GENETIC TESTING: GASTROENTEROLOGIC DISORDERS (NON-CANCEROUS)

OVERVIEW

Genetic testing for gastroenterologic (non-cancerous) disorders may be used to confirm a diagnosis in a patient who has signs and/or symptoms of a specific gastroenterologic disorder. Confirming the diagnosis may alter aspects of management and may eliminate the need for further diagnostic workup. This document addresses genetic testing for common gastroenterologic (non-cancerous) conditions.

POLICY REFERENCE TABLE

Below is a list of higher volume tests and the associated laboratories for each coverage criteria section. This list is not all inclusive.

Coverage Criteria Sections	Example Tests (Labs)	Common CPT Codes	Common ICD Codes	Ref
Known Familial Variant Analysis for Gastroenterologic Disorders				
Known Familial Variant Analysis	Targeted Mutation Analysis for a Known Familial Variant	81403		
Celiac Disease				
HLA-DQ Variant Analysis	HLA-DQ Genotyping (ARUP Laboratories) Celiac Disease HLA Typing (Johns Hopkins Medical Institutions - Pathology Laboratory)	81370, 81375, 81376, 81377, 81382, 81383	K90.0, R10.0-R10.13, R10.3-R10.829, R10.84-R10.9	5, 6, 7, 9
Hereditary Hemochromatosis				

HFE Sequencing and/or Deletion/Duplication Analysis	HFE Sequencing Analysis	81256, 81479	E83.110, E83.118, E83.119, R79.0, E83.19, R16.0	1, 13
Lactase Insufficiency				
MCM6 Targeted Variant Analysis	MCM6 Targeted Mutation Analysis	81479	E73.1	16
Hereditary Pancreatitis				
Hereditary Pancreatitis Multigene Panel	Pancreatitis Panel (CFTR, CTRC, PRSS1, SPINK1) Sequencing (ARUP Laboratories) PRSS1 Targeted Mutation Analysis Test (Prevention Genetics) CTRC Sequencing Analysis SPINK1 Sequencing Analysis	81401, 81404, 81405, 81479	K85.0-K85.92, K86.1, Z83.79	2, 3, 4, 8, 11, 12
Inflammatory Bowel Disease				
Inflammatory Bowel Disease / Crohn's Disease Diagnostic Algorithmic Tests	Prometheus IBD sgi Diagnostic (Prometheus Laboratories)	81479, 82397, 83520, 86140, 88346, 88350, 81499	K50-K52	10, 14
	Inflammatory Bowel Disease Panel (Children's Hospital of Philadelphia)	86255, 86256, 86671, 81499		
Inflammatory Bowel Disease / Crohn's Disease Prognostic Algorithmic Tests	PredictSURE IBD (KSL Diagnostics)	0203U	K50-K52	10
	Crohn's Disease Prognostic Panel (ARUP Laboratories)	83516, 86671, 81499		
	Prometheus Crohn's Prognostic Panel (Prometheus Laboratories	81401, 83520, 86021, 86255, 81499		
Hereditary Inflammatory Bowel Disease / Crohn's Disease Panel Tests	Monogenic Inflammatory Bowel Disease Panel (Invitae)	81479	K50-K52	15, 16, 17, 18, 21, 22, 23,
	Infantile Enterocolitis and Inflammatory Bowel Disease Monogenic (CGC Genetics USA)			

	Very Early Onset Inflammatory Bowel Genomic Panel (Children's Hospital of Philadelphia)			24, 25
	Early Onset Inflammatory Bowel Disease (EGL Genetics)			
Test Specific Not Covered Gastroenterologic Disorders Tests				
Test Specific Not Covered Gastroenterologic Disorders Tests	ASH FibroSURE (LabCorp)	0002M		
	NASH FibroSURE (LabCorp)	0003M		
	EsoGuard™ (Lucid Diagnostics)	0114U		

OTHER RELATED POLICIES

This policy document provides coverage criteria for Genetic Testing for Gastroenterologic Conditions (Non-Cancerous). Please refer to:

- ***Genetic Testing: Hereditary Cancer Susceptibility Syndromes*** for coverage criteria related to germline testing for hereditary cancer syndromes, including Lynch/HNPCC syndrome.
- ***Genetic Testing: Prenatal and Preconception Carrier Screening*** for coverage criteria related to carrier screening in the prenatal, preimplantation, and preconception setting.
- ***Genetic Testing: Prenatal Diagnosis (via amniocentesis, CVS, or PUBS) and Pregnancy Loss*** for coverage related to prenatal and pregnancy loss diagnostic genetic testing for tests intended to diagnose genetic conditions following amniocentesis, chorionic villus sampling or pregnancy loss.
- ***Genetic Testing: Multisystem Inherited Disorders, Intellectual Disability, and Developmental Delay*** for coverage criteria related to diagnostic genetic testing for conditions affecting multiple organ systems.
- ***Genetic Testing: Metabolic, Endocrine, and Mitochondrial Disorders*** for coverage criteria related to genetic testing for MTHFR.

- ***Genetic Testing: General Approach to Genetic Testing*** for coverage criteria related to genetic testing for any non-cancerous GI disorders that is not specifically discussed in this or another non-general policy.

COVERAGE CRITERIA

KNOWN FAMILIAL VARIANT ANALYSIS FOR GASTROENTEROLOGIC DISORDERS

I. Targeted mutation analysis for a known familial variant (81403) for a gastroenterologic disorder is considered **medically necessary** when:

   A. The member has a close relative[1] with a known pathogenic or likely pathogenic variant causing the condition.

II. Targeted mutation analysis for a known familial variant (81403) for a gastroenterologic disorder is considered **investigational** for all other indications.

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CELIAC DISEASE

HLA-DQ Genotyping Analysis

I. *HLA-DQ2* and *HLA-DQ8* variant analysis (81370, 81375, 81376, 81377, 81382, 81383) to rule out celiac disease is considered **medically necessary** when:

   A. The member meets both of the following:

      1. The member has negative serologic and histologic testing, **AND**

      2. The member has signs, symptoms, and or risk factors that indicate at least moderate to high risk of celiac disease (e.g., chronic diarrhea/steatorrhea with unintended weight loss, a first or second degree relative with celiac disease, type 1 DM, autoimmune thyroiditis, Down syndrome, Turner syndrome, pulmonary hemosiderosis), **OR**

B. The member has discordant serologic and histologic (biopsy) findings.

II. *HLA-DQ2* and *HLA-DQ8* variant analysis (81370, 81375, 81376, 81377, 81382, 81383) to rule out celiac disease is considered **investigational** for all other indications.

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HEREDITARY HEMOCHROMATOSIS

HFE Sequencing and/or Deletion/Duplication Analysis

I. *HFE* sequencing and/or deletion/duplication analysis (81256, 81479) to establish a diagnosis of hereditary hemochromatosis is considered **medically necessary** when:

A. The member has abnormal serum iron indices, defined as transferrin saturation greater than or equal to 45% and/or elevated ferritin, indicating iron overload.

II. *HFE* sequencing and/or deletion/duplication analysis (81256, 81479) to screen for hereditary hemochromatosis in the general population is considered **investigational**.

III. *HFE* sequencing and/or deletion/duplication analysis (81256, 81479) to establish a diagnosis of hereditary hemochromatosis is considered **investigational** for all other indications.

