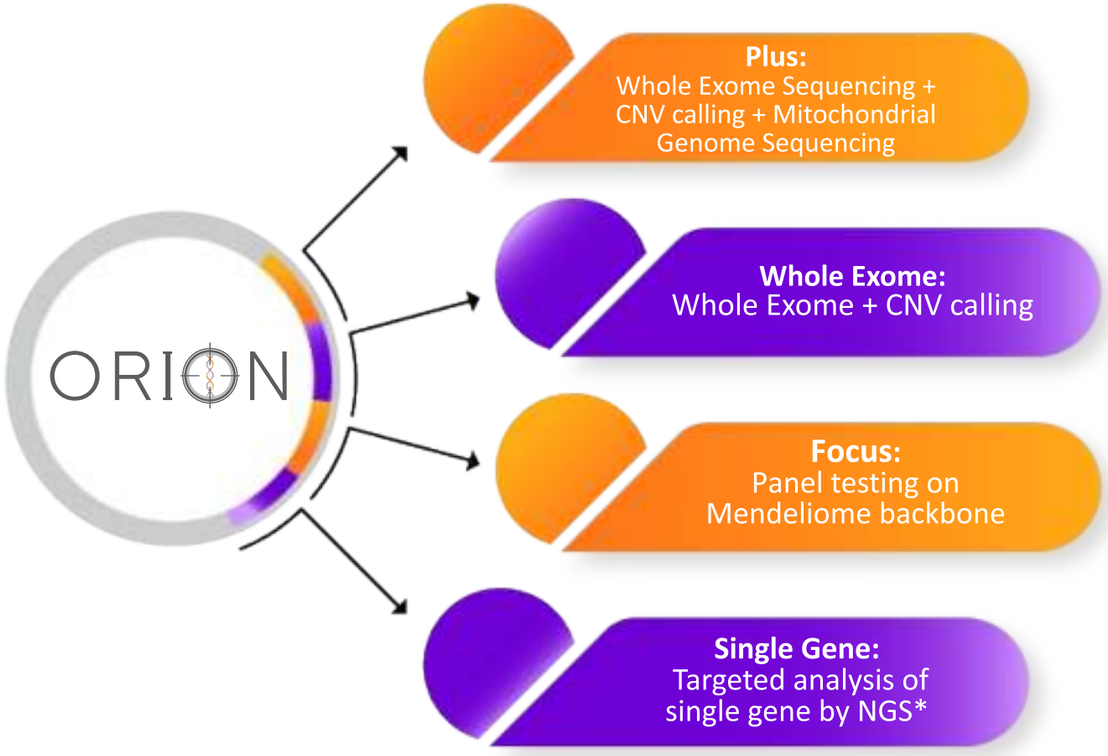
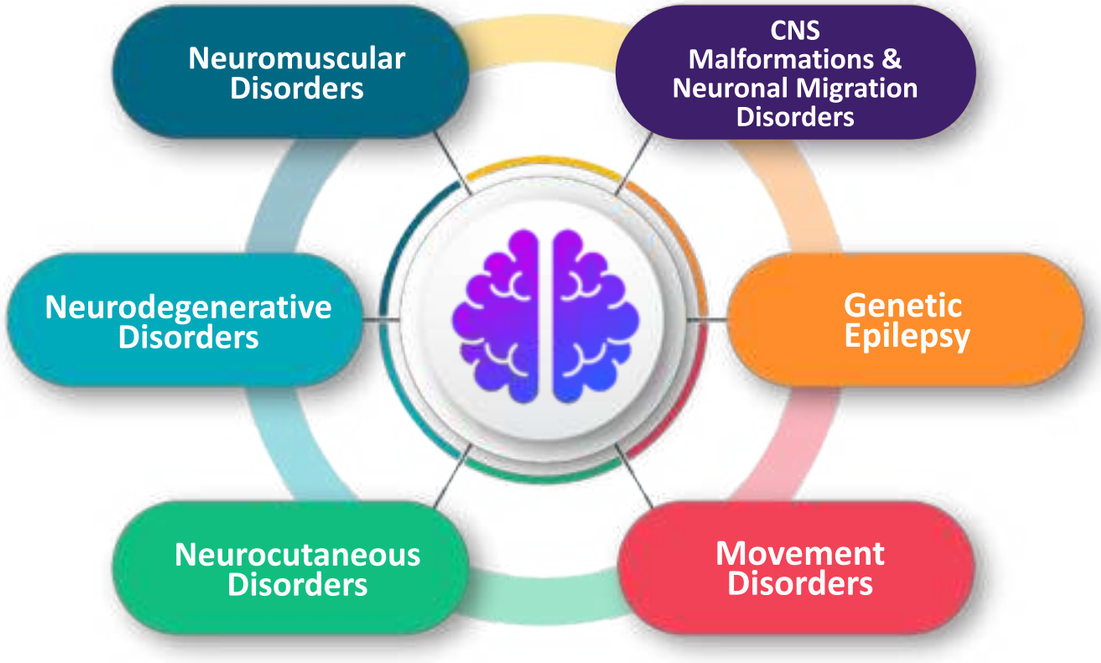
ORION
Intelligent Genomics

NEUROLOGY





*** Kindly contact the laboratory to check the coverage of the gene**

ORION Hightlights

Comprehensive Backbone

MORE THAN JUST EXOME!

