In search of the causes of type 1 diabetes in children with coexisting neurodevelopmental and neurological disorders - genome analysis using next generation - whole exome sequencing (NGS-WES).

Type 1 diabetes (T1D) is an autoimmune disease leading to the progressive destruction of pancreatic β-cells that produce insulin. It affects over 1.2 million children and adolescents worldwide and over 18 000 pediatric patients in Poland. As far as we know, T1D affects genetically predisposed children exposed to yet unknown environmental triggers of autoimmunity. Due to unknown reasons, its prevalence increased significantly worldwide over the past several decades and is still on the rise.

Interestingly, we have observed an increased number of children who have diagnoses or symptoms of neurological/neurodevelopmental disorders that had been admitted with new onset T1D over a past few years. Our observations are supported by the some of recent studies that have shown an increased prevalence of epilepsy ich children with T1D (2-5 times more frequent than in general population), and possible higher prevalence of T1D in children with autism spectrum disorder (ASD). These findings suggest that there might be common genetic background for the above-mentioned comorbidity.

T1D requires regular control of blood glucose, eating habits and activity. Despite new devices and rapid progress in technology children with neurodevelopmental disorders and their caregivers usually find it harder to adjust to the disease regime and are often not comfortable with devices attached directly to their bodies.

In the study we focus on this special group of patients to evaluate clinical characteristics of each patient and to perform whole exome sequencing (WES) of new generation - the most complex, individual genome analysis. NGS is not a low-cost method but using this technique we will be able not only detect single gene mutations but also find and characterize extensive chromosomal aberrations e.g. deletions or insertions, which are particularly common in neurodevelopmental disorders without spending money for additional analyses.

International societies recommend regular check-ups for neurocognitive functioning and have guidelines for children with T1D and coexisting psychological/psychiatric disorders: depression, anxiety and eating disorders. By having extensive knowledge about both T1D and neurological/neurodevelopmental disorders combined from genetic, clinical and previous immunological studies it might be possible to create specific guidelines also for this groups of patients.