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LACTASE INSUFFICIENCY

MCM6 Variant Analysis

I. *MCM6* variant analysis (81479) for the prediction of lactase insufficiency is considered **investigational**.

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HEREDITARY PANCREATITIS

Hereditary Pancreatitis Multigene Panel

I. Hereditary pancreatitis multigene panel analysis (81401, 81404, 81405, 81479) to establish a diagnosis of hereditary pancreatitis is considered **medically necessary** when:

   A. The member meets at least one of the following:

      1. The member has a first[1a]- or second-degree[1b] relative with pancreatitis, **OR**

      2. The member is age 18 years or younger and has had an unexplained, documented episode of acute pancreatitis, **OR**

      3. The member has had recurrent (i.e., two or more separate, documented episodes with hyper-amylasemia) attacks of acute pancreatitis for which there is no explanation (anatomical anomalies, ampullary or main pancreatic strictures, trauma, viral infection, gallstones, alcohol, drugs, hyperlipidemia, etc.), **OR**

      4. The member has unexplained (i.e., idiopathic) chronic pancreatitis, **AND**

   B. The panel includes, at a minimum, the following genes: *PRSS1*, *SPINK*.

II. Hereditary pancreatitis multigene panel analysis (81401, 81404, 81405, 81479) to establish a diagnosis of hereditary pancreatitis is considered **investigational** for all other indications.

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INFLAMMATORY BOWEL DISEASE

Inflammatory Bowel Disease / Crohn's Disease Diagnostic Algorithmic Tests

I. Inflammatory bowel disease diagnostic algorithmic tests (81479, 82397, 83520, 86140, 86255, 86256, 86651, 88346, 88350, 81499) are considered **investigational.**

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Inflammatory Bowel Disease / Crohn's Disease Prognostic Algorithmic Tests

I. Inflammatory bowel disease prognostic algorithmic tests (0203U, 81401, 83516, 83520, 86021, 86255, 86671, 81499) are considered **investigational**.

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Hereditary Inflammatory Bowel Disease / Crohn's Disease Panel Tests

I. Genetic testing for inflammatory bowel disease (81479), including Crohn's disease via a multigene panel is considered medically necessary to confirm a diagnosis and/or determine appropriate treatment when:

   A. Patient has very early onset of typical IBD symptoms before age 2 years, **OR**

   B. Patient is under the age of 18 with aggressive, refractory or unusual IBD presentation, **OR**

   C. Patient is under the age of 18 with IBD symptoms, and also has a family history of IBD or immunodeficiency.

   D. The panel includes at a minimum the following genes: IKBKG, TTC7, ADAM17, NCF2, NCF4, SLC37A4, XIAP, LRBA, CD40LG; WAS, IL10R, IL10, FOXP3

II. Genetic testing for inflammatory bowel disease (81479), including Crohn's disease, via a multigene panel is considered **investigational** for all other indications.

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TEST-SPECIFIC NOT COVERED GASTROENTEROLOGIC DISORDERS TESTS

I. The use of these specific gastroenterologic disorders tests are considered **investigational**:

A. ASH FibroSURE (0002M)
B. NASH FibroSURE (0003M)
C. EsoGuard™ (0114U)

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NOTES AND DEFINITIONS

1. Close relatives include first, second, and third degree blood relatives on the same side of the family:

   a. **First-degree relatives** are parents, siblings, and children

   b. **Second-degree relatives** are grandparents, aunts, uncles, nieces, nephews, grandchildren, and half siblings

   c. **Third-degree relatives** are great grandparents, great aunts, great uncles, great grandchildren, and first cousins

2. **Typical inflammatory bowel disease (IBD) symptoms** include diarrhea, abdominal pain, infections, and bleeding.

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CLINICAL CONSIDERATIONS

Genetic Counseling

Genetic counseling is recommended for patients who are at risk for inherited disorders and who are interested in undergoing genetic testing. Interpreting the results of genetic tests and understanding risk factors can be challenging and genetic counseling helps in the understanding the potential impacts of genetic testing, including possible effects the test results could have on the individual or their family members. Genetic counseling may alter the utilization of genetic testing substantially and has been shown to reduce inappropriate testing and should be performed by an individual with experience and expertise in genetic medicine and genetic testing methods.

