# Clinical Characteristics and Diagnostic Criteria of Maturity-Onset Diabetes Of The Young (MODY) due to Molecular Anomalies of the HNF1A Gene



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| Résumé en anglais | Context: The diagnosis of maturity-onset diabetes of the young type 3 (MODY3), associated with HNF1A molecular abnormalities, is often missed.Objective: The objective of the study was to describe the phenotypes of a large series of MODY3 patients and to reassess parameters that may improve its diagnosis. Design, Setting, and Patients: This retrospective multicenter study included 487 unrelated patients referred because of suspicion of MODY3. Genetic analysis identified 196 MODY3 and 283 non-MODY3 cases. Criteria associated with MODY3 were assessed by multivariate analysis. The capacity of the model to predict MODY3 diagnosis was assessed by the area under the receiver-operating characteristic curve and was further validated in an independent sample of 851 patients (165 MODY3 and 686 non-MODY3). Results: In the MODY3 patients, diabetes was revealed by clinical symptoms in 25% of the cases and was diagnosed by screening in the others. Age at diagnosis of diabetes was more than 25 yr in 40% of the MODY3 patients. There was considerable variability and overlap of all assessed parameters in MODY3 and non-MODY3 patients. The best predictive model was based on criteria available at diagnosis of diabetes, including age, body mass index, number of affected generations, presence of diabetes symptoms, and geographical origin. The area under the curve of the receiver-operating characteristic analysis was 0.81. When sensitivity was set to 90%, specificity was 49%. Conclusions: Differential diagnosis between MODY3 and early-onset type 2 diabetes remains difficult. Whether the proposed model will improve the pick-up rate of MODY3 diagnosis needs to be confirmed in independent populations. |
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