



**Micro  
Health**  
Laboratories

*.....for precious life*

## MicroGEN **GASTROENTEROLOGY** TESTING



**MICRO** GEN™  
PrecisionDx

The Future of Targeted Treatment

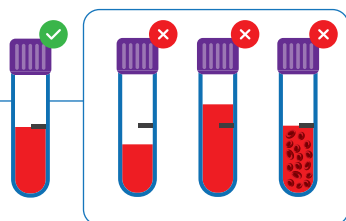
# MicroGEN Gastroenterology Testing

Test code	Test Parameters	Method
GG256	Alagille syndrome 1 (JAG1) deletion/duplication analysis	MLPA
GG257	Alagille syndrome gene panel	NGS
GG258	Cholestasis gene panel	NGS
GG259	Congenital Diarrhea gene panel	NGS
GG260	Congenital hepatic fibrosis gene panel	NGS
GG261	Gastrointestinal Atresia gene panel	NGS
GG262	Gilbert syndrome (UGT1A1) gene analysis (only point mutation analysis)	NGS
GG263	Hemochromatosis gene panel	NGS
GG264	Hirschsprung Disease gene panel	NGS
GG265	Pancreatitis gene panel	NGS
GG266	Polycystic liver disease gene panel	NGS
GG267	Progressive familial intrahepatic cholestasis gene panel	NGS
GG268	Progressive familial intrahepatic cholestasis-3 (ABCB4) deletion/duplication analysis	MLPA
GG269	Tyrosinemia gene analysis	NGS
GG270	UGT1A1 repeat analysis	Fragment Analysis
GG271	Wilson disease (ATP7B) gene analysis	NGS

NGS tests: 28 days, Fragment Analysis tests: 14 days, MLPA tests: 21 days

## Sample

EDTA Blood - 4 mL.



## Method

## TAT

NGS test	28 days
MLPA test	21 days
Fragment Analysis Test	14 days

Required Family History.

Relevant clinical Information and symptoms.

MicroGEN  
**GASTROENTEROLOGY**  
TESTING



# GASTROENTEROLOGY TESTING

## About the Test

Gastroenterology primarily focuses on the treatment and identification of diseases affecting the stomach, small intestine, colon (large intestine), gallbladder, liver, pancreas, and esophagus. It also involves studying, treating, and diagnosing conditions related to the GI tract, including all parts involved in the digestion of food and drinks. Many gastrointestinal diseases often have a genetic basis, and recent research has identified the specific genes responsible for many of these conditions. This has led to the development of genetic tests that can diagnose these diseases and identify asymptomatic family members at risk.

## Benefits of MicroGen Gastroenterology Testing

### Accurate Diagnosis and Prognostic Information:

MicroGen Gastroenterology Genetic testing provides precise subtyping of gastrointestinal diseases. For instance, it can differentiate between idiopathic and hereditary pancreatitis, which is essential for an accurate differential diagnosis. Additionally, it helps distinguish chronic pancreatitis from syndromic disorders that include pancreatitis, offering critical insights for informed treatment and management decisions.

### Risk Assessment and Management:

Identifying the specific genetic cause of a disease allows for a more comprehensive risk assessment. For example, patients with hereditary pancreatitis due to PRSS1 mutations are at an increased risk of developing exocrine and endocrine pancreatic insufficiency and pancreatic cancer, compared to those with idiopathic pancreatitis or pancreatitis linked to CFTR or SPINK1 mutations.

### Family Member Risk Stratification:

Genetic testing can reveal which family members are at risk of developing gastrointestinal diseases. This facilitates early preventive measures, lifestyle recommendations, and routine follow-ups by healthcare professionals. It also supports informed family planning decisions.

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## When to Consider Cardiogenetic Testing

Genetic testing for gastroenterology is typically recommended in the following situations:

- If there is a family history of gastrointestinal (GI) disorders.

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- If you are experiencing symptoms such as chronic abdominal pain, diarrhea, blood in the stool, or unexplained weight loss.

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- If you have been diagnosed with a GI disorder and would like to understand the genetic factors that may be contributing to your condition.

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- If you are planning to start a family and have a known genetic predisposition to GI disorders.

For more information on **MicroGen NGS panels** and their benefits,  
**Please contact us**

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## Precision Diagnostics with MicroGen NGS Panels

Micro Health Laboratories (MHL) utilizes next-generation sequencing (NGS), a cutting-edge molecular genetics method, to analyze patient DNA for genetic variants. MicroGen NGS panels offer simultaneous analysis of a large number of genes, significantly increasing the chances of identifying the genetic cause of diseases with complex or non-specific symptoms. These panels reduce both the time and cost from symptom presentation to diagnosis and enhance diagnostic yield. Additionally, the results can provide information about recurrence risk (the likelihood of having another child with a similar condition) and may also benefit other family members.

**MHL offers over 500+ NGS panels covering all medical specialties.**

These panels are recommended for patients who meet any or multiple of the following criteria:

- **Clinical features**
- **Family history of a particular disorder**
- **Multiple genes linked to the condition**
- **Well-defined disease-associated genes**

(Reference: Genet Med 2015 Jun;17(6): 444-51.doi: 10.1038/gim.2014.122. Epub 2014 Sep 18)

## Comprehensive Analysis

Genetic variants, or changes in the DNA sequence, can be harmful and may cause serious medical conditions, particularly hereditary diseases originating in germ cells and present in all body cells.

