OMI BIOMEDICS
THE POWER OF SCIENCE

OMI RareDx

Whole Genome Sequencing

Clinician Infopack

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Our Story

Our company was founded by life science experts who have personally experienced the challenges of the diagnostic odyssey. After years of grappling with inconclusive tests and missed diagnoses, we established OMI Biomedics to assist clinicians in identifying the root causes of disease early in the diagnostic journey. By collaborating with industry leaders and seasoned professionals with decades of scientific expertise, we remain dedicated to providing genetic testing solutions that can transform the lives of patients and their families. This is our calling.

Our Mission

We aim to bring clarity and support to patients and families on their journey toward a definitive diagnosis by providing high-quality genetic tests that enable healthcare providers to deliver evidence-based, personalized care for improved patient outcomes.

Our Vision

We envision a future where every patient who needs to can access clear answers to their most challenging health questions through the power of genetic testing. By partnering with healthcare professionals and empowering them with the tools and insights they need, we aspire to make genetic testing an integral part of modern medicine—streamlining diagnoses, improving treatment, and enhancing patient lives across the globe.

Unlock insights. Discover Possibilities.

Testing Options

1. OMI RareDx WGS Mono

Whole genome sequencing is performed exclusively on the proband, with the option for Sanger sequencing verification for family members if needed.

2. OMI RareDx WGS Trio

Whole genome sequencing is performed for both the proband and their parents. Data analysis and interpretation focus on the proband's phenotype, and clinical reports are generated accordingly. The parents' genome data is used as a reference to aid in the analysis.

Variants Under Our Scope

- SNV / Indels
- Exon CNV
- Structure variation (>30kb CNVs)
- Triploidy / aneuploidy
- Mitochondrial abnormalities
- Inversion / translocations
- Loss of heterozygosity >5Mb
- Uniparental disomy
- 8 dynamic mutations
- 26 pathogen infections

Product overview

Our genomic testing service uses whole genome sequencing (WGS), offering clinicians a reliable tool to detect a wide range of genetic variations in both nuclear and mitochondrial DNA. This test is designed to assist in identifying complex genetic disorders, especially in cases with suspected but unconfirmed genetic causes. With OMI RareDx WGS, clinicians are empowered by comprehensive data that enhances diagnostic accuracy and supports informed decisions for personalized patient care.

Pathogen microorganism	Classification	Affect Pregnancy	Remarks
Cytomegalovirus (CMV)	dsDNA virus	Y	Torch agent
Toxoplasma (TOX)	protozoan	Y	Torch agent
Herpes simplex virus (HSV)	dsDNA virus	Y	Torch agent
Parvovirus B19 (PVB 19)	ssDNA virus	Y	
Varicella-zoster virus (VZV)	dsDNA virus		
Treponema pallidum	spirochete	Y	Compulsory (GA:0-3, 6-13)
Candida	fungi		
Adenovirus	dsDNA virus		Detected in stillborn tissue
Enterococcus species (Enterococcus faecalis, Enterococcus faecium)	bacteria		Detected in stillborn tissue
Enterobacter sp	bacteria		Detected in stillborn tissue
Escherichia coli	bacteria		Detected in stillborn tissue
Fusobacterium nucleatum	bacteria		Detected in stillborn tissue
Group B Streptococcus species	bacteria		Detected in stillborn tissue; Compulsory (GA:33-36)
Group A Streptococcus species	bacteria		Detected in stillborn tissue
Human herpesvirus-7	dsDNA virus		Detected in stillborn tissue
Human herpesvirus-8	dsDNA virus		Detected in stillborn tissue
Mycoplasma genitalium	mycoplasma		Detected in stillborn tissue
Mycoplasma hominis	mycoplasma		Detected in stillborn tissue
Trypanosoma cruzi	protozoan		Maternal and infant vertical transmission
Plasmodium falciparum	protozoan		Maternal and infant vertical transmission
Klebsiella pneumoniae	bacteria		Detected in stillborn tissue
Staphylococcus aureus	bacteria		Detected in stillborn tissue
Listeria	bacteria		Causes miscarriage, stillbirth, premature birth, congenital infection
Ureaplasma urealyticum			
Chlamydia trachomatis			
Neisseria			

Key Parameters

Testing Depth	20X coverage	Mitochondria 200X coverage	Testing strategy
≥40X	≥95%	≥95%	PE100

≥99.9% specificity guaranteed for all reported variants

Advantages

Superior interpretation

Patient data is analysed and compared against variants in multiple open, commercial, and internal databases, with additional interpretation support from Congenica, the world's leading genomic data analysis platform. **Our team manually reviews each variant one by one to ensure that no potentially relevant variant is overlooked.**

Broad coverage

OMI RareDx Whole Genome Sequencing (WGS) provides comprehensive coverage of the entire genome, enabling the detection of a wide range of genetic variations across both coding and non-coding regions, including single nucleotide variants, structural changes, and mitochondrial DNA alterations.

