



Pathway Diagnostic  
Laboratories

# HEREDITARY NEUROLOGICAL DISORDERS RISK TESTING

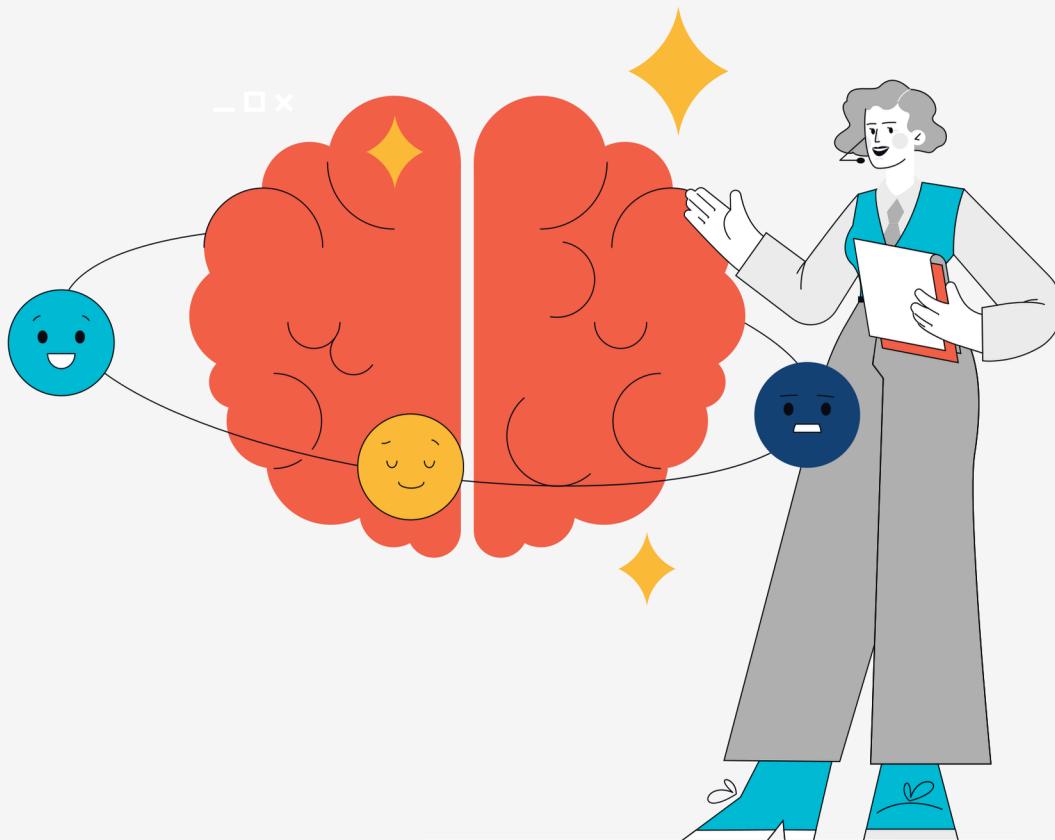


**PATHWAY DIAGNOSTICS LABORATORIES**

# INFORMED DECISION-MAKING: WHY GENETIC TESTING IS CRITICAL FOR MANAGING NEUROLOGICAL DISORDERS

Genetic testing for neurological disorders involves analyzing an individual's DNA to identify genetic variations that may contribute to or cause a neurological condition.

The types of genetic testing available include diagnostic testing, which looks for specific genetic mutations known to cause a particular disorder, and predictive testing, which assesses an individual's risk of developing a condition based on their genetic makeup. Genetic testing can also be used for carrier testing to determine if an individual carries a genetic mutation that could be passed on to their children.



In some cases, genetic testing may not provide a clear diagnosis, but it can help narrow down the possible causes of a neurological condition and inform treatment options.

It is important to note that genetic testing is not always necessary or appropriate for every neurological disorder, and it should be carefully considered in consultation with a healthcare provider or genetic counselor.

