Micro Health Laboratories

.....for precious life

MicroGEN NEUROGENETIC TESTING

MICROGEN PrecisionDx

The Future of Targeted Treatment

MicroGEN Neurogentic Testing

Test code	Test Parameters	Method
GG40	4H syndrome gene panel	NGS
GG41	ABCD1 gene analysis	NGS
GG42	ADGRG1 gene sequencing	NGS
GG43	Adrenoleukodystrophy (ABCD1) gene analysis	NGS
GG44	ADSSL1 gene analysis	NGS
GG45	Aicardi-Goutieres syndrome gene panel	NGS
GG46	Alexander disease (GFAP) gene analysis	NGS
GG47	Alkaptonuria (HGD) gene analysis	NGS
GG48	Amyotrophic Lateral Sclerosis gene panel	NGS
GG49	Arthrogryposis and congenital myasthenic syndrome gene panel	NGS
GG50	Ataxia-telangiectasia (ATM) deletion and duplication analysis	MLPA
GG51	Ataxia-telangiectasia (ATM) gene analysis	NGS
GG52	ATP1A2 and ATP1A3 gene sequencing	NGS
GG53	ATRX gene analysis	NGS
GG54	Autism Spectrum Disorders gene panel (Case–Parent Trio analysis)	NGS
GG55	Autosomal dominant adult-onset demyelinating leukodystrophy (LMNB1) deletion and duplication analysis	MLPA
GG56	Benign infantile epilepsy gene panel	NGS
GG57	Comprehensive Epilepsy gene Panel	NGS
GG58	Brown Vialetto-Van Laere syndrome gene panel	NGS
GG59	Calpainopathy and LGMD2A (CAPN3) deletion and duplication analysis	MLPA
GG60	Canavan disease (ASPA) gene analysis	NGS
GG61	CAPN3 gene sequencing	NGS
GG62	Cerebral Cavernous Malformation gene Panel	NGS
GG63	Charcot-Marie-Tooth 1A and HNPP (PMP22, COX10, TEKT3) deletion and duplication analysis	MLPA
GG64	Charcot-Marie-Tooth and sensory neuropathies gene panel	NGS
GG65	Charcot-Marie-Tooth type 4 (EGR2, GDAP1, NEFL, PRX) deletion and duplication analysis	MLPA
GG66	Charcot-Marie-Tooth type 4C (SH3TC2) deletion and duplication analysis	MLPA
GG67	CLP1 gene sequencing	NGS
GG68	Coenzyme q10 Deficiency gene panel	NGS
GG69	Collagen Type VI-Related Disorders gene panel	NGS
GG70	Congenital Muscular Dystrophy (LAMA2) deletion and duplication analysis	MLPA
GG71	Comprehensive neurology gene panel	NGS
GG72	Congenital Myasthenic Syndromes gene Panel	NGS
GG73	Creatine Metabolism Deficiency gene Panel	NGS
GG74	CSTB gene sequencing	NGS
GG75	Cystic megalencephaly (MLC1) gene analysis	NGS
GG76	Dementia gene Panel	NGS
GG77	Dravet syndrome (SCN1A) deletion and duplication analysis	MLPA
GG78	Dravet syndrome (SCN1A) gene analysis	NGS
GG79	Duchenne Muscular Dystrophy (DMD) deletion and duplication analysis	MLPA
GG80	Duchenne Muscular Dystrophy (DMD) gene sequencing	NGS
GG81	DYSF gene sequencing	NGS
GG82	Dysferlinopathy and LGMD2B (DYSF) deletion and duplication analysis	MLPA
GG83	Dystonia gene panel	NGS
GG84	Dystonia 28 (KMT2B) gene sequencing	NGS
GG85	Early infantile epileptic encephalopathy-4 (STXBP1) deletion and duplication analysis	MLPA
GG86	Early-onset juvenile parkinsonism gene panel	NGS
GG87	Emery-Dreifuss Muscular Dystrophy gene Panel	NGS
GG88	Epileptic encephalopathy gene panel	NGS
GG89	Episodic ataxia gene panel	NGS
GG90	Familial female mental retardation and epilepsy gene panel	NGS
GG91	Familial hemiplegic migraine gene panel	NGS
GG92	Familial temporal lobe epilepsy-1 (LGI1) gene sequencing	NGS
GG93	Friedreich ataxia (FXN) - Repeat expansion analysis	STR
GG94	Fukuyama Congenital Muscular Dystrophy (FKTN) gene sequencing (does not include repeat expansions)	NGS
GG95	GAN and DCAF8 gene sequencing	NGS
GG96	GBE1 gene sequencing	NGS
GG97	GCH1 gene sequencing	NGS

