Report from active participation in 15th Congress of the International Society for Twin Studies; 16 - 19 November 2014 / Hungary, Budapest- Rafał Płoski

The yearly Congress of the International Society for Twin Studies is the largest eve nt specifically devoted to studies of human twins. Monozygotic twins represent an unique model to study genetic vs. environmental influences on virtually any disease/phenotype including cancer. The studied of twins are critically dependent on appropriate registries.
Prof Rafał Płoski presented data during a poster session (POSTER SESSION 1 November 16 th - 17 th). The title of presentation was:" The influence of genomic variability on phenotypic discordance – towards polish registry of monozygotic twins (MZT) discordant for chronic medically relevant conditions." It was co-authored by M. Rydzanicz, J. Kosinska, P. Gasperowicz, P. Stawinski, K. Szymański. The abstract of the presentation is given below:

Introduction/Background Monozygotic twins discordant for a phenotype/disease provide an unique opportunity to dissect genetic/epigenetic factors contributing to human morbidity. The aim of study is to identify genetic and/or epigenetic variability causing phenotypic differences in MZTs. **Patients and Methods** The project will implement genome-scale approaches based on state of the art next-generation sequencing technology such as whole-genome sequencing, whole exome sequencing, and genome-wide methylation analysis. As the first step we have started a search for MZT pairs discordant for chronic diseases through a media campaign in Poland. **Results** So far 30 MZT pairs interested in participation have been identified. In all but one zygosity was positively confirmed through analysis of a panel of 16 STR markers. The recurrent medical conditions present in identified twins include inborn defects (n=5), autism (n=3), allergy (n=2), cancer (n=3), SLE (n=2). Among the non-recurrent diseases the most interesting is a case of biopsy confirmed neuropathy of the Charcot Marie Tooth (CMT) which has been present for ~20 years in a 50 year old MZT but not his brother. **Conclusions** The collection of MZT pairs discordant for medically relevant conditions including clearly monogenic disorders such as CMT is feasible. With the assumption that genomes and epigenomes of MZTs are highly similar, linking the identified genetic/epigenetic differences to the disease phenotype should be much less challenging than in any other type of analysis aiming to establish such relationship or lack of thereof. Thus, we hope that the planned study will reveal novel (epi)mutations/loci causing human diseases.

The discussion related to the presentation resulted in an idea of collaboration between Prof Ploski group and The Hungarian Twin Registry. The further details of the collaboration are expected to be discussed during the visit of dr.Adam Tarnoki to Poland planned in May 2015.

Prof Ploski during poster session.