# GENETIC TESTING FOR NEUROLOGICAL DISORDERS CAN PROVIDE SEVERAL BENEFITS FOR PHYSICIANS AND PATIENTS, INCLUDING:

## EARLY DIAGNOSIS:

Genetic testing can identify genetic mutations that are associated with neurological disorders before the onset of symptoms. This allows for early diagnosis, which can lead to earlier interventions and better outcomes.



## ACCURATE DIAGNOSIS:

Genetic testing can help to confirm a diagnosis, especially when symptoms are non-specific or when there is uncertainty about the underlying cause of the disorder.

## PERSONALIZED TREATMENT:

Genetic testing can identify specific genetic mutations that may respond better to certain treatments, allowing for personalized treatment plans that are tailored to the patient's individual needs.



## FAMILY PLANNING:

For patients with genetic mutations associated with neurological disorders, genetic testing can provide information about the risk of passing on the condition to their children. This can inform family planning decisions and help to prevent the transmission of the disorder to future generations.

## PROGNOSTIC INFORMATION:

Genetic testing can provide prognostic information about the likely course of the disorder, which can help patients and their families prepare for the future.



## CLINICAL TRIALS:

Genetic testing can identify patients who may be eligible for clinical trials of new treatments, which can provide access to novel therapies that may improve outcomes.

Overall, genetic testing for neurological disorders can provide valuable information for physicians and patients, leading to earlier and more accurate diagnoses, personalized treatment plans, and improved clinical outcomes.

# THERE ARE SEVERAL DISEASES AND CONDITIONS RELATED TO GENETIC MUTATIONS IN NEUROLOGICAL RISK GENES.

## SOME OF THE MOST WELL-KNOWN ONES INCLUDE:

### ALZHEIMER'S DISEASE:

Mutations in genes such as PSEN1 and PSEN2 can increase the risk of developing Alzheimer's disease, a progressive and debilitating neurodegenerative disorder that affects memory, thinking, and behavior.

### HUNTINGTON'S DISEASE:

Huntington's disease is a hereditary disorder caused by mutations in the HTT gene. This disease leads to the degeneration of nerve cells in the brain, causing involuntary movements, cognitive impairment, and psychiatric symptoms.

### PARKINSON'S DISEASE:

Mutations in gene LRRK2 have been linked to an increased risk of developing Parkinson's disease, a progressive disorder that affects movement and coordination.

### ATAXIA DISEASE:

Ataxia is a neurological condition that affects coordination and balance. It is caused by damage to the cerebellum, a part of the brain that plays an important role in controlling movement. Mutations in the following genes can lead to the accumulation of abnormal proteins in the cerebellum, which can cause damage and ultimately result in ataxia: ATXN1, ATXN2, ATXN3, CACNA1A, FXN, and TBP.

### ALS (AMYOTROPHIC LATERAL SCLEROSIS):

ALS is a progressive and fatal neurodegenerative disease that affects nerve cells in the brain and spinal cord. Mutations in genes such as SOD1 and TARDBP have been linked to an increased risk of developing ALS.

### CHARCOT-MARIE-TOOTH DISEASE:

This is a group of inherited disorders that affect the peripheral nerves, causing muscle weakness and atrophy. Mutations in genes such as PMP22, GJB1, and MPZ have been linked to different forms of Charcot-Mrie-Tooth disease.

### EPILEPSY:

Mutations in genes such as SCN1A, SCN2A, and GABRG2 have been linked to an increased risk of developing different forms of epilepsy, a neurological disorder that causes seizures.

### RETT SYNDROME:

Rett syndrome is a rare genetic disorder that affects the development of the brain, leading to intellectual disability, seizures, and other neurological problems. It is caused by mutations in the MECP2 gene.



