



**Dhiti Omics Technologies**  
*Insights into health and disease*

Molecular Diagnostics and Genetic Testing



## About us

- Dhiti Omics Technologies is a precision molecular diagnostic company. We are an associate of Genotypic Technology, the pioneer of genomic services in India.
- We deliver ethical and actionable diagnostics to patients pinpointing genetic variants underlying a disease.
- We have a 2,300 sqft facility in Bangalore including a functional wet lab.
- Supported by the ISO 9001:2008 certified genomics facility of Genotypic Technology which houses state-of-the-art genomics platforms – RT-PCR, Microarray, Sanger Sequencer and multiple Next Generation Sequencers.
- Our in-house team of bioinformaticians, automated analysis pipelines and subject matter experts create actionable reports following ACMG guidelines.

## Success Stories in Clinics

### **Propionic Acidemia**

*Inborn error of metabolism*

Couple with recurrent infantile deaths

[Pathogenic mutation in PCCB gene](#)

### **Duchenne Muscular Dystrophy**

*Neuro-muscular disorder*

Young boys with hip muscle weakness, calf muscle hypertrophy, not able to run

[Pathogenic mutation in DMD gene](#)

### **Shah Waardenburg Syndrome**

*Auditory pigmentary Syndrome*

Young male with deafness, blue eyes, constipation

[Pathogenic mutations in SOX10 gene](#)

### **Uncomplicated Hereditary Spastic Paraplegia**

*Genetically diverse neurological disorder*

19 year old male with difficulty in walking, bladder disturbance and weakness in lower limbs

[Pathogenic mutation in SPAST gene](#)

### **GlutaricAcidemia Type IIB**

*Inborn error of metabolism*

4 year old child with developmental delay, normal EEG

[Novel mutation in ETFB gene](#)

### **Leigh Syndrome**

*Rare inherited neuro-metabolic disorder*

3 year old girl with motor regression, progressive imbalance, jerky movements during sleep

[Pathogenic Mutation in FOXRED1 & SURF1](#)

### **chr16p11.2 microdeletion**

3 year old female with macrocephaly, epilepsy, developmental delay and ataxia.

[Likely Pathogenic copy number loss in chr16p11.2](#)

### **Alport Syndrome**

51 year old female with hematuria, proteinuria and hypertension.

[Likely Pathogenic mutation in the COL4A5 gene](#)

### **Polycystic Kidney Disease**

19 year old female suspected to have ADPKD.

[Likely Pathogenic mutation in the PKD1 gene.](#)

### **Neuronal degeneration with Brain Iron Accumulation**

2 year old male with hypotonia and difficulty walking.

[Pathogenic mutation in the PLA2G6 gene.](#)

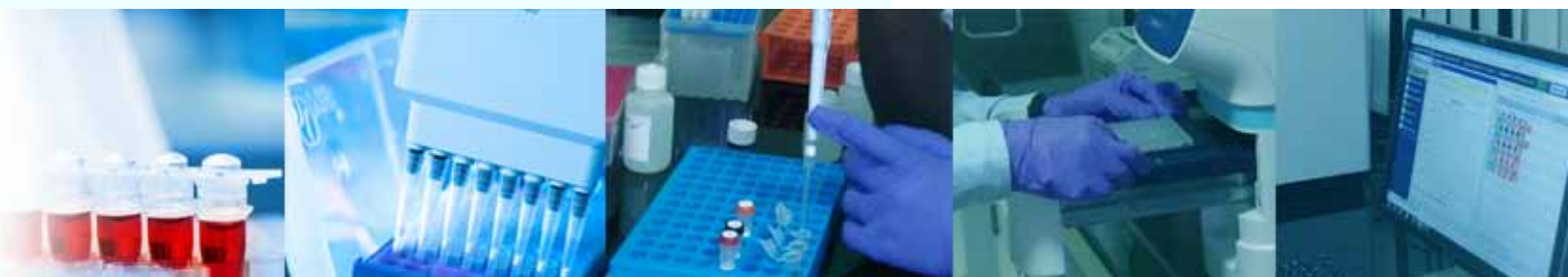
### **Atypical Rett Syndrome**

3.5 year old female with global developmental delay and speech delay.

