

Maturity-Onset Diabetes of the Young Genetic Testing

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Introduction

Maturity-onset diabetes of the young (MODY) genetic testing is addressed by this guideline.

Procedures addressed

The inclusion of any procedure code in this table does not imply that the code is under management or requires prior authorization. Refer to the specific Health Plan's procedure code list for management requirements.

Procedures addressed by this guideline	Procedure codes
GCK deletion/duplication	81479
GCK sequencing	81406
HNF1A deletion/duplication	81479
HNF1A sequencing	81405
HNF4A deletion/duplication	81479
HNF4A sequencing	81406

Maturity-Onset Diabetes of the Young

Procedures addressed by this guideline	Procedure codes
MODY gene analysis	81400
	81401
	81402
	81403
	81404
	81405
	81406
	81407
	81408
	81479
MODY multigene panel	81479

Criteria

Introduction

Requests for maturity-onset diabetes of the young (MODY) genetic testing are reviewed using the following criteria.

For gene testing in non-MODY contexts (e.g., neonatal diabetes, familial hyperinsulinism, etc.), refer to the general policies, *Genetic Testing to Diagnose Non-Cancer Conditions* and *Genetic Testing by Multigene Panels*, as appropriate.

HNF1A Sequencing and Deletion/Duplication Analysis

- Genetic Counseling:
 - Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND
- Previous Genetic Testing:
 - No previous HNF1A gene sequencing or deletion/duplication analysis, and
 - No known MODY mutation in biologic relative, AND
- Diagnostic Testing for Symptomatic Individuals:
 - Member has a diagnosis of diabetes prior to 35 years of age, and
 - Member has a biological parent with diabetes, and

- Member does NOT have symptoms consistent with a specific condition or specific gene mutation, and
- Member does NOT have any of the following features:
 - Extra-pancreatic manifestations (e.g., congenital malformations and other signs of syndromic diabetes), or
 - Pancreatic autoantibodies suggestive of type 1 diabetes, or
 - Body mass index (BMI) greater than or equal to 35 kg/m², or
 - Acanthosis nigricans, AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy.

HNF4A Sequencing and Deletion/Duplication Analysis

- Genetic Counseling:
 - Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND
- Previous Genetic Testing:
 - No previous HNF4A gene sequencing or deletion/duplication analysis, and
 - No known MODY mutation in biologic relative, and
 - Member has previous HNF1A testing with no deleterious mutation found, AND
- Diagnostic Testing for Symptomatic Individuals:
 - Member meets criteria for HNF1A testing, AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy.

GCK Sequencing and Deletion/Duplication Analysis

- Genetic Counseling:
 - Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND
- Previous Genetic Testing:
 - No previous GCK gene sequencing or deletion/duplication analysis, and
 - No known MODY mutation in biologic relative, AND
- Diagnostic Testing for Symptomatic Individuals:
 - Member meets criteria for HNF1A testing and has had previous HNF1A testing with no deleterious mutation found, or
 - Member has a personal history of the following features presenting outside of pregnancy:
 - Persistent, stable elevation of fasting blood glucose (5.5-8 mmol/L), and
 - HbA1C that is no more than mildly elevated (less than or equal to 7.5%), and
 - At least one oral glucose tolerance test demonstrates a small increment (less than 4.6 mmol/L), or

- Member has a personal history of the following features in the context of gestational diabetes:
 - Persistent elevation of fasting blood glucose (5.5-8 mmol/L) before, during, and after pregnancy, and
 - At least one oral glucose tolerance test demonstrates a small increment (less than 4.6 mmol/L) either during or after pregnancy, AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy.

Sequencing and Deletion/Duplication Analysis of ABCC8, BLK, CEL, HNF1B, INS, KCNJ11, KLF11, NEUROD1, PAX4, and PDX1

Individual testing of these genes for the purpose of diagnosing MODY is not medically necessary.

