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A Survey of Monogenic Diabetes in Switzerland

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Introduction

The diagnosis of Monogenic diabetes (MD) is important for appropriate genetic counselling, accurate prognosis and therapy. Because its features are non-specific, MD remains under diagnosed.

Objective

To establish the number of patients with MD in Switzerland and select patients for genetic testing of MD using the patient's clinical phenotype via the MODY probability calculator.

Methods

A questionnaire was sent to members of the Swiss Society of Endocrinology and Diabetes and the Swiss Society for Pediatric Endocrinology and Diabetes. Data requested included age & method of diagnosis, family history of diabetes, ethnic origin, BMI, HbA1c, autoimmune status, C-peptide and associated features.

Results

We identified 72 patients (40 females), median age 23 years (12 days-47 years). Of these 39/72 (54 %) had genetic testing for *GCK, HNF1B, HNF1α, HNF4A* genes. *GCK* was the commonest mutation followed by *HNF1A* then *HNF1B* mutations. The MODY calculator was used for all patients to calculate the risk of developing MD except in 5 patients (due to missing information). This index has been not applicable for patients older 35 years. For all patients this index was higher than 20% in 73% of case.

Conclusion

In Switzerland 46% of the suspected MD forms had no genetic testing and medical treatment may therefore not be optimal. The use of MODY calculator in our study was very helpful and sensitive in making diagnosis of suspected MD in most patients, but it has not allowed a distinction between different forms of MD.