# GENE PANEL FOR HEREDITARY NEUROLOGICAL DISORDERS RISK (164-GENE PANEL )

ADNP, AFF2, ALDH7A1, ANG, APTX, ARX, ASPA, ASXL1, ATN1, ATP1A2, ATP7B, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BCL11A, BSCL2, C12orf4, CACNA1A, CACNA1C, CC2D1A, CDKL5, CHD2, CNOT3, CNTN6, COL4A1, COL4A3BP, CSNK2A1, CSTB, CTNND2, DHCR7, DPYD, EGR2, EHMT1, EN2, EZH2, FBXO11, FMR1, FOXG1, FOXP1, FTSJ1, FXN, GABRG2, GAMT, GARS, GATM, GBA, GCH1, GRIN2A, GRN, HEXA, HFE, HSPB1, HTT, IKBKAP, KCNQ2, KDM5C, L1CAM, LRRK2, MAPT, MBOAT7, MECP2, MED12, MTHFR, MTM1, NDP, NDUFA1, NLGN3, NLGN4X, NOTCH3, NSD1, NTRK1, NTRK2, PABPN1, PCDH19, PDGFB, PDHA1, PIK3CA, PINK1, PMP22, PNKD, POLG, PPP2R2B, PRRT2, PSEN1, PTEN, REEP1, SCN1A, SCN1B, SCN2A, SCN8A, SCO2, SGCE, SLC16A2, SLC2A1, SLC6A8, SLC9A6, SMN1, SMN2, SOD1, SPG11, STXBP1, SYNGAP1, TARDBP, TBP, TCF4, TH, THAP1, TOR1A, TPP1, TSC1, TSC2, TTR, UBA1, ZEB2, ZNF41, ACADM, APOE, APP, ARSA, ATM, BCKDHA, BCKDHB, BCS1L, BLM, C10orf2, COQ2, COX10, DGUOK, ERBB4, FANCC, FUS, G6PC, GAA, GALT, GBE1, GJB1, HBB, MCOLN1, MFN2, MPV17, MPZ, NPC1, OPA1, OPTN, PAH, PDSS2, PLCG2, POLG2, PRNP, PSEN2, RRM2B, SCO1, SETX, SLC25A4, SPAST, SPTLC1, SUCLA2, SUCLG1, TAZ, TK2, TYMP.

## TEST SPECIFICATIONS

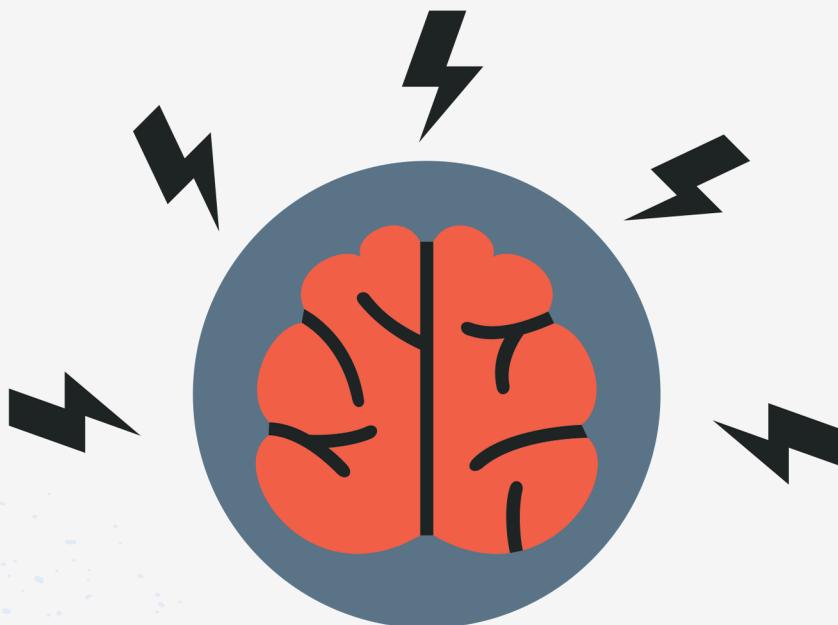
**Acceptable sample requirements**  
Buccal Swab or Saliva

**Turnaround time - 2-3 weeks**  
Coverage  $>96\%$  at 20x



**Reporting**  
Likelypathogenic and Pathogenic variants

**Customization**  
Customizable Gene List



# WHAT TO DO WHEN HEREDITARY NEUROLOGICAL RISKS GENETIC TESTING COMES BACK POSITIVE: **UNDERSTANDING THE IMPLICATIONS AND TAKING ACTION**

*If a doctor receives a positive report for the genetic testing of hereditary neurological disorders, there are several next steps they may take:*

## **CONFIRMING THE DIAGNOSIS:**

If a genetic mutation is identified, doctors may confirm the diagnosis through further testing or clinical evaluation to determine the extent of the disease or condition.