MODY Multigene Panel Testing

- Genetic Counseling:
 - Pre- and post-test genetic counseling by an appropriate provider (as deemed by the Health Plan policy), AND
- Previous Genetic Testing:
 - No previous MODY genetic testing, and
 - No known MODY mutation in biologic relative, AND
- Diagnostic Testing for Symptomatic Individuals:
 - Member has a diagnosis of diabetes prior to 35 years of age, and
 - Member has a family history of diabetes consistent with autosomal dominant inheritance, and
 - Member does NOT have symptoms consistent with a specific condition or specific gene mutation, and
 - Member does NOT have any of the following features:
 - Extra-pancreatic manifestations (e.g., congenital malformations and other signs of syndromic diabetes), or
 - Pancreatic autoantibodies suggestive of type 1 diabetes, or
 - Body mass index (BMI) greater than or equal to 35 kg/m², or
 - Acanthosis nigricans, AND
- Rendering laboratory is a qualified provider of service per the Health Plan policy.

Billing and Reimbursement

Introduction

This section outlines the billing requirements for tests addressed in this guideline. These requirements will be enforced during the case review process whenever appropriate. Examples of requirements may include specific coding scenarios, limits on allowable test combinations or frequency and/or information that must be provided on a claim for automated processing. Any claims submitted without the necessary information to allow for automated processing (e.g. ICD code, place of service, etc.) will not be reimbursable as billed. Any claim may require submission of medical records for post service review.

- ABCC8, BLK, CEL, HNF1B, INS, KCNJ11, KLF11, NEUROD1, PAX4, and PDX1 analysis are not separately reimbursable for the purposes of MODY testing.
- Any individual gene or multi-gene panel is only reimbursable once per lifetime.
- When otherwise reimbursable, the following limitations apply:
 - When a panel is being performed, it is only reimbursable when billed with a single, appropriate panel procedure code (e.g., 81479*).
 - When use of a panel code is not possible, each billed component procedure will be assessed independently.
 - In general, only a limited number of panel components that are most likely to explain the member's presentation will be reimbursable. The remaining panel components will not be reimbursable.
 - When the test is billed with multiple stacked codes, only the following genes may be considered for reimbursement:
 - HNF1A
 - GCK
 - HNF4A

Note: *The panel code(s) listed here may not be all-inclusive. For further discussion of what is considered an appropriate panel code, please refer to the guideline *Laboratory Billing and Reimbursement*.

What is MODY?

Maturity-onset diabetes of the young (MODY) is a type of monogenic diabetes characterized by non-insulin-dependent diabetes and early onset (usually before age 35 years).¹⁻³

Incidence

As of 2019, approximately 37.3 million people in the United States had diabetes, or 11.3% of the population.⁴ The most common types of diabetes are type 1 and type 2. The genetic basis of these types of diabetes is largely unknown. The disease is thought to be the result of a combination of multiple genetic and environmental risk factors.⁴ Monogenic forms of diabetes are rare, accounting for approximately 1-4% of all diabetes cases.^{1,2}

Symptoms

Diabetes is a disorder that results in elevated blood glucose. Over time, the disorder can cause various health problems, including diseases of the heart, kidneys, eyes, and nervous system.

Cause

Monogenic forms of diabetes are caused by a mutation in a single gene. There are at least 14 known MODY genes. Three genes account for the majority of cases.¹⁻³

- MODY3: Mutations in the hepatocyte nuclear factor-1 alpha (HNF1A) gene are the most common cause of MODY, accounting for 30-65% of all cases. This type is characterized by a progressive insulin secretory defect due to beta-cell failure. Laboratory evaluations are negative for pancreatic islet cell antibodies (ruling out type 1) and glycosuria is detectable even at low blood glucose levels (<10 mmol/l). Treatment of choice for people with this type of MODY is sulfonylureas, and a majority of individuals can be transferred from insulin to oral agents.
- MODY2: Mutations in the glucokinase gene (GCK) are the next most common cause of MODY, accounting for approximately 30-50% of cases. GCK encodes the glucokinase enzyme, which acts as the pancreatic glucose sensor. Mutations result in lifelong, stable, mild fasting hyperglycemia. HbA1C values are usually just above the high normal range. People with GCK mutations rarely require treatment. This type of MODY may be detected during pregnancy, when glucose tolerance testing is routinely performed.
- MODY1: Mutations in the hepatocyte nuclear factor-4 alpha (HNF4A) gene cause a clinical presentation similar to HNF1A. However, mutations in this gene are much less common (less than 10% of MODY). Age of onset may be later, and there is not a low renal threshold. HNF4A mutations can also cause high birth weight in newborns and transient neonatal hypoglycemia. These individuals are also more sensitive to sulfonylurea treatment.

