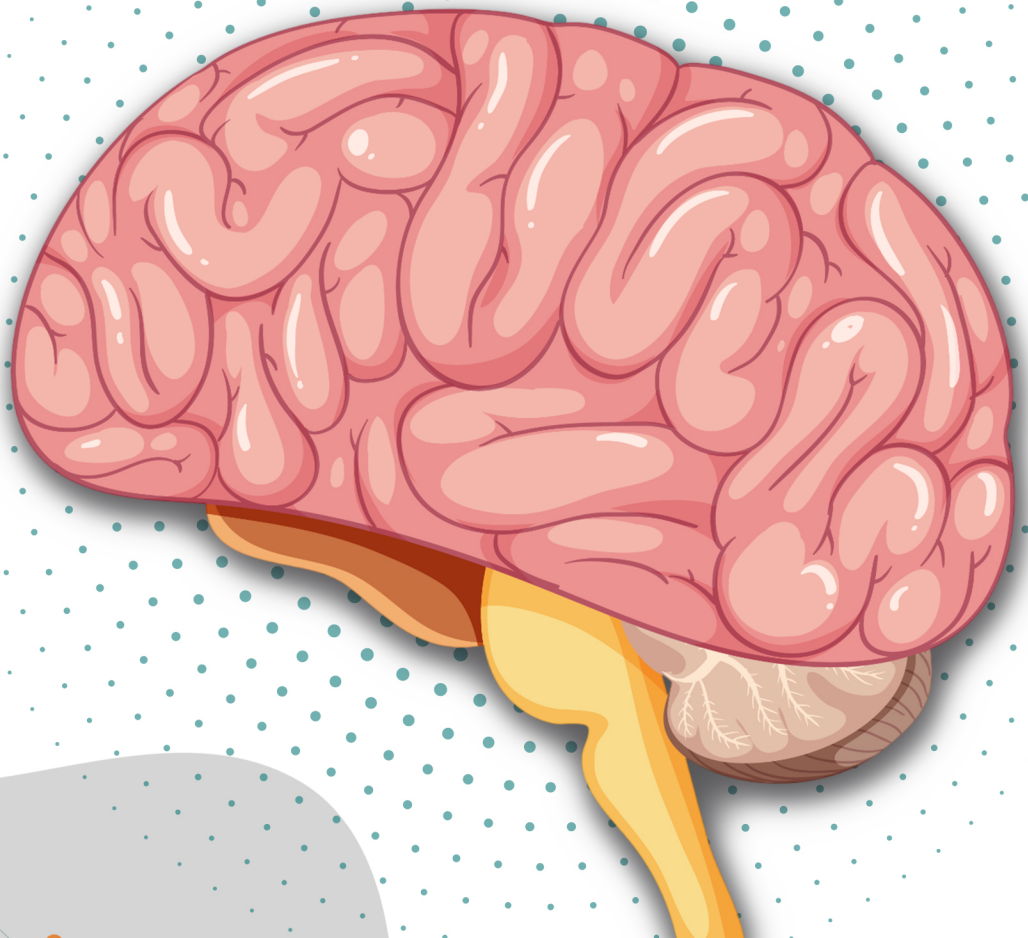


Comprehensive Genetics for **Neurological Conditions**



Comprehensive Neurological Panel

Many neurological conditions from developmental delays to movement disorders have a genetic origin. Our comprehensive neurology panel uses advanced genetic testing to identify changes in your DNA associated with a wide range of inherited neurological disorders. This precision allows for an accurate genetic diagnosis, enabling one self to make informed decisions about care, management, and family planning. With over **30 million*** people in India are living with neurological disorders, the need for clear answers has never been greater.

*PMID: 25591669

Panels Covered

Epileptic Disorders

Our testing can identify inherited mutations linked to both syndromic and non-syndromic epilepsy, helping guide more personalized treatment strategies.

Examples include: **Epileptic encephalopathy, Benign Infantile Familial Seizures (BIFS), Lennox-Gastaut Syndrome etc.**

Suitable Test: WES

Neuromuscular Disorders

Disorders that affect the peripheral nervous system. These disorders affect the ability to perform voluntary movements.

