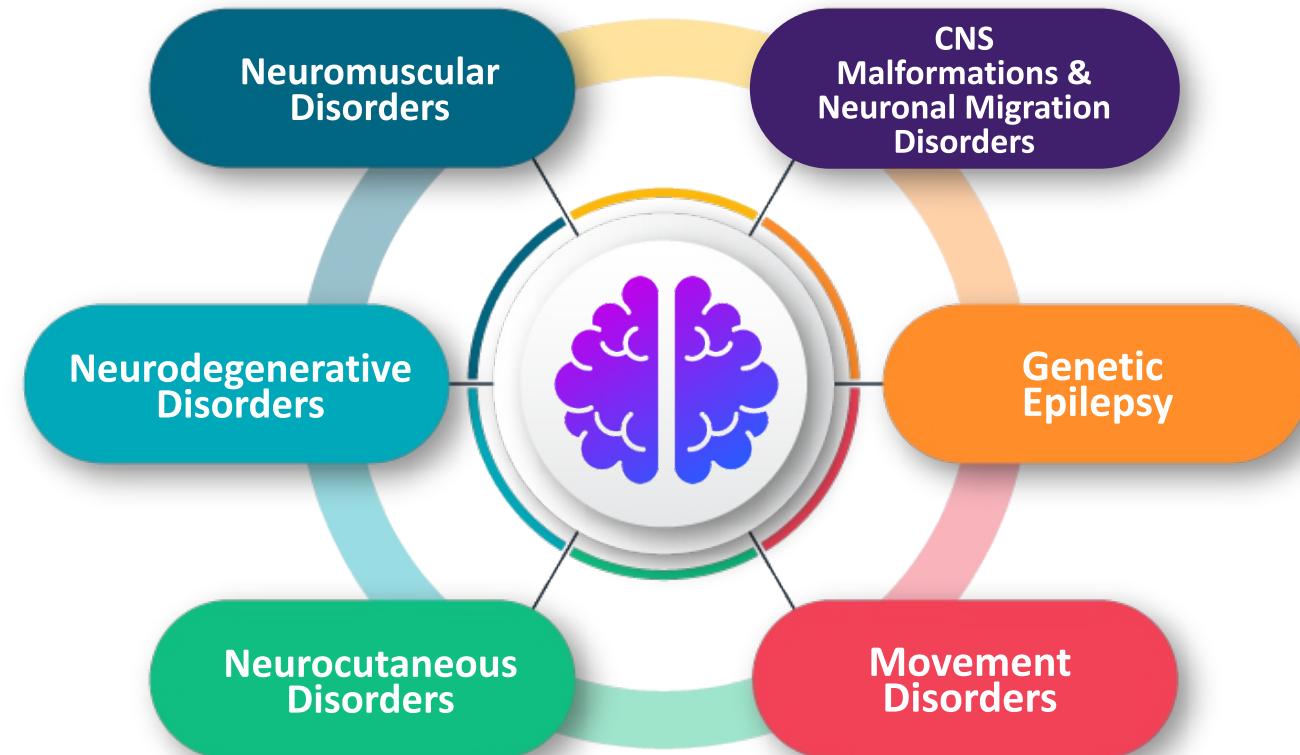