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Test code	Test Parameters	Method
GG98	Giant axonal neuropathy-1 (GAN) gene analysis	NGS
GG99	Gordon Holmes syndrome (RNF216 and OTUD4) gene sequencing	NGS
GG100	Hereditary essential tremor (FUS and TENM4) gene sequencing	NGS
GG101	Hereditary spastic paraplegia gene panel	NGS
GG102	HINT1 gene sequencing	NGS
GG103	Holoprosencephaly gene Panel	NGS
GG104	Huntington's disease (HTT) - Repeat expansion analysis	STR
GG105	Hyperekplexia gene panel	NGS
GG106	Hypomyelination syndrome gene panel	NGS
GG107	Idiopathic Generalized and Focal Epilepsy gene Panel	NGS
GG108	IGHMBP2 gene sequencing	NGS
GG109	Joubert syndrome gene panel	NGS
GG110	KCNT1 gene sequencing	NGS
GG111	Krabbe disease (GALC) deletion and duplication analysis	MLPA
GG112	Krabbe disease (GALC) gene analysis	NGS
GG113	Leukodystrophy and Leukoencephalopathy gene Panel	NGS
GG114	LGMD and Congenital Muscular Dystrophy gene Panel	NGS
GG116	Lissencephaly gene panel	NGS
GG117	Macrocephaly and Overgrowth Syndrome gene Panel	NGS
GG118	MECP2 gene sequencing	NGS
GG119	Metabolic Epilepsy gene Panel	NGS
GG120	Metabolic Myopathy and Rhabdomyolysis gene Panel	NGS
GG121	Metachromatic leukodystrophy gene panel	NGS
GG122	MFSD8 gene sequencing	NGS
GG123	Microcephaly and Pontocerebellar Hypoplasia gene Panel	NGS
GG124	Migraine gene Panel	NGS
GG125	Muscular dystrophy and congenital myopathy gene panel	NGS
GG126	MYH3 gene sequencing	NGS
GG127	Myotonia congenita (CLCN1) deletion and duplication analysis	MLPA
GG128	Myotonia congenita gene panel	NGS
GG129	NCL and Progressive Myoclonic Epilepsy Panel	NGS
GG130	NDUFS7 gene sequencing	NGS
GG131	Nemaline Myopathy gene Panel	NGS
GG132	Neurodegeneration due to cerebral folate transport deficiency (FOLR1) gene sequencing	NGS
GG133	Neurodegeneration with brain iron accumulation 2B (PLA2G6) deletion and duplication analysis	MLPA
GG134	Neurodegeneration with brain iron accumulation gene panel	NGS
GG135	Neurofibromatosis (NF1 and NF2) gene analysis	NGS
GG136	Neurofibromatosis type 1 (NF1) deletion and duplication analysis	MLPA
GG137	Neurofibromatosis type 1 (NF1) gene analysis	NGS
GG138	Neurofibromatosis type 2 (NF2) deletion and duplication analysis	MLPA
GG139	Neurofibromatosis type 2 (NF2) gene analysis	NGS
GG140	Neuronal ceroid lipofuscinosis gene panel	NGS
GG141	Neuronal migration disorder gene panel	NGS
GG142	Neuro-Ophthalmology gene Panel	NGS
GG143	Neurotransmitter disorders gene panel	NGS
GG144	NKX2-1 gene sequencing	NGS
GG145	NOTCH3 (CADASIL) gene analysis	NGS
GG146	PANK2 gene sequencing	NGS
GG147	Pantothenate kinase-associated neurodegeneration (PANK2) deletion and duplication analysis	MLPA
GG148	Parkinson Disease gene Panel	NGS
GG149	Periodic Paralysis gene Panel	NGS
GG150	PIEZO2 gene sequencing	NGS
GG151	PLP1 gene sequencing	NGS
GG152	PMP22 deletion and duplication analysis	MLPA
GG153	PMP22 gene analysis (inflammatory demyelinating polyneuropathy screen)	NGS
GG154	Polymicrogyria gene Panel	NGS
GG155	Pontocerebellar hypoplasia gene panel	NGS
GG156	Porphyria gene Panel	NGS