Efficient results

A single OMI RareDx WGS test can yield more detailed results than single or multi gene panel testing, whole exome sequencing (WES), mitochondrial whole genome sequencing, chromosomal microarray analysis (CMA), and copy number variant sequencing (CNVSeq). This approach accelerates the diagnostic process for complex diseases, saving both time and costs associated with comprehensive diagnosis and treatment.

Expert support

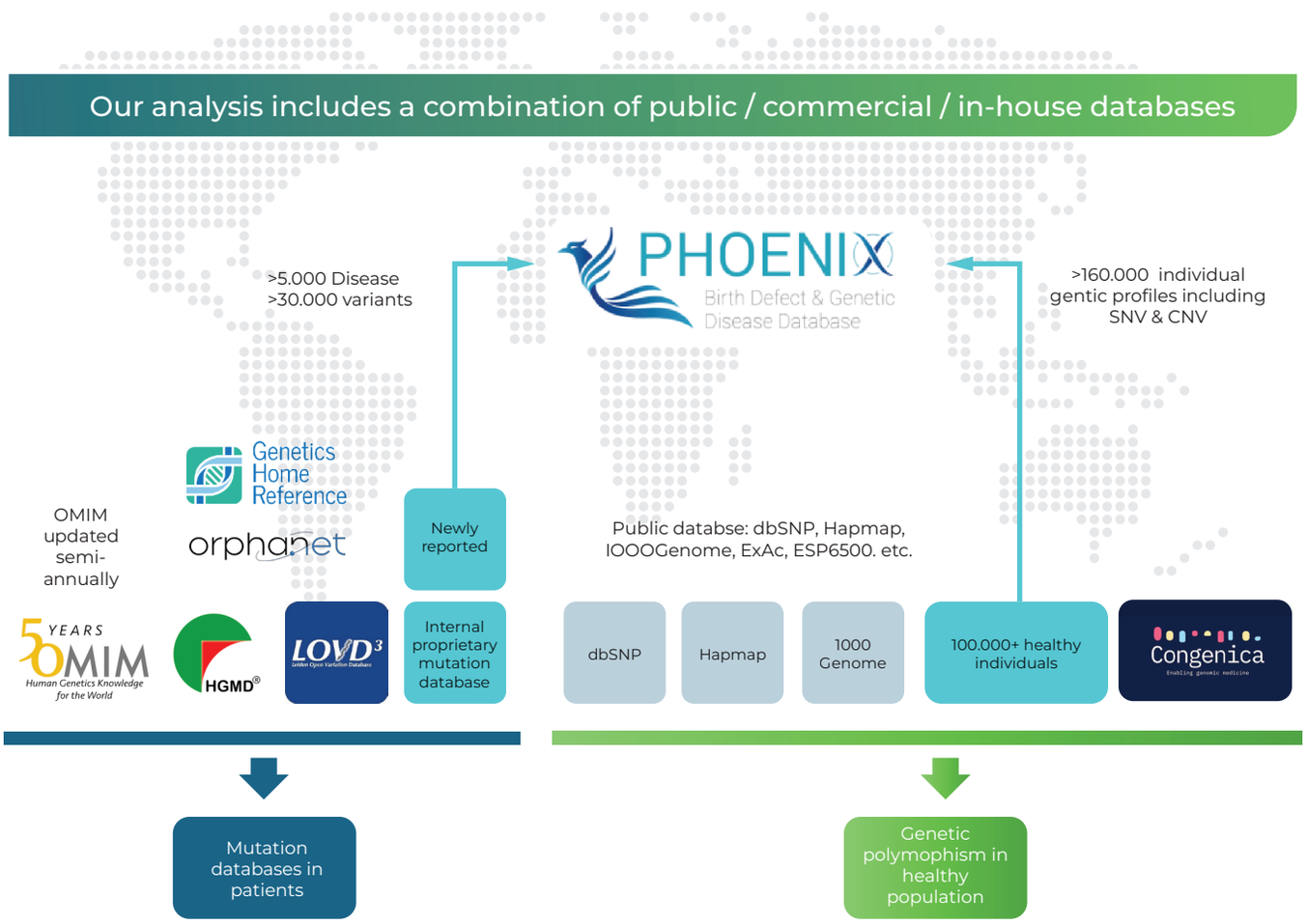
Our experienced team of geneticists, bioinformaticians, and genetic counsellors is available to guide and clarify any aspect of the genetic testing process.

