**Abstract Title:** The genetic spectrum of maturity-onset diabetes of the young in the Lithuanian diabetic population

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**Topic:** 07. Metabolic and Mitochondrial Disorders

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**Keywords:** MODY/ GCK

Background/Objectives:

The most common type of monogenic diabetes is maturity-onset diabetes of the young (MODY) that affects 1–5% of all patients with diabetes mellitus. Identification of the MODY subtype is important due to subtype-related differences in clinical course and progression, response to treatment and for the genetic counseling. The aim of the study was to determine frequency and genetic spectrum of MODY in the Lithuanian diabetic population selected according to the criteria.

Methods:

The study included 55 patients from the diabetic population. Patients were referred for genetic testing based on the results of MODY probability calculator developed by Exeter University. For each individual with the diagnostic criteria for MODY, a panel of 14 MODY genes was screened using targeted next generation sequencing assay.

Results:

The (likely) pathogenic variants confirming a diagnosis of MODY were identified in 17 (30.9%) individuals. The majority of patients (88.2%) were found to harbour (likely) pathogenic variants in the GCK gene, one patient (5.9%) had pathogenic variant in ABCC8 gene and one patient (5.9%) carried (likely) pathogenic variants in both GCK and ABCC8 genes. Also, a variant of uncertain significance in PAX4 gene was detected in one individual.

Conclusion:

The most prevalent MODY subtype in the selected Lithuanian diabetic population is GCK-MODY and only few patients were found having mutations in ABCC8 and PAX4 genes. Translational biology, integrative genomic research and studies on both monogenic and polygenic forms of diabetes are required, which will broaden our understanding in terms of pathophysiology and treatment of diabetes.