Report from active participation in the 49th European Human Genetics Conference, June 6-9, 2015, Glasgow, UK – Krystian Jazdzewski

The conference was one of the premier events in the field of human genetics with over 3.000 participants, over 215 oral presentations, 13 workshops, 8 educational sessions, and more than 150 exhibiting companies.

Title of presentation: Association between GWAS-derived rs966423 genetic variant and overall mortality in patients with differentiated thyroid cancer

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In the presented work, we analyzed five germline genetic variants (rs116909374, rs965513, rs944289, rs966423, rs2439302) that have previously been associated in genome-wide association study (GWAS) with increased risk of thyroid cancer, but their role in mortality of patients with thyroid cancer has not been established. Our work was a retrospective study of 1836 patients with differentiated thyroid cancer (1643 women and 193 men) with a median age at diagnosis of 49 years (and an overall median follow-up time of 8.7 years (after initial treatment at a single comprehensive cancer center between 1990-2013. Among the 5 analyzed variants, rs966423 was associated with increased mortality, which was 6.4% vs. 3.7% in TT-carriers vs. CC/CT carriers (P=0.017). Deaths per 1000 person-years were 6.81 vs. 4.01 in TT vs. CC/CT patients (HR=1.6; *P=0.038)* after adjustment for age at diagnosis, and sex. Importantly, the association of rs966423 with mortality remained significant when lymph-node metastasis, extrathyroidal invasion, angioinvasion and distant metastasis were included in the model (HR=1.89; *P=0.014)*. As a result we showed that rs966423-TT genotype was significantly associated with increased overall mortality among patients with thyroid cancers. Contrary to *BRAF* mutation and other somatic changes putatively associated with patients mortality, the status of germline rs966423 is known before the treatment, and might be used in management of mortality risk by means of modification of therapy, nevertheless it requires further investigation in large prospective studies before it is ready for clinical application.

Krystian Jazdzewski during the poster session