The remaining genes are rare causes of MODY, each accounting for less than 5% of cases.¹⁻³

- MODY5: Caused by heterozygous mutations in HNF1B. The vast majority of HNF1B mutations cause Renal Cysts and Diabetes Syndrome, which is associated with

diabetes, renal cysts, genitourinary malformations, pancreatic atrophy, hyperuricemia, and abnormal liver function tests.

- MODY8: Caused by heterozygous mutations in CEL. Affected individuals also have pancreatic exocrine dysfunction (diabetes-pancreatic-exocrine dysfunction syndrome).
- Others include: MODY4 (PDX1/IPF-1), MODY6 (NEUROD1), MODY7 (KLF11), MODY9 (PAX4), MODY10 (INS), MODY11 (BLK), MODY12 (ABCC8), MODY13 (KCNJ11), and APPL1 (MODY14).

Other monogenic causes of pediatric diabetes include the following (not meant to be an all-inclusive list):^{1,5,6}

- Permanent neonatal diabetes mellitus (PNDM), defined as persistent hyperglycemia in the first 6 months of life. It is most commonly caused by mutations in the ABCC8, KCNJ11, and INS genes. Biallelic mutations in GCK, PDX1, and RFX6 are less common causes.
- Transient neonatal diabetes mellitus (TNDM), which accounts for ~50% of all neonatal diabetes. Affected individuals are at risk for recurrence later in life. 70% of TNDM cases are due to 6q24 methylation defects, while other genetic causes include variants in ABCC8 and KCNJ11.
- Cystic fibrosis, caused by biallelic CFTR mutations (for more information, see test-specific guideline, *Cystic Fibrosis Genetic Testing*)
- Immune dysregulation, polyendocrinopathy, and enteropathy, X-linked (IPEX syndrome), due to mutations in FOXP3
- Maternally inherited diabetes and deafness (MIDD), caused by mutations in mitochondrial genes: MT-TL1, MT-TK, or MT-TE
- Wolcott-Rallison syndrome, due to mutations in EIF2AK3
- Wolfram syndrome, caused by mutations in WFS1 and less often CISD2
- Other genes associated with PNDM and extra-pancreatic features include GATA6, GLIS3, IER3IP1, NEUROG3, PTF1A, and RFX6.

Inheritance

MODY is typically inherited in an autosomal dominant manner.

Autosomal dominant inheritance

In autosomal dominant inheritance, individuals have 2 copies of the gene and only one mutation is required to cause disease. When a parent has a mutation, each offspring has a 50% risk of inheriting the mutation. Males and females are equally likely to be affected.

Mutations that occur de novo in an affected individual, reduced penetrance, and variable expressivity have been reported.³ Thus, the absence of a family history does not, by itself, rule out a diagnosis of MODY.

Diagnosis

Diabetes evaluations may include assessment of pancreatic autoantibodies, plasma glucose levels, hemoglobin A1C assessment (HbA1C), and oral glucose tolerance testing (OGTT). For young individuals in whom a diagnosis of type 1 or type 2 diabetes is considered unlikely, genetic testing for monogenic diabetes may be considered, especially in the presence of a strong family history.⁴

Management

Like other forms of diabetes, monogenic diabetes is treated with diet, oral antidiabetic agents, and/or insulin, as required for blood sugar regulation.³ Most individuals with MODY are not insulin-dependent. Knowledge of the specific genetic cause of MODY may help guide management.