80% of our detected variants are not reported in the literature

We provide a thorough clinical interpretation with expert manual review to ensure no potentially significant variant is overlooked

Patient variants are cross-linked to variant-disease combinations with known phenotype description and molecular basis reported in multiple databases

- Manual review of every variant one-by-one by experts
- All variants are evaluated according to ACMG guidelines
- The mutation database is updated routinely on a semi-annual basis



Who May Benefit?

Patient Profile

OMI RareDx is recommended for cases where a genetic disease is suspected to contribute to some or all of the patient's symptoms.

- Broad, complex, or non-specific symptoms that don't indicate a clear disease or phenotype
- Suspected chromosomal imbalances, microdeletion or microduplication syndromes
- Clinical suspicion of mitochondrial disease
- Prior genetic testing did not provide a conclusive diagnosis, e.g., a negative microarray finding
- Difficult diseases requiring differential diagnosis

Common disorders

- Autism Spectrum Disorder
- Intellectual disability
- Congenital abnormalities
- Developmental delay
- Metabolic disorder
- Epilepsy
- Vision defects
- Cardiomyopathy
- Muscular weakness

Profile Guidance

We are available to provide guidance on the appropriateness of genetic testing for your patients.

Our Whole Genome Sequencing Services are Available for Patients **Worldwide**

Our team can support sample pick up from anywhere in the world.

OMI RareDx has serviced patients from over 30 countries.

Whole Genome Sequencing Brings Clarity

Timely and accurate diagnosis can change everything. Whole Genome Sequencing (WGS) is transforming the diagnostic process—shortening time to diagnosis, revealing hidden causes, and opening doors to individualized care strategies. Empower your clinical decisions with a single, comprehensive test designed to deliver insights that matter.

for Patients and Their Care Teams

Why Use WGS Early in the Diagnostic Journey?

Accelerated Diagnosis

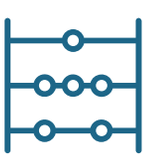
Avoid years of uncertainty. WGS often delivers answers within 2–3 months, enabling earlier access to interventions and support.

Superior Diagnostic Yield

WGS identifies a causative variant in up to 60% of patients with unexplained neurodevelopmental conditions—far surpassing standard panels.

Clinically Actionable Insights

A confirmed diagnosis informs tailored therapeutic strategies, surveillance, and long-term care plans.



Comprehensive and Non-Repetitive

Capture the full genomic landscape in a single test—WGS detects SNVs, CNVs, structural variants, and non-coding mutations, minimizing the need for sequential testing.

Guided Communication and Planning

Facilitates transparent dialogue with families and care teams, equipping them with a clear roadmap for next steps.