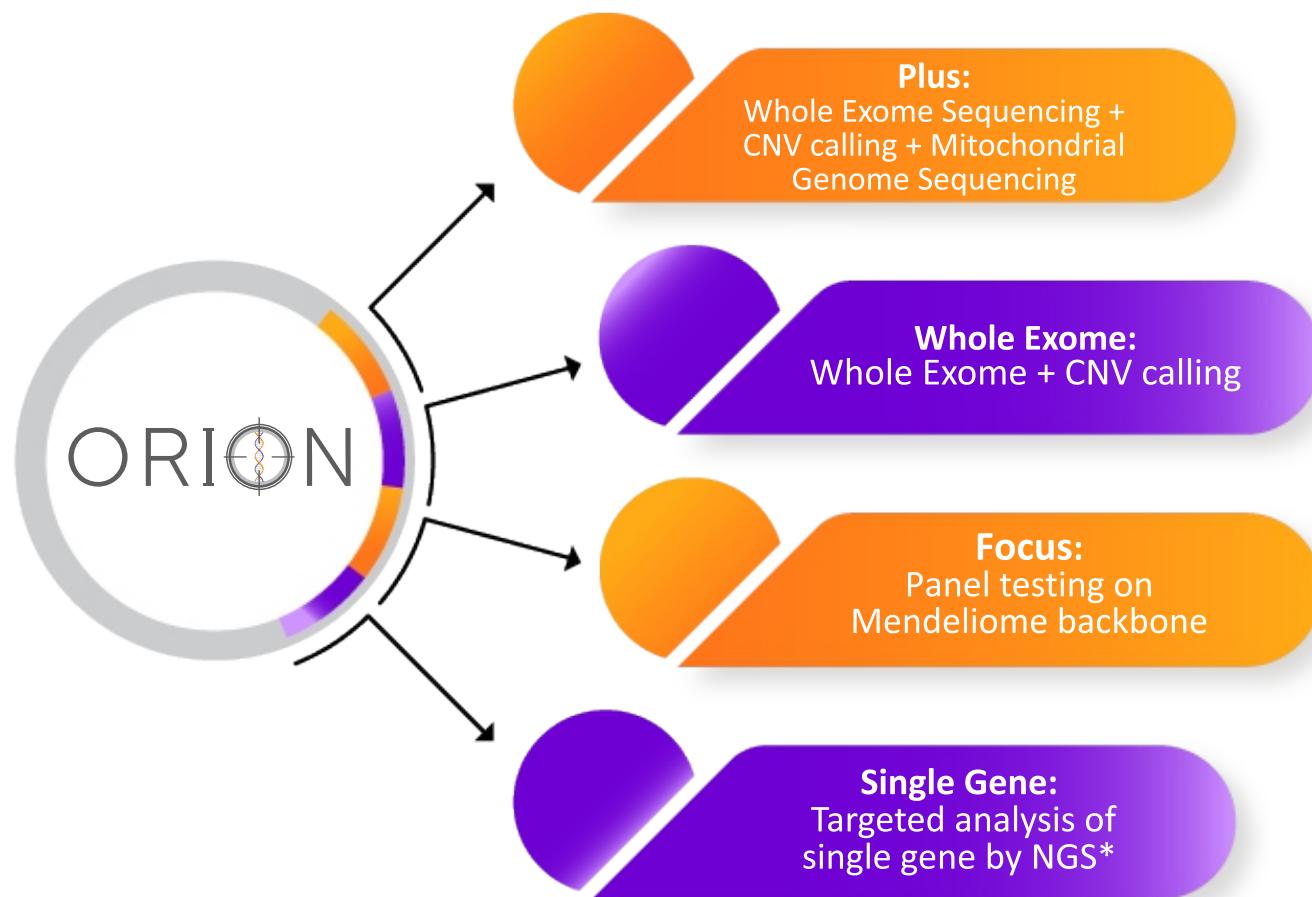




