Micro Health Laboratories

.....for precious life

MicroGEN DERMATOLOGY TESTING

MICROGEN PrecisionDx

The Future of Targeted Treatment


Microgen
DERMATOLOGY
TESTING

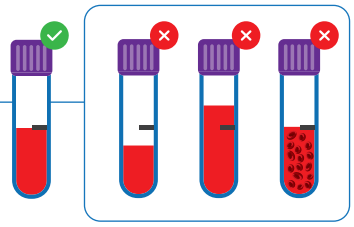
MicroGen Dermatology Testing

Test code	Test Parameters	Method
GG195	MicroGen Acute intermittent porphyria - HMBS gene analysis	NGS
GG196	MicroGen Adams-Oliver Syndrome gene panel	NGS
GG197	MicroGen Albinism gene panel	NGS
GG198	MicroGen ATP2A2 gene sequencing	NGS
GG199	MicroGen Cutis Laxa gene panel	NGS
GG200	MicroGen Dyskeratosis Congenita gene panel	NGS
GG201	MicroGen Ectodermal dysplasia gene panel	NGS
GG202	MicroGen EDA, EDAR, EDARADD gene analysis	MLPA
GG203	MicroGen Ehlers-Danlos Syndrome gene panel	NGS
GG204	MicroGen Epidermolysis bullosa gene panel	NGS
GG205	MicroGen Erythropoietic protoporphyria-1 (FECH) gene sequencing	NGS
GG206	MicroGen Harlequin ichthyosis (ABCA12) gene analysis	NGS
GG207	MicroGen Hereditary Acrodermatitis Enteropathica gene panel	NGS
GG208	MicroGen Hereditary Melanoma and Skin Cancer gene panel	NGS
GG209	MicroGen Hermansky-Pudlak Syndrome gene panel	NGS
GG210	MicroGen Ichthyosis gene panel	NGS
GG211	MicroGen Kindler syndrome (FERMT1) gene sequencing	NGS
GG212	MicroGen KRT5 gene sequencing	NGS
GG213	MicroGen Neurofibromatosis gene panel	NGS
GG214	MicroGen Pachyonychia Congenita gene panel	NGS
GG215	MicroGen Palmoplantar Keratoderma gene panel	NGS
GG216	MicroGen PLEC gene sequencing	NGS
GG217	MicroGen Progeria and Progeroid Syndromes gene panel	NGS
GG218	MicroGen Sjogren-Larsson syndrome (ALDH3A2) gene analysis	NGS
GG219	MicroGen Tuberous Sclerosis gene panel	NGS
GG220	MicroGen TYR deletion/duplication analysis	NGS
GG221	MicroGen Waardenburg Syndrome gene panel	NGS
GG222	MicroGen Xeroderma pigmentosum gene panel	NGS
GG223	MicroGen ZMPSTE24 gene sequencing	NGS

NGS tests: 28 days, MLPA tests: 21 days

Sample

EDTA Blood - **4 ml.**



Method

TAT

NGS test	28 days
MLPA test	21 days

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

MICROGEN™ PrecisionDx

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DERMATOLOGY TESTING

About the Test

Dermatological conditions can range from minor to extremely complex. Patients with genetic diseases affecting the skin may not be aware of these inherited conditions. Many dermatological conditions have a genetic basis influenced by both inherited traits and environmental factors. These diseases often exhibit a broad range of symptoms and severity and are frequently polygenic, involving multiple genes.

MicroGen dermatology genetic testing can assess a patient's risk of developing serious skin disorders, other cancers, and pigmentation abnormalities. Our comprehensive genetic tests can identify disease-causing variants, providing essential information that can improve patient care and inform treatment choices.

Benifits of **MicroGen**
Dermatology Testing

- **Accurate diagnosis and prognostic information:** MicroGen dermatology genetic testing enables the differentiation between isolated ocular/oculocutaneous albinism and various syndromic forms, such as Hermansky-Pudlak, Chediak-Higashi, Griscelli, and Waardenburg syndromes. Additionally, it provides valuable prognostic insights into visual impairment associated with oculocutaneous albinism and predicts the outcomes of syndromic albinism, including the severity of neurological or immunodeficiency in different types of Griscelli syndrome.

- **Subtyping Diseases:** Provides precise subtyping of hereditary dermatological diseases, leading to more accurate diagnoses.

- **Precision Diagnosis:** Provides a precise genetic diagnosis, guiding personalized treatment plans and management strategies.

- **Family Planning:** Identifies the mode of inheritance and family risk. It assists in understanding genetic risks for future generations and supports decision-making regarding family planning.

- **Early Intervention:** Enables early access to therapies and clinical trials tailored to specific genetic mutations, potentially improving outcomes.

- **Lifestyle Recommendations:** Provides advice on lifestyle changes to manage or reduce the risk of developing dermatological conditions.

For more information on MicroGen dermatological genetic testing and its benefits, please contact us.

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.

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.

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.