Whole Exome

CNV Calling

Mitochondrial Genome

Customized Gene Curation:

TOWARDS EVIDENCE BASED MEDICINE

Phenotype Specific

Regularly Updated Gene Lists

Expert Variant Annotation:

ADDING RELEVANCE TO REPORTING!

Gene-Disease Association

Variant-Disease Association

Coverage:

INCLUSIVE OF

>19,000

>19,000 genes

100X

100X Mean Depth

>95%

>95% Coverage @20X

Enhanced Variant Calling:

ENRICHING ANALYSIS!

Customized Pipeline

Extensively Validated

Best-in-Class Reports:

PEER REVIEWED BY:

Bionformatician

PhD Scientist

Genetic Counselor

Clinical Geneticist

ORION Focus

Panel Testing on Mendeliome Backbone

CNS Malformations & Neuronal Migration Disorders

- Agenesis of Corpus Callosum
- Aqueductal Stenosis - X linked (MASA syndrome)
- Ciliopathies
- Craniosynostosis Syndromes
- Holoprosencephaly
- Lissencephaly
- Macrocephaly & Microcephaly
- Neuronal Migration Disorder
- Pontocerebellar Hypoplasia

Neurocutaneous Disorders

- Neurofibromatosis
- RASopathies
- Tuberous Sclerosis

Genetic Epilepsy

- Epileptic Encephalopathy
- Familial Hemiplegic Migraine
- Febrile Seizures
- Hyperekplexia
- Inborn Errors of Metabolism
- Infantile spasms
- Myoclonic Epilepsy

Movement Disorders

- Ataxia (Non Repeat Expansion type)
- Dystonia
- Hereditary Spastic Paraplegia
- Neurotransmitter Disorders
- Parkinsonism
- Periodic paralysis
- Paroxysmal dyskinesia

Neuromuscular Disorders

- Myopathies
- Muscular Dystrophy
- Myotonia Congenita and Periodic Paralysis
- Peripheral Neuropathies
- Spinal Muscular Atrophy (non-SMN type)