[Pathogenic mutation in the MECP2 gene.](#)

## Available Diagnostic Tests

Name of Panel	What do we test?
<b>Inherited Disorder</b>	
Clinical Exome Profiling	Screening for 5000+ genes that are known to harbor disease-causing mutations
Inherited Diseases Panel	Tests for 552 genes which harbor mutations responsible for severe, recessive pediatric-onset inherited diseases
Whole Exome Sequencing	Sequences Coding regions of the entire genome
One Seq Panel	Detects mutations and copy number variations
<b>Copy Number Variations</b>	
CGH Array	Copy number changes including deletions and amplifications
<b>Nephrology</b>	
Inherited Common Nephrological Disorders	Tests for 57 genes associated with inherited renal disorders like Alport Syndrome, PKD, FSGS, aHUS, Hyperoxaluria, Nephrotic Syndrome, Medullary Cystic Disease
<b>Neurology</b>	
Neuromuscular Panel	This panel identifies point mutations as well as deletion and duplications in some common genes in neuromuscular disorders like Duchenne Muscular Dystrophy .
Mitochondrial Gene Sequencing	Sequences all mitochondrial genes
<b>Oncology</b>	
BRCA-1 & BRCA-2 -Mutation Profiling	Mutation profiling of BRCA 1 and BRCA 2 genes.
BRCA-1 & BRCA-2-MLPA Analysis	Del/Dup of BRCA 1 and BRCA 2 genes by MLPA
Tumour Hotspot Profiling	Screens hotspots in 48 well-known oncogenes and tumour suppressor genes to detect disease-causing mutations
<b>Cardiology</b>	
Cardiac Channelopathy Panel	Multiplexed assay for detecting mutations in 33 genes known to cause cardiac channelopathies.
<b>Metabolic Disorders</b>	
Metabolic Disorder Panel	A comprehensive diagnosis of more than 120 Metabolism errors. It screens coding regions of 300+ genes
<b>Carrier Testing</b>	
Carrier testing	Carrier Testing of a mutation in a family member of patient
<b>Pre-Natal Validation</b>	
Prenatal Validation	A sample for which the proband/parental mutation testing has already been done
<b>Single Gene Test</b>	
Beta thalassemia	Tests 5 most common hotspots specific to the Indian population
Single Gene Test	Test for specific genes
<b>Customised Gene Panel</b>	
Customized Gene Panel	Upon request, we can design specific gene panels
<b>Launching Soon</b>	
Autism Array	Detects copy number variation implicated in autism



## Why us?

- Team of experienced scientists with expertise in genomics in collaboration with subject matter experts
- Extensive bioinformatics analysis to identify genetic variant underlying the disease
- Currently working with leading clinics and hospitals in multiple cities

We also offer financing options for the tests in collaboration with a leading financial institution.

Write to us for more details.

## Shipment

### Sample Requirement

#### All tests (except Tumor Mutation Profiling)

5 ml blood, collected in EDTA tube and stored in refrigerator (2 – 8 °C)

#### Tumor Mutation Profiling

Frozen tissue sections or Formalin-fixed Paraffin-embedded (FFPE) tissue block

Tumor Mutation Profiling Trusted aid for an oncologist

Samples should have at least 70% tumor cell population

#### Pre-Natal Samples

Amniotic Fluid (20 ml)

Please send the samples to our Bangalore facility (address given below) in cool pack (2 – 8 °C)

To know more on the diagnostics services offered, write to us at [contact@dhitiomics.com](mailto:contact@dhitiomics.com) or call 8971278811

You can also register via [www.dhitiomics.com](http://www.dhitiomics.com) to have a specialist call you.



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