Example: **Limb Girdle Muscular Dystrophy; Spinal Muscular Atrophy; Duchenne Muscular Dystrophy etc.**

Suitable Test: MLPA, WES

Neurodegenerative Disorders

Inherited neurodegenerative conditions involve the gradual loss of neuron function. Identifying a genetic cause can help in understanding disease progression and in accessing targeted therapies or clinical trials.

Examples include: **Adrenoleukodystrophy, Canavan Disease etc.**

Suitable Test: WES

Neurometabolic Disorders

Caused by enzyme or vitamin deficiencies, these disorders often appear in infancy and affect brain development and function. Early genetic diagnosis is critical for timely dietary or medical management.

Examples include: **Maple Syrup Urine Disease, Neuronal Ceroid Lipofuscinosis, Krabbe Disease etc.**

Suitable Test: WES

Movement Disorders

Genetic movement disorders impact coordination, fluency, and control of body movements. Genetic testing helps clarify complex symptoms and supports accurate diagnosis and treatment planning.

Examples include: **Spinocerebellar Ataxia, Tourette's Syndrome, Juvenile Parkinsonism etc.**

Suitable Test: WES, Fragment Analysis

Mitochondrial Disorders

These are caused by mutations in mitochondrial or nuclear DNA and can affect multiple organs with highly variable symptoms. Genetic testing plays a crucial role in establishing an accurate and definitive diagnosis.

Examples include: **Mitochondrial Myopathy, Myoclonic Epilepsy with Ragged Red Fibers, Leber's hereditary optic neuropathy, etc.**

Suitable Test: WES-Mt DNA

Intellectual Disability & GDD

Analysing genes associated with neurodevelopment provides precise molecular diagnosis for the patient this explains the cause of developmental challenges and associated features.

Examples include: **Angelman , Sotos , Rett , Prader-Willi and Fragile X syndrome, etc.**

Suitable Test: WES, CMA, Fragment Analysis

Autism Spectrum Disorder

SNVs, CNVs and indel gene variants cause disturbed synaptic function and chromatin remodeling resulting in the condition. Panel analyses a curated set of genes known to be associated with ASD to provide families with outcome importance for early intervention.

Suitable Test: WES, CMA, FRAT

Neurocutaneous Disorders

Neurological disorders that have cutaneous manifestations.

These are lifelong conditions that can cause tumors to grow inside the brain, spinal cord, organs, skin, and bones.

Examples include: **Neurofibromatosis Type 1 & 2 (NF1, NF2), Tuberous Sclerosis Complex (TSC), etc.**

Suitable Test: WES

Who Should Consider Testing?

- Children with **developmental delays** (speech, motor, cognitive milestones).
- Individuals diagnosed with or suspected of **Autism Spectrum Disorder (ASD)**.
- Children with **intellectual disability** or **learning difficulties**.
- Unexplained **seizures/epilepsies**.
- **Behavioral conditions** with developmental concerns.
- Family history of **genetic or neurodevelopmental disorders**.
- Presence of **multiple congenital anomalies or dysmorphic features**.

When Should Testing Be Done?

- When developmental milestones are **delayed or regressing**.
- After a **clinical diagnosis of ASD or intellectual disability**.
- When standard investigations do not explain symptoms.
- Before planning further medical or therapeutic interventions.
- Planning pregnancy with a history of neurodevelopment disorder in the family.

Test specification

PARAMETERS	WES	CMA	FRAT	MLPA	Fragment Analysis
Platform	Illumina Novaseq 6000	Illumina Infinium GSA -24 v3.0	VMMR Sandwich ELISA	3500xL Genetic Analyzer	3500xL Genetic Analyzer
Variant Type	SNVs, CNVs, MtDNA Mutations	Copy Number Variants	Folate receptor α antibodies	Large Deletion/ Duplications	Triple Repeat Expansions
Sensitivity & Specificity	99 - 100%	99 - 100%	95%	99.9%	99.9%
Sample Type	Blood in EDTA tube	Blood in EDTA tube	Blood in Serum tube	Blood in EDTA tube	Blood in EDTA tube
Turn Around Time	4 weeks	3-4 weeks	2 weeks	3 weeks	3 weeks

Other Services



Pharmacogenomics

Genetics-guided drug selection



Preventive Genomics

Predicting health risks before symptoms appear




Reproductive Genomics

Pre-conception to Post-natal genetic care




Gutmicrobiome Test

Understanding gut health and balance

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SCAN TO
KNOW MORE