MicroGEN Neurogentic Testing

Test code	Test Parameters	Method
GG157	PRKN gene deletion and duplication analysis	MLPA
GG158	PRKRA gene sequencing	NGS
GG159	Progressive myoclonic epilepsy gene panel	NGS
GG160	PSEN1 gene sequencing	NGS
GG161	Pyridoxine-dependent epilepsy (ALDH7A1) gene analysis	NGS
GG162	RANBP2 gene sequencing	NGS
GG163	Rett Syndrome (MECP2) deletion and duplication analysis	MLPA
GG164	Rett Syndrome gene panel	NGS
GG165	RSRC1 gene sequencing	NGS
GG166	SACS gene sequencing	NGS
GG167	Sandhoff disease (HEXB) gene sequencing	NGS
GG168	Septo-Optic Dysplasia Panel	NGS
GG169	SETX gene sequencing	NGS
GG170	SMN and NAIP deletion and duplication analysis	MLPA
GG171	Spastic Paraplegia gene Panel	NGS
GG172	Spinal Muscular Atrophy (SMN1) gene analysis	Sanger
GG173	Spinal Muscular Atrophy (SMN1 and SMN2) deletion and duplication analysis	MLPA
GG174	Spinal Muscular Atrophy gene Panel	NGS
GG175	Spinocerebellar ataxia 1 (ATXN1) repeat expansion analysis	STR
GG176	Spinocerebellar ataxia 12 (PPP2R2B) repeat expansion analysis	STR
GG177	Spinocerebellar ataxia 2 (ATXN2) repeat expansion analysis	STR
GG178	Spinocerebellar ataxia 3 (ATXN3) repeat expansion analysis	STR
GG179	Spinocerebellar ataxia 6 (CACNA1A) repeat expansion analysis	STR
GG180	Spinocerebellar ataxia 7 (ATXN7) repeat expansion analysis	STR
GG181	Spinocerebellar ataxia repeat expansion analysis: SCA1, SCA2, SCA3, SCA6, SCA7,SCA12	STR
GG182	SPTBN2 gene sequencing	NGS
GG183	Tay-Sachs disease (HEXA) deletion and duplication analysis	MLPA
GG184	Tay-Sachs disease (HEXA) gene analysis	NGS
GG185	TBC1D24 gene analysis	NGS
GG186	TCF4 gene sequencing	NGS
GG187	TIA1 and DES genes sequencing	NGS
GG188	TSC1 and TSC2 gene analysis	NGS
GG189	TSC1 deletion and duplication analysis	MLPA
GG190	TSC2 deletion and duplication analysis	MLPA
GG191	Tuberous Sclerosis gene Panel	NGS
GG192	X-linked adrenoleukodystrophy (ABCD1) deletion and duplication analysis	MLPA
GG193	X-linked Intellectual Disability gene Panel	NGS
GG194	X-linked spastic paraplegia-2 (PLP1) deletion and duplication analysis	MLPA