# NEUROLOGY



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

# ORION Highlights

## ● Comprehensive Backbone

MORE THAN JUST EXOME!



Whole Exome



CNV Calling



Mitochondrial Genome

## ● Coverage:

INCLUSIVE OF



>19,000 genes



100X Mean Depth



>95% Coverage @20X

## ● Customized Gene Curation:

TOWARDS EVIDENCE BASED MEDICINE



Phenotype Specific



Regularly Updated Gene Lists



Customized Pipeline



Extensively Validated

## ● Expert Variant Annotation:

ADDING RELEVANCE TO REPORTING!



Gene-Disease Association



Variant-Disease Association

## ● Best-in-Class Reports:

PEER REVIEWED BY:



Bioinformatician



PhD Scientist



Genetic Counselor



Clinical Geneticist

## 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 Paraparesis
- 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

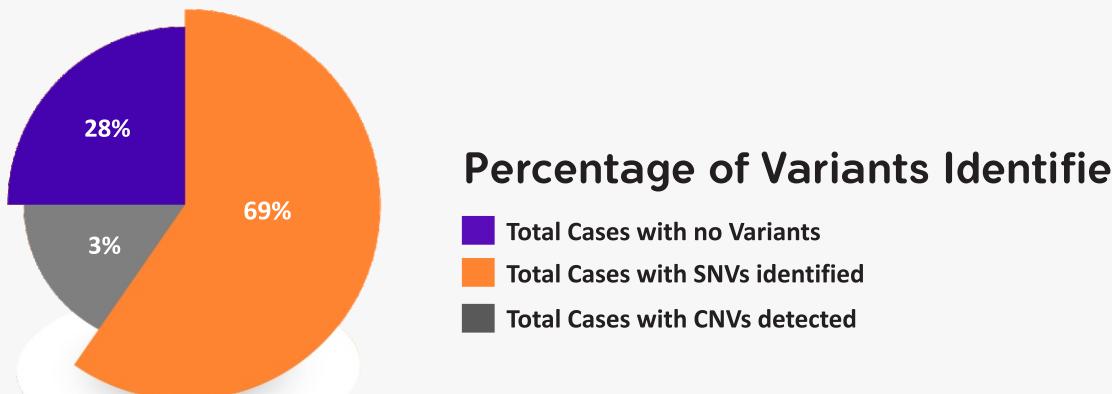
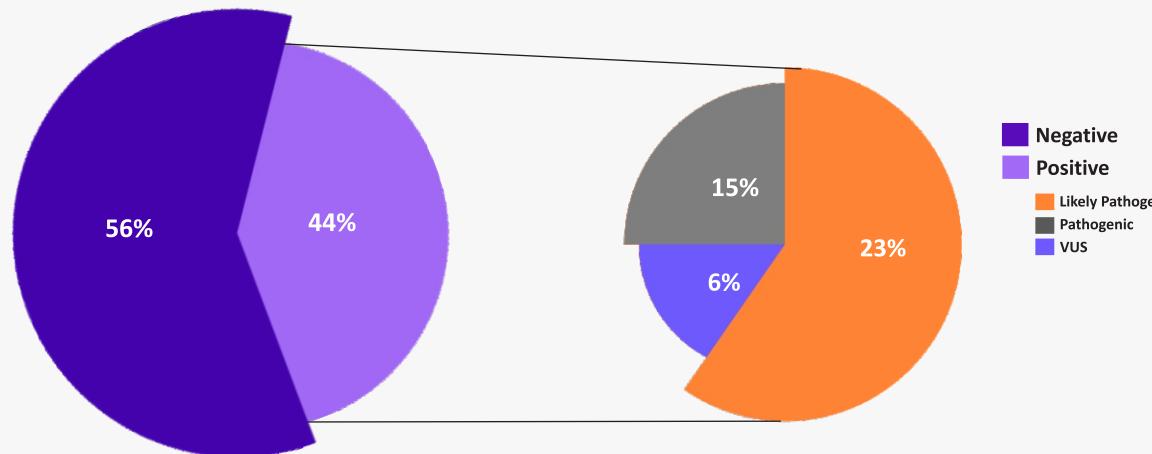
- Alzheimer and Dementia
- Amyotrophic Lateral Sclerosis
- Basal Ganglia calcification
- Neurometabolic
- Leukodystrophy and Leukoencephalopathies
- Neurodegeneration with Brain Iron Accumulation (NBIA)