Survival

Survival of affected individuals was reduced when compared with unaffected relatives, specifically with regard to cardiovascular-related causes of death.⁷

Test information

Introduction

Testing for MODY may include single gene sequence analysis, single gene deletion/duplication analysis, or multigene panels of various sizes.

Next Generation Sequencing Assay

Next generation sequencing (NGS), which is also sometimes called massively parallel sequencing, was developed in 2005 to allow larger scale and more efficient gene sequencing. NGS relies on sequencing many copies of small pieces of DNA simultaneously and using bioinformatics to assemble the sequence. Sequence analysis detects single nucleotide substitutions and small (several nucleotide) deletions and insertions. Regions analyzed typically include the coding sequence and intron/exon boundaries. Promoter regions and intronic sequences may also be sequenced if disease-causing mutations are known to occur in these regions of a gene.

Deletion and Duplication Analysis

Analysis for deletions and duplications can be performed using a variety of technical platforms including exon array, Multiplex ligation-dependent probe amplification (MLPA), and NGS data analysis. These assays detect gains and losses too large to be identified through standard sequence analysis, often single or multiple exons or whole genes.

Multi-Gene Testing Panels

The efficiency of NGS has led to an increasing number of large, multi-gene testing panels. NGS panels that test several genes at once are particularly well-suited to conditions caused by more than one gene or where there is considerable clinical overlap between conditions making it difficult to reliably narrow down likely causes. Additionally, tests should be chosen to maximize the likelihood of identifying mutations in the genes of interest, contribute to alterations in management for an individual, and/or minimize the chance of finding variants of uncertain clinical significance.

MODY multigene panels include a wide variety of genes associated with MODY and monogenic diabetes in general. Some panels may also include genes associated with other types of monogenic diabetes and glycemic disorders, such as neonatal diabetes, syndromic diabetes, and familial hyperinsulinism.

Guidelines and evidence

American Association of Clinical Chemistry

American Association of Clinical Chemistry (AACL, 2023) stated:⁸

- "Routine determination of genetic markers such as HLA genes or single nucleotide polymorphisms (SNP) is of no value at this time for the diagnosis of management of type 1 diabetes. Typing for genetic markers and the use of genetic risk scores are recommended for individuals who cannot be clearly classified as having type 1 or type 2 diabetes. A (moderate)"
- For selected diabetes syndromes, including neonatal diabetes and MODY (maturity onset diabetes of the young), valuable information including treatment options can be obtained with definition of diabetes-associated mutations. A (moderate)"
- "There is no role for routine genetic testing in people with type 2 diabetes. These studies should be confined to the research setting and evaluation of specific syndromes. A (moderate)"

American Diabetes Association

The American Diabetes Association (ADA, 2025) stated:⁹

- "Children and young adults who do not have typical characteristics of type 1 or type 2 diabetes and family history of diabetes in successive generations (suggestive of an autosomal dominant pattern of inheritance) should have genetic testing for maturity-onset diabetes of the young (MODY)." (B)
- "The diagnosis of monogenic diabetes should be considered in children and adults diagnosed with diabetes in early adulthood with the following findings:"
 - "Diabetes diagnosed within the first 6 months of life"

- Diabetes without typical features of type 1 or type 2 diabetes (negative diabetes-associated autoantibodies, no obesity, and lacking other metabolic features, especially strong family of diabetes)
- Stable, mild fasting hyperglycemia (100-150 mg/dL [5.6-8.5 mmol/L]), stable A1C between 5.6% and 7.6% (between 38 and 60 mmol/mol), especially if no obesity"

European Molecular Genetics Quality Network

The European Molecular Genetics Quality Network (EMQN, 2008) made the following recommendations for testing:²