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BACKGROUND AND RATIONALE

Practice Guidelines and Position Statements

Genetic Testing for Celiac Disease

American College of Gastroenterology

The guidelines from the American College of Gastroenterology (2013) addressing the diagnosis and management of celiac disease (CD) stated the following on human leukocyte antigen (HLA) gene testing:

1. HLA-DQ2/DQ8 testing should not be used routinely in the initial diagnosis of CD [celiac disease] (Strong recommendation, moderate level of evidence).
2. HLA-DQ2/DQ8 genotyping testing should be used to effectively rule out the disease in selected clinical situations (Strong recommendation, moderate level of evidence).
3. Examples of such clinical situations include but are not limited to:
    a. Equivocal small-bowel histological finding (Marsh I-II) in seronegative patients
    b. Evaluation of patients on a gluten-free diet in whom no testing for CD was done before gluten-free diet

c. Patients with discrepant celiac-specific serology and histology

d. Patients with suspicion of refractory CD where the original diagnosis of celiac remains in question.

American College of Gastroenterology published guidelines (2018) addressing the management of Crohn's disease in adults state that genetic and routine serologic testing is not indicated to establish the diagnosis of Crohn's disease.

American Gastroenterological Association Institute

The American Gastroenterological Association Institute (2006) issued a position statement on the diagnosis and management of CD. Regarding serologic testing, the Institute concluded that, in the primary care setting, the transglutaminase immunoglobulin (Ig) A antibody test is the most efficient single serologic test for diagnosing CD.5, The guidelines indicated that the antiendomysial antibodies IgA test is more time-consuming and operator dependent than the tissue transglutaminase (tTG). If IgA deficiency is strongly suspected, testing with IgG anti endomysial antibody (EMA) and/or tTG IgG antibody test is recommended. If serologic test results are negative and CD is still strongly suspected, providers can test for the presence of the disease-associated HLA alleles and, if present, perform a small intestinal mucosal biopsy. Alternatively, if signs and symptoms suggest that small intestinal biopsy is appropriate, patients can proceed to biopsy without testing for HLA alleles.

U.S. Preventive Services Task Force

The US Preventative Service Task Form (2017) released guidelines on screening adults and children for CD. These guidelines reviewed the use of tTG IgA testing followed by an intestinal biopsy to screen asymptomatic patients. Genotype testing was not discussed. The overall conclusion of this review was that the current balance of evidence was insufficient to assess benefits and harms resulting from screening for CD.

<u>Genetic Testing for Hereditary Hemochromatosis</u>

American College of Gastroenterology

In 2019, practice guidelines from the American College of Gastroenterology made the following statement on genetic testing for hereditary hemochromatosis:

> "We recommend that family members, particularly first-degree relatives, of patients diagnosed with HH should be screened for HH (strong recommendation, moderate quality of evidence)."

Genetic Testing for Hereditary Pancreatitis

American College of Gastroenterology

In 2013, the American College of Gastroenterology guidelines on management of acute pancreatitis included the following statement: "Genetic testing may be considered in young patients (<30 years old) if no cause [of acute pancreatitis] is evident, and a family history of pancreatic disease is present (conditional recommendation, low quality of evidence).

In 2015, the American College of Gastroenterology Clinical Guideline: Genetic Testing and Management of Hereditary Gastrointestinal Cancer Syndromes recommended genetic testing of patients with suspected familial pancreatic cancer to include analysis of BRCA1/2, CDKN2A, PALB2, and ATM. Evaluation for Peutz-Jeghers Syndrome, Lynch Syndrome, and HP-associated genes should be considered if personal and/or family history criteria are met for the syndrome.

American Pancreatic Association

In 2014, the American Pancreatic Association published Practice Guidelines in Chronic Pancreatitis: Evidence-Based Report on Diagnostic Guidelines. A classification guideline for the etiology of chronic pancreatitis (CP) includes genetic mutations in PRSS1, CFTR, SPINK1, and others.

