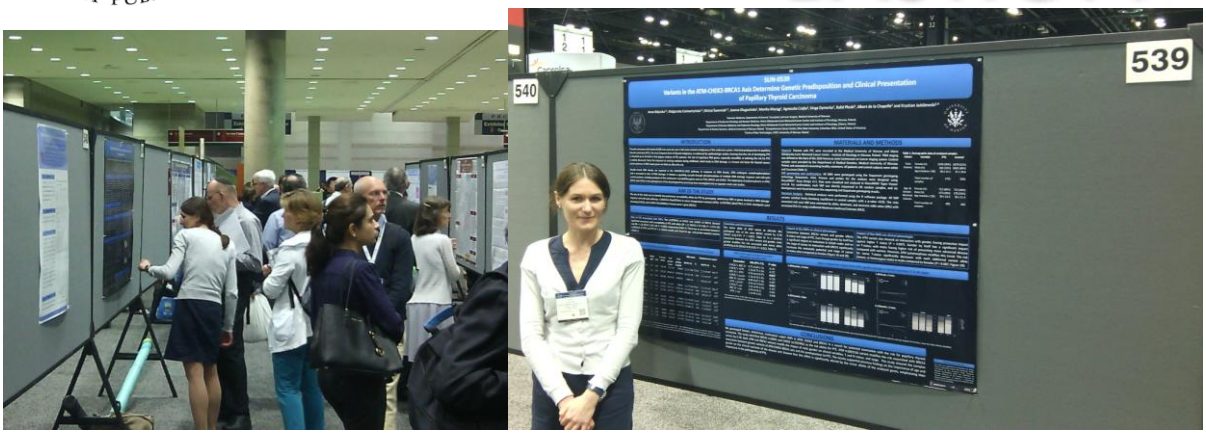
Report from active participation in 16th International Congress of Endocrinology & The Endocrine Society's 96th Annual Meeting & Expo (ICE/ENDO) Chicago, USA – Anna Wojcicka

The 16th Congress of Endocrinology gathered over 10,000 endocrinologists and basic scientists from all over the world. I had pleasure to present our data during a poster session devoted to the thyroid-related research, held on the 22nd of June.

Title of presentation: Variants in the ATM-CHEK2-BRCA1 Axis Determine Genetic Predisposition and Clinical Presentation of Papillary Thyroid Carcinoma

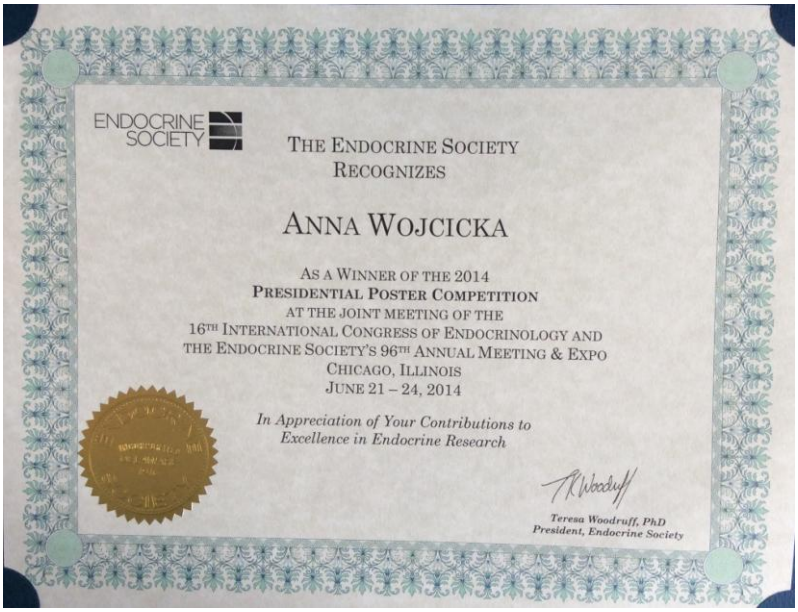
Authors: **Anna Wójcicka** (presenting author), Małgorzata Czetwertyńska, Michał Świerniak, Joanna Długosińska, Monika Maciąg, Agnieszka Czajka, Kinga Dymecka, Rafał Płoski, Albert de la Chapelle and Krystian Jażdżewski

The project involved identification of polymorphisms tailoring the genetic predisposition to papillary thyroid carcinoma (PTC). The risk of developing this kind of cancer, the most frequent thyroid malignancy, is elevated up to 8.6-fold in the first-degree relatives of PTC patients, which could be explained by polygenic action of low-penetrance alleles. Among the etiological culprits, exposure to ionizing radiation during childhood is a known risk factor for thyroid cancer. The mechanisms by which ionizing radiation promotes carcinogenesis consist mainly of its ability to induce DNA double-strand breaks. In mammalian cells, double-strand DNA breaks activate the ataxia-telangiectasia-mutated (ATM) kinase, which phosphorylates and activates checkpoint yeast homolog 2 (*CHEK2)*. Subsequently, CHEK2 phosphorylates the breast cancer 1 gene (*BRCA1)* and triggers DNA repair or, if failed, leads to cellular apoptosis. The functionality of the ATM-BRCA1-CHEK2 pathway is affected by polymorphisms and mutations within the involved genes, underlying inefficient DNA repair and leading to tumorigenic changes within the cells. The aim of this study was to identify low-penetrance susceptibility alleles for PTC by genotyping deleterious SNPs in genes involved in DNA damage-response and cell-cycle control pathways: *ATM*, *BRCA1* and *CHEK2*. We identified *CHEK2* rs17879961 (OR=2.2, *P*=2.37e-10) and *BRCA1* rs16941 (odds ratio [OR]=1.16, *P*=0.005) as risk alleles for PTC. The *ATM* rs1801516 variant modifies the risk associated with *BRCA1* variant and both *ATM* and *BRCA1* variants modify the impact of male gender on clinical variables of cancer patients. The study demonstrates a complex association between genetic variants of the ATM-CHEK2-BRCA1 axis and predisposition to PTC. Furthermore, the study supports previous findings on the importance of age and gender for the clinical outcome of the disease and shows that this effect is significantly altered by the minor alleles of the analysed genes, emphasizing their importance in the pathogenesis of PTC.



Anna Wojcicka during the poster session

The poster received the Presidential Poster Competition Award of Endocrine Society.


ENDOCRINE SOCIETY

THE ENDOCRINE SOCIETY
RECOGNIZES

ANNA WOJCICKA

AS A WINNER OF THE 2014
PRESIDENTIAL POSTER COMPETITION
AT THE JOINT MEETING OF THE
16TH INTERNATIONAL CONGRESS OF ENDOCRINOLOGY AND
THE ENDOCRINE SOCIETY'S 96TH ANNUAL MEETING & EXPO
CHICAGO, ILLINOIS
JUNE 21 – 24, 2014

In Appreciation of Your Contributions to Excellence in Endocrine Research

Teresa Woodruff, PhD
President, Endocrine Society