

Diagnosing Maturity-Onset Diabetes of the Young

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Maturity-onset diabetes of the young (MODY) is a heterogeneous group of disorders with autosomal dominant inheritance that occurs irrespective of body mass index and is usually diagnosed before 25 years of age.¹ MODY accounts for an estimated 1–2% of all diabetes.² As cases of diabetes rise among youth, clinicians should consider MODY when diagnosing diabetes in youth and young adults.

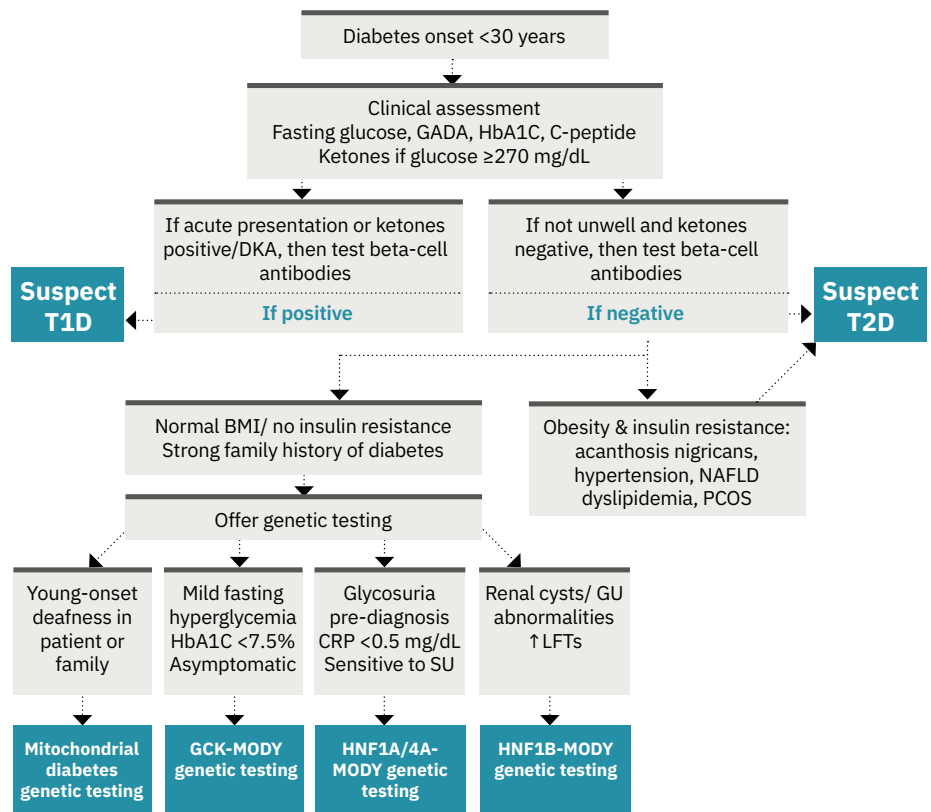
A molecular diagnosis is essential to ensure the most appropriate treatment and, consequently, the most optimal long-term prognosis. Approximately 80% of patients with MODY are misclassified as type 1 diabetes (T1D) or type 2 diabetes (T2D), and there is often a delay of more than 10 years between the onset of diabetes and molecular diagnosis.² Youth and young adults presenting with hyperglycemia but without key features of T1D or T2D should have additional testing for MODY.¹ Figure 1 provides an algorithm for identifying key features of T1D (autoantibodies) and T2D (obesity, insulin resistance) before testing for MODY.²

SEARCH for Diabetes in Youth, the first systematic study of MODY prevalence, found that 8% of children with diabetes who were autoantibody negative and C-peptide positive had mutations in HNF1A, HNF4A, or glucokinase (GCK); these mutations account for 95% of all MODY cases.³ Of the 47 MODY-mutation carriers, only 3 (6%) were correctly diagnosed.³

Familial inheritance and genetic testing play a critical role in correctly diagnosing, managing, and treating MODY.¹ For example, HNF1A- or HNF4A-MODY are typically treated with sulfonylureas, except when contraindicated during pregnancy.^{3,4} GCK-MODY often does not require lifelong treatment, except for consideration of insulin therapy during pregnancy.⁴

For more information, access Cardi-OH’s expanded resource on [youth-onset diabetes](#).

Figure 1. Diagnostic Algorithm for MODY²



BMI=body mass index, CRP=C-reactive protein, DKA=diabetic ketoacidosis, GADA=glutamic acid dehydrogenase antibodies, GCK=glucokinase, GU=genitourinary, LFTs=liver function tests, NAFLD=non-alcoholic fatty liver disease, PCOS=polycystic ovarian syndrome, SU=sulfonylurea, T1D=type 1 diabetes, T2D = type 2 diabetes.

Adapted from *When to consider a diagnosis of MODY at the presentation of diabetes: aetiology matters for correct management*

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