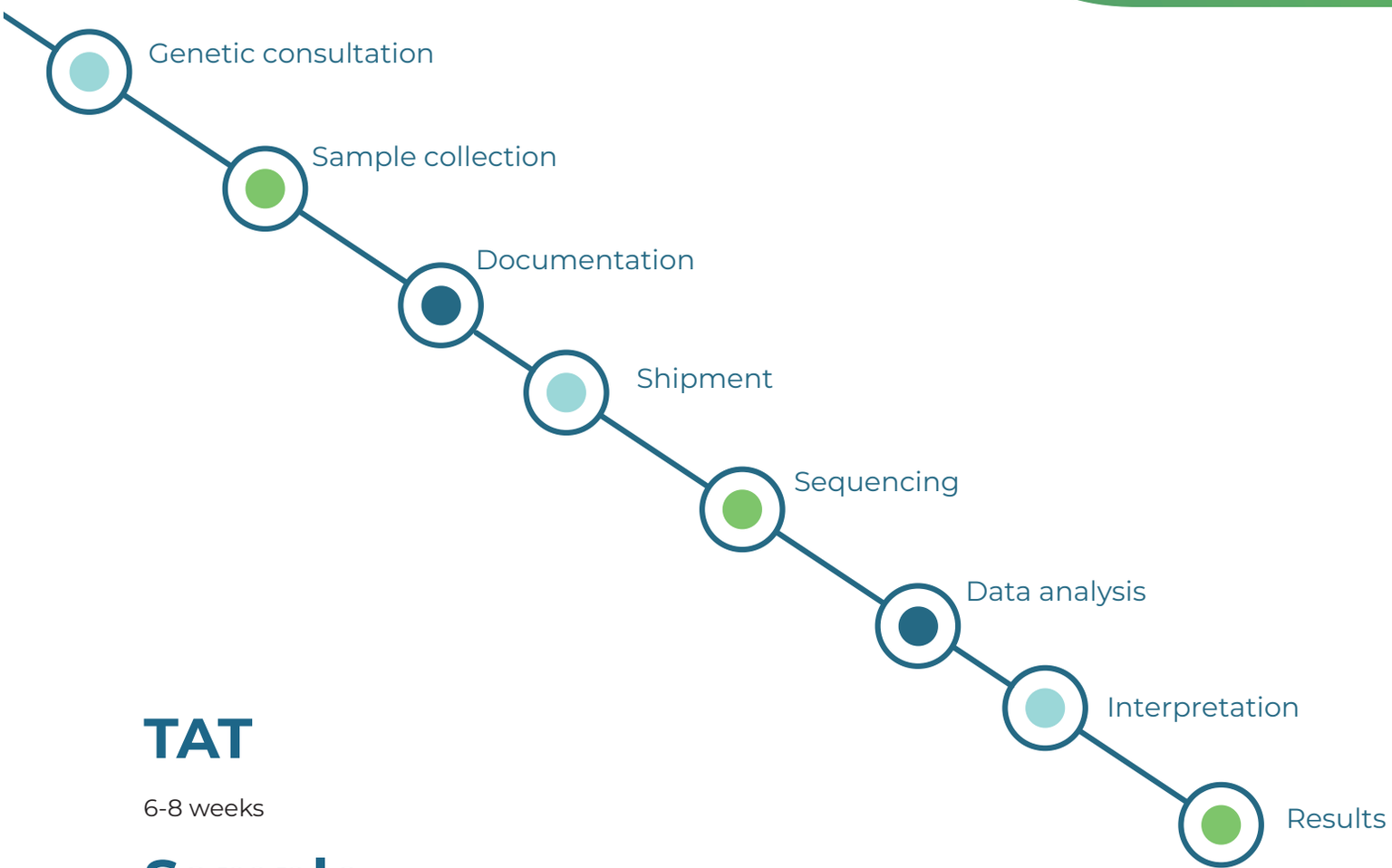
Case Studies: Whole Genome Sequencing in Pediatric Autism Spectrum Disorder (ASD)/ Neurodevelopmental Disorders (NDDs)

Patient	WGS Genetic Finding	Precision-Driven Clinical Impact
5-year-old boy – Global developmental delay, seizures, and ASD features; prior microarray & Fragile X testing normal.	SLC6A8 c.619C>T (p.Arg207Ter); Hemizygous mutation → Creatine Transporter Deficiency (OMIM #300036) [1]	**Targeted Therapy**: Initiated high-dose creatine, arginine, and glycine supplementation to address brain creatine deficiency and improve developmental outcomes.
8-year-old girl – ASD diagnosis with extreme macrocephaly (>99th percentile head circumference); prior gene panel inconclusive.	PTEN c.697C>T (p.Arg233Ter); Heterozygous mutation → PTEN Hamartoma Tumor Syndrome (OMIM #601728) [2]	**Surveillance & Management**: Initiated annual thyroid ultrasound and cancer screening protocol for early detection of benign and malignant growths associated with PTEN mutations.
4-year-old boy – Severe hyperactivity and speech regression; initial diagnosis of autism.	SGSH c.734G>A (p.Arg245His) and c.892C>T (p.Pro298Ser); Compound heterozygous → Sanfilippo Syndrome Type A (OMIM #252900) [3]	**Prognosis & Specialized Care**: Reclassified as a progressive lysosomal storage disorder; care shifted to symptom management, neurodevelopmental monitoring, and eligibility for clinical trials.

References

1. Salomons, G. S., et al. (2001). X-linked creatine-transporter gene (SLC6A8) defect: a new creatine-deficiency syndrome. *Am J Hum Genet*, 68(6), 1497–1500. https://doi.org/10.1086/320595
2. Marsh, D. J., et al. (1999). PTEN mutation spectrum and genotype-phenotype correlations in BRRS suggest a single entity with Cowden syndrome. *Hum Mol Genet*, 8(8), 1461–1472. https://doi.org/10.1093/hmg/8.8.1461
3. Weber, B., et al. (2001). Mutation analysis in mucopolysaccharidosis type IIIA: 12 novel mutations identified in the SGSH gene. *Hum Mutat*, 18(3), 264–265. https://doi.org/10.1002/humu.1204

Workflow



TAT

6-8 weeks

Sample

Peripheral blood (>2S mL) or Genomic DNA (>3pg (30ng/pL))

Request a demo report

Healthcare professionals interested in our service can request a demo report to evaluate the capabilities and insights provided by our genomic testing.

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Technical limitations: This method may not detect all pathogenic mutations. Please contact us for further information.