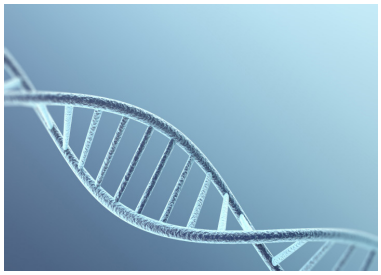

Refining Focus

How Focused NGS is Improving Next-Generation Sequencing Diagnostics

NGS Next Generation Sequencing

- Multi-gene panels
- Whole Exome Sequencing
- Focused Next Generation Sequencing

What is a multi-gene panel?

Multi-gene panels include the sequencing of select genes that share a common phenotype. Greenwood Diagnostic Labs have over 50 multi-gene panels to choose from.

What is Whole Exome Sequencing?

Whole exome sequencing is a test that captures the entire exome with additional coverage for genes with known Mendelian disease associations. In other words, it analyzes and interprets over 20,000 genes!

The standard WES test includes trio analysis with parents.

The average read depth for each exome is **>150x**

What is Focused NGS?

Focused NGS is a customizable Next Generation Sequencing testing option. At Greenwood Diagnostic Labs, Focused NGS allows for cutomized testing tailored to patient's phenotype for genes that may not be offered individually or as an existing panel. Focused NGS allows to clinicians to select up to 20 genes for analysis.

>30% of patient's have a pathogenic or likely pathogenic finding consistent with the reported phenotype.

Focused NGS Diagnostic Yields

(Abnormal results: pathogenic + likely pathogenic)

"This option makes it possible to customize the gene sequencing process by allowing the clinical team to pick and choose one or more genes of greatest interest based on the patient's phenotype. Since the sequence data are generated on an exome platform, it also provides the flexibility to request analysis of the remaining exome data."

David Everman, MD

Focused NGS provides among the highest diagnostic yields of any sequencing

Benefits of Focused NGS

- Clinicians have the freedom to select only the genes they're interested in testing
- Reflex to WES is available for negative cases at a reduced cost
- Analysis and reporting are streamlined
- Option to reflex to the QUICK Analysis

Greenwood Diagnostic Labs
Giving Greater Care

Greenwood Diagnostic Labs

Next Generation Sequencing Panels	# of Genes
Aortic Dysfunction or Dilation and Related Disorders Panel	20
Bardet-Biedl Syndrome Panel	26
Brugada Syndrome Panel	18
Central Hypoventilation Syndrome Panel	3
Charcot-Marie-Tooth Hereditary Neuropathy Panel	54
Cholestasis Panel	73
Comprehensive Cardiac Panel	108
Comprehensive Pulmonary Panel	124
Cone-Rod Dystrophy Panel	37
Congenital Contractures Panel	57
Congenital Stationary Night Blindness Panel	15
Connective Tissue Disorders Panel	35
Cornelia de Lange Syndrome Panel	5
Craniosynostosis Panel	8
Dilated & Arrhythmogenic Cardiomyopathy Panel	51
Dyskeratosis Congenita Panel	14
Early Infantile Epileptic Encephalopathy Panel	86
Epilepsy/Seizure Panel	165
Familial Hypercholesterolemia Panel	4
Hearing Loss Panel	91
Hereditary Spastic Paraplegia Panel	79
Hermansky-Pudlak Syndrome & Pulmonary Fibrosis Panel	40
Hydrops, Non-immune Panel	87
Hypertrophic Cardiomyopathy Panel	24
Kallmann Syndrome & Hypogonadotropic Hypogonadism Panel	39

Next Generation Sequencing Panels	# of Genes
Leber Congenital Amaurosis Panel	24
Long QT Syndrome Panel	18
Lysosomal Storage Disorders Panel	75
Macular Degeneration Panel	24
Maturity-onset Diabetes of the Young Panel (MODY), or Hyperinsulinism Panel	14
Mitochondrial Depletion Panel	23
Neuromuscular Disorders Panel	144
Neuronal Ceroid Lipofuscinoses Panel	9
Ocular Albinism & Hermansky-Pudlak Syndrome	18
Optic Atrophy and Early Glaucoma Panel	34
Overgrowth/Macrocephaly Panel	16
Periodic Fever Panel	14
Peroxisomal Biogenesis Disorders Panel	12
Primary Ciliary Dyskinesia & Cystic Fibrosis Panel	42
Pulmonary Arterial Hypertension Panel	22
RASopathy Panel	23
Retinitis Pigmentosa Panel	92
Rett/Angelman Syndrome Panel	21
Rhabdomyolysis & Metabolic Myopathies Panel	47
Skeletal Dysplasia Panel	11
Syndromic Autism Panel83	83
Tuberous Sclerosis Panel	2
Vascular Malformations Panel	21
X-Linked Intellectual Disorders (XLID) Panel	114

Focused NGS

Custom Panel (2-20 Genes)
Custom Individual Analysis (1 Gene)

QUICKAnalysis

A free NGS-reflex analysis that rapidly screens the full exome for pathogenic variants when panels results are negative.

Whole Exome Sequencing

Prenatal Exome Sequencing
Singleton Analysis
Trio Analysis

106 Gregor Mendel Circle
Greenwood, SC 29646

+ 1.800.473.9411

ggc.org/diagnostic-lab