- Testing for GCK mutations (presentation outside of pregnancy):
 - Persistent, stable elevation of fasting blood glucose (5.5-8 mmol/l)
 - HbA1c just above the upper limit of normal (rarely exceeds 7.5%)
 - Oral glucose tolerance testing demonstrates a small increment (4.6 mmol/l is often used to prioritize testing)
 - May have a family history consistent with autosomal dominant inheritance
- Testing for GCK mutations (for evaluation of gestational diabetes):
 - Persistent elevation of fasting blood glucose (5.5-8 mmol/l) before, during and after pregnancy
 - At least one oral glucose tolerance test with an increment of <4.6 mmol/l (either during or after pregnancy)
- Testing for HNF1A mutations:
 - Young-onset diabetes (<25 years old)
 - Non-insulin-dependent diabetes
 - Family history of diabetes (at least two generations)
 - Absence of pancreatic islet autoantibodies
 - Glycosuria at blood glucose levels <10 mmol/l
 - Marked sensitivity to sulfonylureas
 - Features suggestive of monogenic diabetes (lack of obesity or evidence of insulin resistance, absence of acanthosis nigricans, etc)
- Testing for HNF4A mutations:
 - Should be considered when HNF1A analysis is normal but the clinical features are strongly suggestive of HNF1A
 - "When diabetic family members have marked macrosomia (>4.4 kg at term) or if diazoxide-responsive neonatal hyperinsulinism has been diagnosed in the context of familial diabetes."
 - "Macrosomic babies with diazoxide-responsive hyperinsulinism and a strong family history of diabetes should be considered for HNF4A mutation screening."

- Syndromic forms of diabetes, including HNF1B and CEL mutations, "are not included in these guidelines since testing is guided by the non-endocrine pancreatic or extra-pancreatic clinical features."

International Society for Pediatric and Adolescent Diabetes

The International Society for Pediatric and Adolescent Diabetes (ISPAD, 2022) made the following recommendations:¹

- "Testing for GCK-MODY, which is the commonest cause of persistent, incidental hyperglycemia in the pediatric population, is recommended for mild stable fasting hyperglycemia that does not progress." (B)
- "In familial autosomal dominant symptomatic diabetes, mutations in the HNF1A gene (HNF1A-MODY) should be considered as the first diagnostic possibility" (B)
- "Specific features can suggest subtypes of MODY, such as renal developmental disease or renal cysts (HNF1B-MODY), macrosomia and/or neonatal hypoglycemia (HNF4A-MODY), exocrine pancreatic dysfunction or pancreatic cysts (CEL-MODY), or hearing impairment and maternal inheritance of diabetes (mitochondrial diabetes)" (C)
- "Obesity alone should not preclude genetic testing in young persons, especially if:
 - Family history is strongly suggestive of autosomal dominant inheritance of diabetes
 - If some affected family members are NOT obese
 - And/or, there are no other features of metabolic syndrome." (C)
- "Features that suggest monogenic diabetes in children initially thought to have T1D [Type 1 diabetes] are listed below. . . none of these are pathognomonic and should be considered together rather than in isolation:"
 - "Diabetes presenting before 6 months of age (as T1D is extremely rare in this age group), or consider NDM [neonatal diabetes mellitus] if the diagnosis is between 6 and 12 months and there is no evidence of autoimmunity or if the person with diabetes has other features such as congenital defects, or an unusual family history."
 - "Family history of diabetes in one parent and other first-degree relatives of that affected parent."
 - "Absence of islet autoantibodies, especially if checked at diagnosis."
 - "Preserved β -cell function, with low insulin requirements and detectable C-peptide (either in blood or urine) over an extended partial remission phase (at least 5 years after diagnosis)."
- "In young people, T2D [Type 2 diabetes] often presents around puberty and the majority are obese. As there is no diagnostic test for T2D and because obesity has become so common in children, children and adolescents with monogenic diabetes may also be obese and can be very difficult to distinguish from T2D. One recent study found that 3% of obese youth with presumed T2D in fact carried pathogenic

monogenic diabetes variants. Features that suggest monogenic diabetes in young people with suspected T2D are listed below:"

