Rare Disease Diagnosis Decision Support Tool:

Broad Clinical Phenotype

TO FIND A UNIFYING DIAGNOSIS FOR PATIENTS WITH MULTIPLE MEDICAL ISSUES

- Multiple Congenital Anomalies
- Failure To Thrive
- Dysmorphic Features
- Autism Spectrum Disorders
- Neurodevelopmental Disorders
- Intellectual Disability
- Seizure Disorders
- Extensive Differential Diagnosis
- Atypical Presentations
- Dual Diagnoses

CMA
Assesses copy number variants and associated conditions

Chromosomal Microarray Analysis (CMA)

- 15+ years of experience and >80,000 studies
- Extensive database and knowledge
- Exon-by-exon coverage of over 4200 genes
- 57,000 oligos to detect absence of heterozygosity, uniparental disomy, or consanguinity

WES
Assesses coding region of the genome for sequence variants

Whole Exome Sequencing (WES)

- 9+ years of experience and >20,000 studies
- Extensive database and knowledge
- Diagnostic rates range from 25-40%, greater for Trio WES cases
- Rapid options (TAT of 10 days)

WGS
Assesses entire genome for copy number and sequence variants

Whole Genome Sequencing (WGS)

- One of the first labs to offer clinically (February 2019)
- Extensive experience as sequencing lab for the Undiagnosed Diseases Network
- Diagnostic rates of 40% or greater is expected for new patients

Disclaimer: The above provides general recommendations for appropriate testing to consider based on presenting phenotype. Optimal test selection and strategy is best determined by the clinical health care provider. Metabolic, mitochondrial, and/or repeat expansion disorders may need an additional separate assessment.

BAYLOR GENETICS