## **INFORM THE PATIENT AND FAMILY:**

The doctor will inform the patient and their family members of the diagnosis and the implications of the genetic test results. They will also discuss the possibility of genetic counseling.



## **DEVELOP A TREATMENT PLAN:**

The doctor will develop a treatment plan tailored to the specific disorder and the patient's individual needs. This may include medications, therapies, and lifestyle changes.

## **MONITOR THE PATIENT:**

The doctor will monitor the patient's condition over time to ensure that the treatment plan is effective and to adjust it as necessary.

## **OFFER GENETIC COUNSELING:**

The doctor may refer the patient and their family members to a genetic counselor to discuss the risk of passing on the disorder to future generations and options for family planning.

It is important to note that there is currently no cure for most hereditary neurological disorders, but early diagnosis and appropriate management can help to slow the progression of the disorder and improve the patient's quality of life.

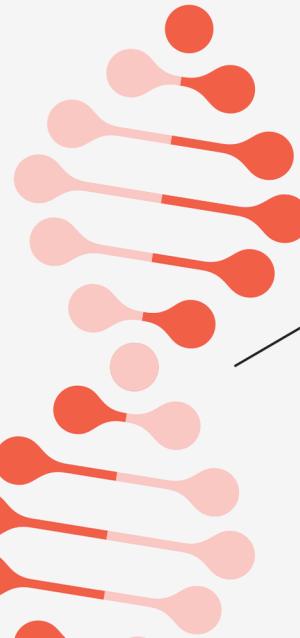
# UNLOCKING THE SECRETS OF YOUR GENETIC HEALTH COMPREHENSIVE GENETIC TESTING AT **PATHWAY DIAGNOSTICS LABORATORIES**

**PATHWAY DIAGNOSTICS LABORATORIES MAY BE A GOOD CHOICE  
FOR GENETIC TESTING FOR SEVERAL REASONS:**



## **HIGH-QUALITY TESTING**

Pathway Diagnostics Laboratories uses advanced technology and have experienced technicians to ensure that the testing is performed to the highest standards

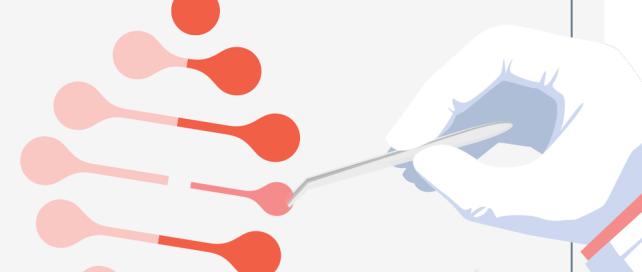


Pathway Diagnostics Laboratories has a fast turn around time for test results. This means that doctors can quickly get the information they need to make informed decisions about their patients' treatment.



## **COMPREHENSIVE TESTING OPTIONS**

We offer a wide range of genetic testing options, including tests for Diabetes predict, Cancer genetics, Cardiovascular, Drug-Gene Interactions and Neurological disorders.



## **COLLABORATIVE APPROACH**

Pathway Diagnostics Laboratories works closely with doctors to ensure that the testing is tailored to the specific needs of each patient. They provide personalized support and guidance throughout the testing process.



# DECODING THE MYSTERY OF GENETIC TESTING: A PATIENT'S ROADMAP TO BETTER HEALTH

Genetic testing is a medical test that examines your DNA to identify any changes or mutations that may be associated with a specific disease or condition. This guide aims to provide you with an overview of genetic testing, its significance, testing outcomes, and how it could be beneficial to you and your family members.

## WHAT IS **GENETIC TESTING?**

DNA is the genetic material that contains the instructions for the development, growth, and function of all living organisms. Genetic testing can provide information about inherited diseases or conditions, predispositions to certain diseases, and the likelihood of passing these conditions onto your children.