- "Lack of consistent severe obesity among affected family members."
- "Lack of consistent acanthosis nigricans and/or other markers of metabolic syndrome (hypertension, low HDL-cholesterol, etc.) among affected family members."
- "Family history of diabetes in one parent and other first-degree relatives of that affected parent, especially if any affected family member lacks obesity and other markers of metabolic syndrome."
- "Unusual distribution of fat, such as central fat with thin or muscular extremities."
- "From a clinical perspective, specific clinical scenarios when a diagnosis of monogenic diabetes should be considered include:"
 - "Diabetes presenting before 6 months of age, which is known as NDM."
 - "Autosomal dominant familial mild hyperglycemia or diabetes."
 - "Diabetes associated with extra-pancreatic features (such as, for example, congenital heart or gastrointestinal defects, brain malformations, severe diarrhea, or other autoimmune conditions in a very young child)."
 - "Monogenic IR [insulin resistance] syndromes (see below: characterized by high insulin levels or high insulin requirements; abnormal distribution of fat with a lack of subcutaneous fat, especially in extremities; dyslipidemia, especially high triglycerides; and/or significant acanthosis nigricans)."
- "Three genes are responsible for the majority of MODY cases (GCK, HNF1A, and HNF4A) ... At least 14 different genes have been reported to cause diabetes with a MODY-like phenotype, and some panels will include all these genes, or possibly also many other genes associated with exceedingly rare recessive causes. It is reasonable to consider including syndromic causes such as mitochondrial diabetes, as diabetes can often be the first presenting feature and a molecular diagnosis can thereby guide monitoring and treatment of other associated features. In the modern era of expanded testing by many different laboratories, caution must be used when interpreting test results, as often there is very little information available to support the causality of rare variants in uncommon subtypes."

Selected Relevant Publications

A 2018 expert-authored review stated that MODY has an onset in adolescence or young adulthood, typically less than 35 years.³

- "Molecular genetic testing approaches to determine the associated MODY gene can include a combination of gene-targeted testing (serial single-gene or multigene panel) and comprehensive genomic testing (chromosomal microarray analysis or exome sequencing), depending on the phenotype."

- "Serial single-gene testing. Sequence analysis of the most likely genes is performed first. If no pathogenic variant is found, gene-targeted deletion/duplication analysis to detect exon-sized deletions could be considered, especially for those genes (CEL, GCK, HNF1A, HNF1B, and HNF4A) in which whole-gene or multiexon deletions have been identified."
- "A MODY multigene panel that includes the 14 known MODY-related genes and other genes of interest is most likely to identify the genetic cause of MODY at the most reasonable cost while limiting identification of variants of uncertain significance and pathogenic variants in genes that do not explain the underlying phenotype."
 1. "The genes included in the panel and the diagnostic sensitivity of the testing used for each gene vary by laboratory and are likely to change over time."
 2. "Some custom laboratory-designed multigene panels may include genes not associated with MODY but possibly associated with other types of monogenic diabetes; other custom laboratory-designed panels may not include the genes that rarely cause MODY."
 3. "In some laboratories, panel options may include a custom laboratory-designed panel and/or custom phenotype-focused exome analysis that include genes specified by the clinician."
 4. "Methods used in a panel may include sequence analysis, deletion/duplication analysis, and/or other non-sequencing-based tests. Note: Given that whole-gene and/or multiexon deletions have been identified in GCK, HNF1A, HNF1B, and HNF4A, a multigene panel that also includes deletion/duplication analysis is recommended."

Note:

This benefit/harm statement only applies to those jurisdictions that do not have Medicare guidance. Based upon the guidelines and evidence provided in the clinical policy, following EviCore's criteria for maturity-onset diabetes of the young (MODY) testing will ensure that testing will be available to those members most likely to benefit from a genetic diagnosis. For those not meeting criteria, it ensures alternate diagnostic strategies are considered. However, it is possible that some members who have the condition, but have non-standard features, will not receive an immediate approval for testing.

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