# ORION Advantage

## 1. Improved Diagnostic Yield

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

### Diagnostic Yield with Variants according to ACMG Class



## 2. Improved Diagnostic Potential

Proband: 9 months/ Female

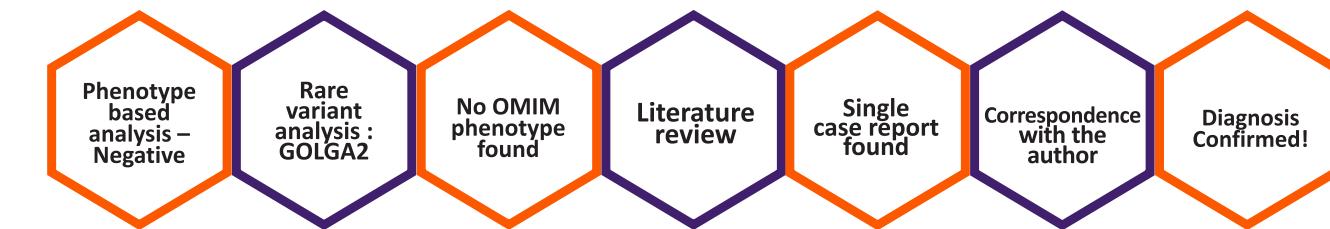
Consanguinity: Yes

Clinical Indications: Global Developmental delay

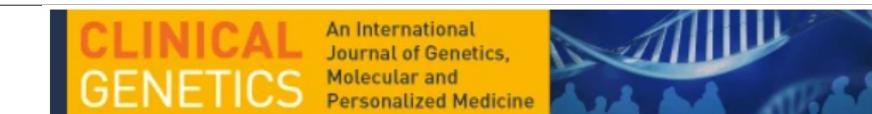
Investigations: EEG showed low amplitude reads.

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



SHORT REPORT

Bi-allelic loss of function variants in GOLGA2 are associated with a complex neurological phenotype: Report of a second family

Udhaya Kotecha, Mehul Mistri, Nidhi Shah, Parth S. Shah, Vandana A. Gupta

First published: 23 August 2021 | <https://doi.org/10.1111/cge.14053>

### 3. Enhanced CNV Calling

**Proband:** 1 year / Male

**Consanguinity:** Yes

**Clinical Indications:** Hearing loss, visual impairment, seizures, neuroregression, dysmorphism

**Suspected Diagnosis:** Zellweger syndrome

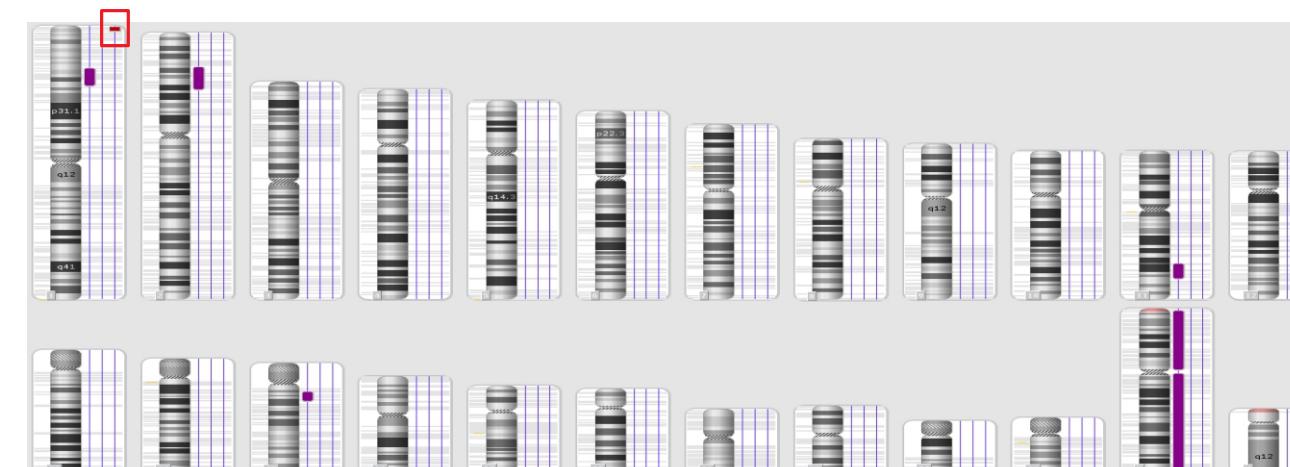
Whole Exome did not identify a single nucleotide variant **BUT**

**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)<sup>#</sup>
- Chorionic Villous Biopsy (CVS)<sup>#</sup>

#### 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

#### Genetic Testing from the Comfort of Your Home in 5 Steps



Enquire



Pre-Test  
Counselling



Take Your  
Test



Download  
Report



Post Test  
Counseling

Powered By



**Neuberg Center For Genomic Medicine (NCGM)**  
Near GTPL House, Opp. Armedia, Sindhu Bhavan Road,  
Bodakdev, Ahmedabad, Gujarat, India - 380059

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