## WHY IS **GENETIC TESTING SIGNIFICANT?**

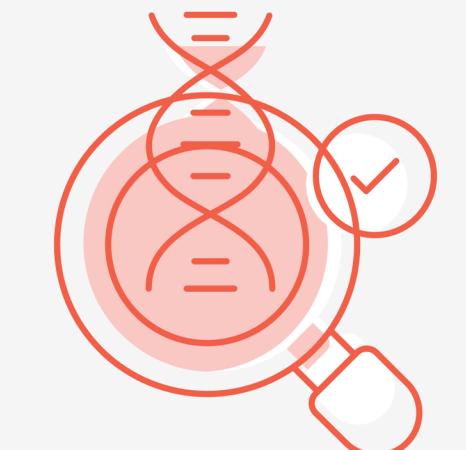
*Genetic testing can provide valuable information that can help you and your healthcare provider make informed decisions about your health. It can help to:*

### **DIAGNOSE GENETIC DISEASES:**

Genetic testing can identify changes or mutations in genes that are associated with specific diseases or conditions. This information can help healthcare providers diagnose genetic diseases and develop appropriate treatment plans.

### **INFORM REPRODUCTIVE DECISIONS:**

Genetic testing can help individuals and couples make informed decisions about family planning. If a genetic mutation is identified, it can be used to determine the likelihood of passing the condition onto future children.

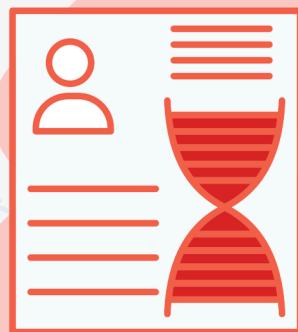


### **DETERMINE THE RISK OF DEVELOPING A DISEASE:**

Some genetic mutations are associated with an increased risk of developing certain diseases or conditions. Genetic testing can help identify these mutations, allowing for early intervention or prevention.

### **PERSONALIZE TREATMENT PLANS:**

Genetic testing can provide information about how an individual may respond to certain medications or treatments. This information can help healthcare providers personalize treatment plans for better outcomes.



## TESTING OUTCOMES

The results of genetic testing can be positive, negative, or uncertain. A positive result indicates that a genetic mutation associated with a specific disease or condition was identified. A negative result means that no mutations were identified. An uncertain result means that the test did not provide a definitive answer and further testing may be necessary.



## HOW CAN GENETIC TESTING BE BENEFICIAL TO YOU AND YOUR FAMILY MEMBERS?

*Genetic testing can be beneficial to you and your family members in several ways, including:*

### EARLY DETECTION AND TREATMENT:

Genetic testing can help identify conditions at an early stage, allowing for earlier treatment and better outcomes.



### FAMILY PLANNING:

Genetic testing can help individuals and couples make informed decisions about family planning, such as whether to have children or how to manage the risk of passing on a genetic condition.



### RISK REDUCTION:

Genetic testing can help identify individuals at increased risk for certain diseases or conditions, allowing for early intervention and lifestyle changes to reduce the risk.



### PERSONALIZED TREATMENT:

Genetic testing can provide information about how an individual may respond to certain medications or treatments, allowing for personalized treatment plans for better outcomes.



IF YOU ARE CONSIDERING GENETIC TESTING, IT IS IMPORTANT TO CHECK WITH YOUR HEALTH INSURANCE PROVIDER TO DETERMINE YOUR COVERAGE.

**YOU CAN DO THIS BY:**

**REVIEWING YOUR INSURANCE POLICY:**

Check your insurance policy or contact your insurance provider to see if genetic testing is covered and under what circumstances.

**CONSULTING WITH YOUR HEALTHCARE PROVIDER:**

Talk to your healthcare provider about whether genetic testing is medically necessary and covered by your insurance.

**SEEKING PRIOR AUTHORIZATION:**

Some insurance plans may require prior authorization for genetic testing, which means your healthcare provider will need to submit a request to your insurance provider for approval.



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