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

MICROGEN™ PrecisionDx

The Future of Targeted Treatment

NEUROGENETIC TESTING

About the Test

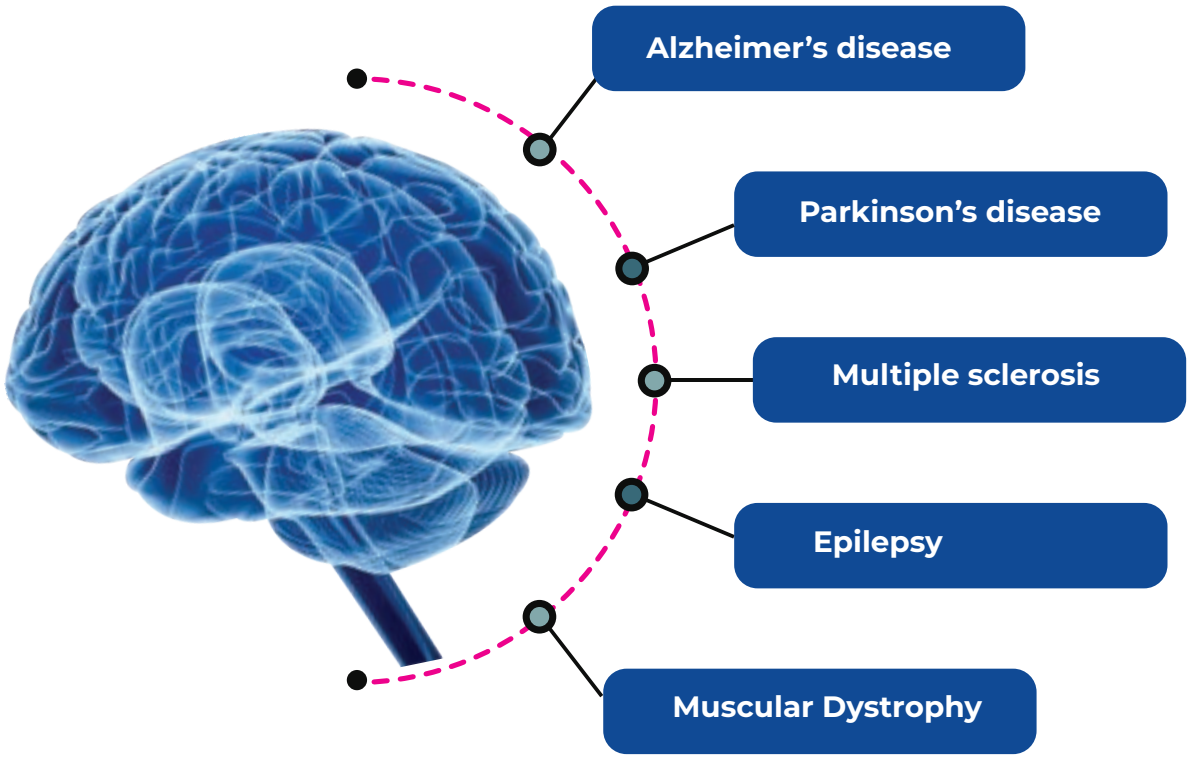
Neurological disorders (NDs) are a leading cause of disability and illness worldwide, with a significant impact on quality of life. These disorders affect various parts of the nervous system, from the brain to peripheral nerves, often stemming from structural, biochemical, or electrical abnormalities.

Many neurological disorders have a genetic component, influenced by both inherited traits and environmental factors. Genetic testing has emerged as a crucial tool for diagnosing these conditions, providing insights into risk assessment and familial inheritance patterns. Thus, a more precise diagnosis of many neurological disorders is now possible, and genetic testing can be considered earlier in the diagnostic procedure.

Common
Neurological Disorders

Examples include Alzheimer's disease, Parkinson's disease, multiple sclerosis, epilepsy, and muscular dystrophy, among others. These conditions can lead to diverse symptoms and disabilities, affecting individuals differently based on genetic and environmental factors.

In 2021, the top ten neurological conditions contributing to health loss included stroke, dementia, migraine, and epilepsy. While neurological disorders generally affect men more, conditions like migraine and dementia disproportionately impact women.



When to get tested
for Neurological Disorders?

- **Early Clinical Signs:** Testing is warranted when there are initial signs indicating specific neuroanatomical changes or disease pathology.
- **Age of Onset:** Testing is crucial to determine the age at which symptoms first appear, aiding in disease identification and management.
- **Mode of Inheritance:** Understanding the genetic inheritance pattern (autosomal dominant, autosomal recessive, X-linked, etc.) is essential for diagnosis and familial risk assessment.
- **Extra-Neural Signs:** Testing is recommended when there are additional signs affecting organs outside the nervous system (e.g., eyes, skin, connective tissues, visceral organs).

Why is it important to test for **Neurological Disorders?**

- **Prevalence in Pediatric Neurology:** NDs are relatively common in pediatric practice, often involving Mendelian inheritance patterns affecting the nervous system.
- **Accurate Diagnosis:** Through neuroimaging, biochemical assays, and genetic tests, NDs can be accurately diagnosed, enabling tailored treatment plans and interventions.

Who should consider **Neurogenetic testing?**

- **Individuals with Neurological Symptoms:** Testing is appropriate for anyone presenting with symptoms such as developmental delays, seizures, movement disorders, or cognitive impairments.
- **Individuals with Family History:** Those with a family history of neurological disease should consider testing to assess genetic risk and potential inheritance.
- **Individuals with Sporadic Symptoms:** Even in the absence of family history, testing may be necessary when symptoms strongly suggest a specific genetic neurological disorder.

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

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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