Etiological diagnosis of diabetes in Italian diabetic children and adolescents

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Introduction

The prevalence of type 2 diabetes (T2D) and monogenic diabetes (MD) is still poorly described in pediatric population. T2D is increasing worldwide, with differences between Countries and ethnicities, while MD is reported to account for less than 5% of cases. We aimed to estimate the prevalence of T2D and MD in a large pediatric population of diabetic patients in Italy.

Subjects and methods

3,076 patients, diabetes onset January 2007 - December 2012 (age at diagnosis < 18 yrs) from 14 Italian Tertiary Centers for Pediatric Diabetes. Diabetes was classified as T1D, T2D, MD, syndromic diabetes, and secondary diabetes on the basis of the **diagnostic work-up** displayed in tab. 1. (at the bottom) The diagnosis of MD or syndromic diabetes was always confirmed by genetic testing.

Table 1. **Diagnostic work-up**

1. Biochemistry: C-peptide, HbA1c, T1D autoantibodies
2. Examination: a. family history, b. physical examination, c. signs of syndromic diabetes (e.g. optic atrophy, etc.)

Onset within 6 months of age, **neonatal diabetes**, otherwise:
if deficient insulin secretion, sporadic ➔ **T1D** was considered as first clinical diagnosis (confirmed by autoantibodies)
if at least 2 consecutive generations with diabetes and negative T1D autoantibodies ➔ **MODY** as first clinical diagnosis (c.d.)
if any sign suggestive of **syndromic diabetes** ➔ appropriate genetic testing
If no insulin requirement, overweight, normal/high c-peptide, features of insulin resistance (not compulsory) ➔ **T2D** as first c.d.

Results. Results (Table 2) are compared with those of papers with similar aims.

Table 2.	*Present study*	*SEARCH study**	*Norwegian registry°*	*DPV-Wiss data§*
Type 1 DM	**2,813 (91,4%)**	6,668 (85.65%)	2,725 (98.87%)	38,934 (95.5%)
Type 2 DM	**35 (1.1%)**	837 (10.88%)	1 (0.04%)	562 (1.4%)
MODY	**180 (5.8%)** *(5 MODY1, 159 MODY2, 13 MODY3, 3 MODY5)*	not reported in the main paper	26 (0.94%)	339 (0.83%) (only 263 genetically confirmed, 0.63%)
Syndromic DM	**9 (0.3%)**		not evaluated	922 (2.2%) reported as secondary or other genetic form
Neonatal DM	**16 (0.6%)**		4 (0.14%)	
Secondary DM	**23 (0.7%)**	127 (1.65%)	not evaluated	
Other	**see above**	63 (0.82%)	not reported	not reported
Total population	*3,076*	*7,695*	*2,756*	*40,757*

Discussion

T1D is the most frequent diagnosis in Italian diabetic patients <18yrs. A striking disparity, likely due to different lifestyle and genetic background, is observed between the rate of T2D of this study (1.1%, in keeping with European reports of 0.24-1.4%) and the SEARCH data from US (about 11%). At further variance with other Western countries (e.g. Norwegian registry, the DPV-Wiss study, the SEARCH study), the prevalence of MD in Italy is very high. This could depend on the fact that broader attention is devoted to MD in Italy than in the US, and also on the fact that genetic testing is easily accessible and free of charge. The close follow-up of patients with incidental hyperglycemia likely accounts for the very high rate of GCK/MODY2 mutations, the most frequent MODY type in Italy.

Main references: * Pettitt DJ et al. Diab Care 2014;37:402 ° Irgens HU et al. Diabetologia 2013;56:1512 § Schober E et al. Diab Med 2009;26:466

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