Identifying these disease-causing variants is essential for accurate diagnosis, prognosis, and determining the most effective treatments for patients.

NGS enables the thorough analysis of thousands of clinically relevant target genes, providing results quickly enough to support timely clinical decisions. NGS can detect various types of DNA variants, including point mutations (nucleotide substitutions) and small insertions or deletions.

MHL uses targeted sequencing to identify both known variants linked to specific genetic disorders and novel variants in disease-associated genes.

## Adherence to Best-Practice Guidelines

The identified variants are reported following international best-practice guidelines, including those from the American College of Medical Genetics (ACMG) and Clinical Molecular Science Standards (CMSS).

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# MicroGen Carrier Sequencing

For cases where the genetic cause is unknown, MHL offers the MicroGen Carrier Sequencing. This test covers all protein-coding regions, including the intron-exon boundary regions of approximately 23,000 genes, as well as mitochondrially encoded genes. The sequencing provides uniform coverage across the exome with a mean depth of over 80-100x, ensuring that more than 98% of targeted base pairs are covered at  $\geq 10x$ . The MicroGen Carrier Seq enables the detailed detection and analysis of both single nucleotide variants (SNVs) and copy number variants (CNVs), with a sensitivity range of 75-99% for CNVs, depending on the length and zygosity of the deletion or duplication.

## Comprehensive Reporting

Each report includes a detailed description of the methods used, references to publications that support the medical and scientific findings, and recommendations for follow-up analyses for specific diseases. We provide thorough reporting of pathogenic variants, likely pathogenic variants, and variants of uncertain significance (VUS), ensuring that all clinically relevant information is communicated.

MHL provide high-quality sequencing and best-in-class data analysis - interpreted and communicated in comprehensive medical reports. Our multidisciplinary team of experts, including consultant geneticists, genetic counselors, genome analysts, and bioinformaticians, is involved in the interpretation and validation of genetic variants, ensuring the highest standards of accuracy and reliability. Our experienced professionals meticulously interpret genomic data, providing clear and actionable results. This integrated approach ensures that every analysis is conducted with precision, offering clear insights and recommendations for patient care.

## Medical Genetic Counseling

MHL offer expert medical genetic counseling as an integral part of the genetic testing journey. Genetic counseling is a communicative process designed to support patients and their families both before and after genetic testing. This service is educational, impartial, and nondirective. Before any genetic test is conducted, genetic counselors gather a detailed family history, explain the testing methods to be used, and discuss the risks, benefits, limitations, and implications of a potential genetic diagnosis.

After receiving genetic test results, genetic counseling assists both the specialist physician and the patient in interpreting the findings. Patients are informed about the potential consequences of the results, including the likelihood of developing the genetic disorder, the risk of passing it on to future children, and strategies to prevent, reduce, or manage these risks. Our goal in providing counseling is to equip patients with a deeper understanding of their results, enabling them to make more informed decisions regarding their health and future.

Reference: Nat Rev Genet. 2018 Dec;19(12):735-736. doi: 10.1038/s41576-018-0057-3. Ann Lab Med. 2018 Jul;38(4):291-295. doi: 10.3343/alm.2018.38.4.291.

## Limitations

Genetic testing plays a crucial role in the diagnostic process, but it doesn't always provide a clear answer. In some cases, a genetic variant may exist but not be identified due to limitations in current medical knowledge or testing technology. Accurate interpretation of test results may also depend on understanding the true biological relationships within a family. Failing to disclose these relationships accurately may lead to incorrect interpretations, misdiagnoses, or inconclusive test outcomes.

**Contextual Interpretation:** It's important to consider that test results are interpreted in the context of clinical findings, family history, and other laboratory data. Genetic testing only reports variations in genes potentially related to the proband's medical condition. Rare polymorphisms can result in false negative or positive results, and misinterpretation may occur if the provided information is inaccurate or incomplete.

**Detection Limitations:** Certain events, such as CNVs (detection range 75-99%), translocations, repeat expansions, and chromosomal rearrangements, may not be reliably detected by MicroGen Panel testing. Additionally, variants in untranslated regions, promoters, and intronic regions are not assessed using this method. Deep intronic variants are not assessed by this method.

**Accuracy Considerations:** While genetic testing is highly accurate, rare instances of inaccurate results may occur due to various factors. These factors may include mislabeled samples, incorrect clinical or medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, blood transfusion, or mosaicism (where a genetic change is present in only a small percentage of cells, making it undetectable by the test).

**Variant Annotation Discrepancies:** The population allele frequencies and in silico predictions for the GRCh38 version of the human genome are obtained by lifting over the coordinates from the hg19 genome build. Since existing population allele frequencies (e.g., 1000 Genomes, ExAC, gnomAD-Exome) are available only for the hg19 genome version, some discrepancies in variant annotation may occur due to complex changes in certain regions of the genome.



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