American Society of Clinical Oncology

In 2018, the American Society of Clinical Oncology (ASCO) published "Evaluating Susceptibility to Pancreatic Cancer: ASCO Provisional Clinical Opinion". The ASCO reported that cancer-unaffected individuals should be offered genetic risk evaluation if they are members of families with an identified pathogenic cancer susceptibility gene variant, from families that meet criteria for genetic evaluation for known hereditary syndromes that are linked to pancreatic cancer, and from families that meet criteria for familial pancreatic cancer. ASCO further considered what surveillance strategies should be used for individuals with a predisposition to pancreatic ductal adenocarcinoma to screen for pancreatic and other cancers. Surveillance can be considered for individuals who are first-degree relatives of individuals with familial pancreatic cancer and/or individuals with a family history of pancreatic cancer who carry a pathogenic germline variant in genes associated with predisposition to pancreatic cancer.

Genetic Testing for Hereditary Inflammatory Bowel Disease / Crohn's Disease Panel Tests

UpToDate (Higuchi LM and Bousvaros A, 2021)

Clinical features that raise suspicion for monogenic IBD include

- Young age of onset (eg, younger than six years, particularly younger than age two years)
- Family history of IBD and/or immunodeficiency in multiple family members, particularly with male predominance, or consanguinity
- Recurrent infections or unexplained fever
- Associated features of autoimmunity (eg, arthritis, primary sclerosing cholangitis, anemia, or endocrine dysfunction)
- Very severe IBD and/or resistance to conventional therapies for IBD
- Symptoms or signs suggesting hemophagocytic lymphohistiocytosis (hepatomegaly, fever, cytopenias, high ferritin)
- Lesions of the skin, nails, or hair
- Current or past history of cancer in the patient

Infants or young children presenting with these features warrant careful evaluation for monogenic IBD and consultation with an immunologist

UpToDate (Snapper SB and McGovern DPB, 2021)

"Very early onset IBD — There is a diverse spectrum of rare genetic disorders that produce IBD-like intestinal inflammation caused by genetic variants that have a large effect on gene function and typically present in infancy [89-91]. Over 60 unique monogenic IBD disorders have been described that affect mucosal homeostasis in diverse ways, including:

- Epithelial barrier and response defects (eg, IKBKG, TTC7, ADAM17)
- Dysfunction of neutrophil granulocytes (eg, NCF2, NCF4, SLC37A4)
- Hyper- and autoinflammatory disorders (eg, XIAP)
- Complex defects in T- and B-cell function (eg, LRBA, CD40LG; WAS)
- Regulatory T cells and IL-10 signaling (eg, IL10R, IL10, FOXP3)

Some of these disorders with gene defects that affect predominantly hematopoietic cells (eg, IL-10R, IL-10, XIAP, FOXP3) respond to stem cell transplantation."

European Society of Paediatric Gastroenterology, Hepatology and Nutrition (ESPGHN)

Summary of clinical features that should prompt considering a monogenic inflammatory bowel disease workup (Red flag signs)
Age of inflammatory bowel disease (IBD) presentation

- <2 years IBD symptom onset
- <6 years IBD symptom onset in particular when other red flag signs are present

Family history

- Affected family member with a suspected monogenic disorder
- Consanguinity
- Multiple family members with early-onset IBD

Comorbidity and extraintestinal manifestations are particularly relevant for monogenic IBD diagnostic considerations when rare or atypical for patient age irrespective of organ manifestation.

- Recurrent severe infections or atypical infections consistent with diagnostic criteria of a primary immunodeficiency
- Hemophagocytic lymphohistiocytosis
- Autoimmune features in particular features of Immunodysregulation polyendocrinopathy enteropathy X-linked syndrome
- Malignancies
- Multiple intestinal atresias

British Society of Gastroenterology

We suggest that genetic testing for monogenic disorders should be considered in adolescents and young adults who have had early onset (before 5 years of age) or particularly aggressive, refractory or unusual IBD presentations.

European Crohn's and Colitis Organization (ECCO) and ESPGHN

1. In infants younger than 2 years, allergic colitis, immunological disorders and monogenic forms of colitis should be excluded.
2. Unusual disease evolution, history of recurrent infections, HLH, and non-response to multiple IBD medications may indicate an underlying genetic defect which should prompt genetic and/or immunological analyses at any age during childhood.

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