Micro Health Laboratories

.....for precious life

MicroGEN ENDOCRINOLOGY TESTING

MICROGEN PrecisionDx

The Future of Targeted Treatment

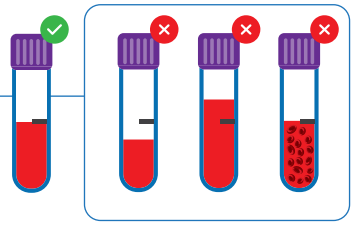
Microgen

ENDOCRINE TESTING

MicroGEN Endocrinology Testing

Test code	Test Parameters	Method
GG224	Abnormal genitalia and disorders of sex development panel	NGS
GG225	Alstrom syndrome (ALMS1) gene analysis	NGS
GG226	Androgen receptor (AR) deletion/duplication analysis	MLPA
GG227	Androgen receptor (AR) gene analysis	NGS
GG228	Comprehensive monogenic diabetes gene panel	NGS
GG229	Congenital adrenal hyperplasia CYP21A2 (21-OH) deletion/duplication analysis	MLPA
GG230	Congenital adrenal hyperplasia CYP21A2 (21-OH) gene analysis	NGS
GG231	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency (HSD3B2) gene analysis	NGS
GG232	Congenital adrenal hyperplasia gene panel	NGS
GG233	Congenital hypopituitarism gene panel	NGS
GG234	CYP11B1 gene sequencing	NGS
GG235	GATA3 gene sequencing	NGS
GG236	Glucocorticoid deficiency gene panel	NGS
GG237	Hereditary pancreatitis gene panel	NGS
GG238	HNF1B gene sequencing	NGS
GG239	Hypercholesterolemia gene panel	NGS
GG240	Hyperlipidemia gene panel	NGS
GG241	Hyperparathyroidism gene panel	NGS
GG242	Hypoglycemia, hyperinsulinism and ketone metabolism gene panel	NGS
GG243	Hypomagnesemia gene panel	NGS
GG244	Hypothyroidism and resistance to thyroid hormone gene panel	NGS
GG245	Kallmann syndrome gene panel	NGS
GG246	Leptin deficiency (LEP) gene analysis	NGS
GG247	Maturity-Onset Diabetes of the Young (MODY) & neonatal diabetes gene panel	NGS
GG248	MC4R gene sequencing	NGS
GG249	MCM6 gene sequencing	NGS
GG250	MKRN3 gene sequencing	NGS
GG251	Monogenic and syndromic obesity gene panel	NGS
GG252	NROB1, ABCD1, AIRE gene sequencing	NGS
GG253	Premature ovarian failure gene panel	NGS
GG254	Pseudohypoparathyroidism Ia (GNAS) gene sequencing	NGS
GG255	SRD5A2 gene analysis	NGS

Sample



EDTA Blood - **4 ml.**

- ☑ Required Family History.
- ☑ Relevant clinical Information and symptoms.

Method	TAT
NGS test	28 days
STR	14 days
MLPA test	21 days
Sanger sequencing	28 days

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About **the Test**

Hereditary endocrine disorders impact the endocrine system, a network of glands that produce and release hormones to help control or coordinate many body processes. Changes in various endocrine glands can cause a range of symptoms. For example, insufficient cortisol production in the adrenal glands can result in adrenal insufficiency, leading to symptoms such as fatigue, digestive issues, dehydration, and skin alterations.

The main functions of the endocrine glands and tissues can be broadly grouped into several categories:

- **Reproduction and sexual differentiation**
- **Development and growth**
- **Maintenance of the internal environment**
- **Metabolism and nutrient supply regulation**

A single hormone may affect more than one of these functions, while each function may be controlled by several hormones.

Common **Endocrine Disorders**

- **Adrenal Insufficiency:** Caused by low cortisol levels. Symptoms include fatigue, stomach upset, dehydration, and skin changes.
- **Pituitary Disorders:** Conditions such as Cushing's disease and gigantism result from hormone imbalances in the pituitary gland.
- **Thyroid Disorders:** Conditions such as hyperthyroidism and hypothyroidism arise from overproduction or underproduction of thyroid hormones.
- **Androgen Insensitivity Syndrome (AIS):** Pathogenic variants in the androgen receptor lead to AIS, characterized by a female phenotype in individuals with an XY karyotype.

When to get tested
for Endocrine Disorders?

Consider genetic testing for endocrine disorders if you experience any of the following:

- **Ambiguous genitalia**
- **Dysgenic gonads**
- **Short stature**
- **Obesity with or without associated developmental delay**
- **Hyperglycemia with a strong family history**
- **Hyperthyroidism or hypothyroidism**

Who should consider
Endocrine testing?

Genetic testing is advised if:

- **Hormone levels are excessively high or low, indicating a potential endocrine disease or disorder.**
- **The body does not respond correctly to hormones.**
- **There is a family history of endocrine disorders.**

Early detection and diagnosis of hormone-related diseases can greatly enhance prognosis.

Benefits of MicroGen
Endocrine testing

- **Precision Diagnosis: Provides a precise genetic diagnosis, guiding personalized treatment plans and management strategies.**
- **Family Planning: Offers valuable information for family members about genetic risks and reproductive choices.**
- **Lifestyle Recommendations: Provides advice on lifestyle changes to manage or reduce the risk of developing endocrine-related conditions.**

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 these 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.

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 ⩾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.

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).

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
Counselling

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.

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

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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