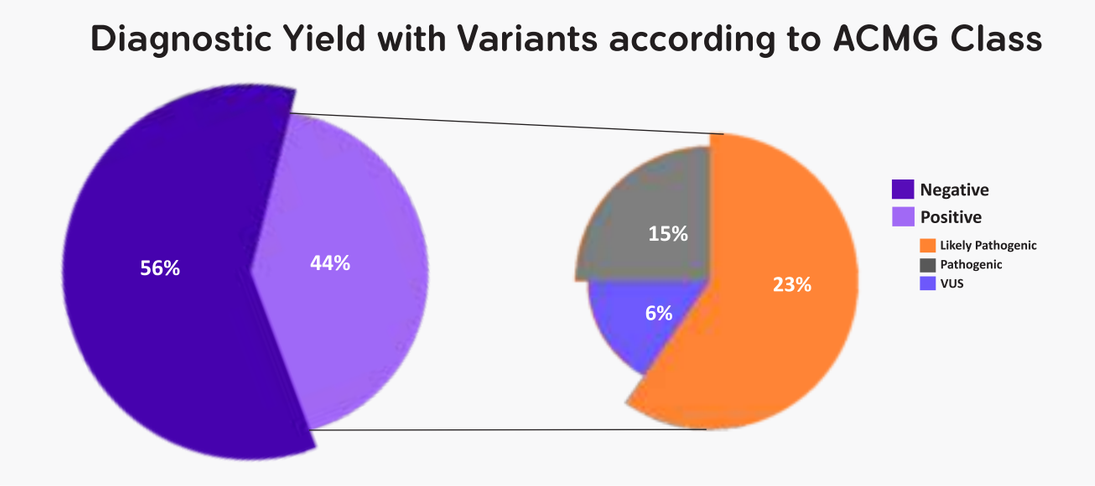
Neurodegenerative Disorders

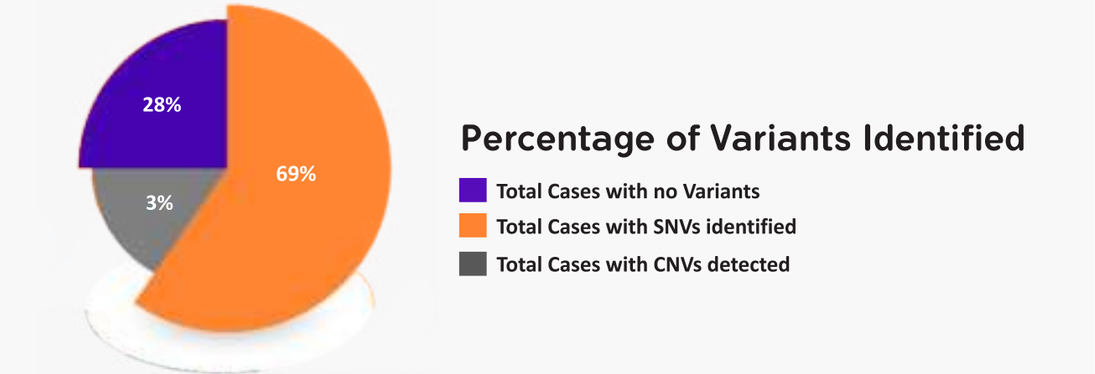
- Alzheimer and Dementia
- Amyotropic Lateral Sclerosis
- Basal Ganglia calcification
- Neurometabolic
- Leukodystrophy and Leukoencephalopathies
- Neurodegeneration with Brain Iron Accumulation (NBIA)

ORION Advantage

1. Improved Diagnostic Yield

Performance data at NCGM for Neurology (2020-2021): 1000 patients





2. Improved Diagnostic Potential

Proband: 9 months/ Female
Consanguinity: Yes
Clinical Indications: Global Developmental delay
Investigations: EEG showed low amplitude reads.
Family history: Non contributory

Gene & Transcript	Location	Variant	Zygosity	Classification	Disease	Inheritance
GOLGA2 (NM_004486.4)	Exon 22	c.2251>T (p.Gln751Ter)	Homozygous	Uncertain Significance	Unspecified / All Highly Penetrant Disorders	Autosomal Recessive

Case Report Publication

3. Enhanced CNV Calling

Proband: 1 year / Male
Consanguinity: Yes
Clinical Indications: Hearing loss, visual impairment, seizures, neuroregression, dysmorphism
Suspected Diagnosis: Zellweger syndrome
Whole Exome did not identify a single nucleotide variant **BUT**
A copy number variant was identified:

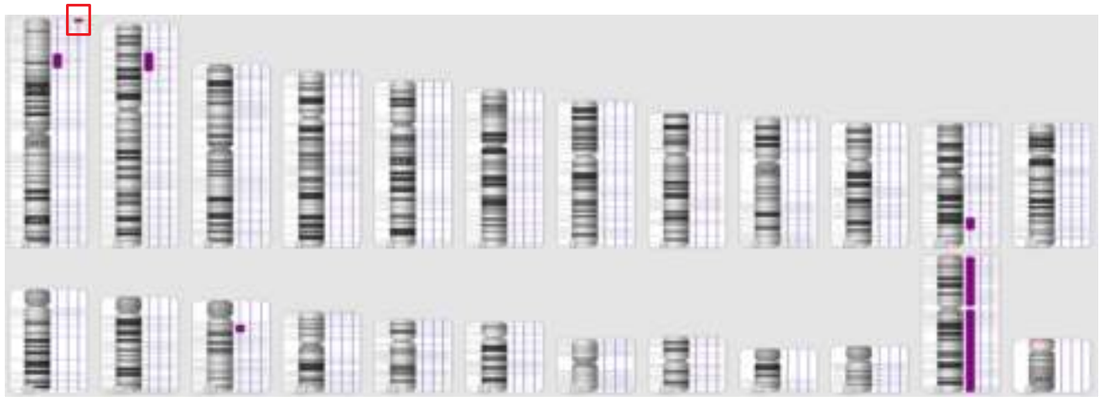
CNV Variant

Chromosome	Location	Copy Number Variation	Copy Number	Classification	Disease
Chromosome1	Chr:1_?861321_?3809560	Deletion	1	Pathogenic	Chromosome 1p36 deletion syndrome

Confirmation of the identified CNV by Chromosomal Microarray:

SEGMENT INFORMATION SUMMARY OF FINDINGS

Type	Chr	Size (kbp)	Marker count	OMIM Gene Count	Microarray Nomenclature	Interpretation
Loss	1	3765.356	257	56	arr[GRCh37] 1p36.33p36.32(849466_4614822)x1	Pathogenic



Validated For :

- Single nucleotide variants
- Indels upto 30bp
- Copy number variants: Multi Exon (more than 3 exons) deletions and duplications

Does Not Include:

- Triplet Repeat Expansions
- Methylation Abnormalities
- Somatic Mutations

Acceptable Samples:

- EDTA Whole Blood
- Dried Blood Spots (DBS)
- Amniotic fluid (AF)#
- Chorionic Villous Biopsy (CVS)#

We Also Offer:

- Fragile X (TP-PCR)
- Spinal Muscular Atrophy (MLPA)
- Duchenne Muscular Dystrophy (MLPA)
- Chromosomal Microarray (315k, 750k)

Turn Around Time for ORION : 28 Business Days

 # Kindly contact the lab to check prerequisites

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