ACMG 2014
LEARN WHAT IS NEW IN LABORATORY GENETICS FROM MAYO MEDICAL LABORATORIES

CHROMOSOMAL MICROARRAY: Prenatal testing
FRIEDREICH ATAXIA: Frataxin concentration to diagnose and monitor patients
OLIGOSACCHARIDOSES AND MUCOPOLYSACCHARIDES: Improved screening approaches
LYSOSOMAL STORAGE DISORDERS: Comprehensive test menu
HEREDITARY COLON CANCER SYNDROMES: Targeted testing and next generation sequencing
CLIR POST-ANALYTICAL TOOLS: Lowering false positives and improving differential diagnosis

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### NEW TESTS AVAILABLE FROM MAYO MEDICAL LABORATORIES

**CYTOGENETICS**
- CMAPD  Prenatal Chromosomal Microarray

**BIOCHEMICAL GENETICS**
- PLSD  Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot
- TALDO  Polyols, Quantitative, Urine
- GATOL  Galactitol, Quantitative, Urine
- OLIWB  Oligosaccharidoses Screen, Leukocytes
- OLITC  Oligosaccharidoses Screen, Fibroblasts
- BAFS  Bile Acids, Fractionated and Total, Serum
- I2SBS  Iduronate-2-sulfatase, Blood Spot
- I2SW  Iduronate-2-sulfatase, Whole Blood

**MOLECULAR GENETICS**
- HURLS  Hurler Syndrome, Full Gene Analysis
- HUNTS  Hunter Syndrome, Full Gene Analysis
- PPN  Acute Porphyria, Multi-Gene Panel
- MP3AS  Mucopolysaccharidosis IIIA, Full Gene Analysis
- MP3BS  Mucopolysaccharidosis IIIB, Full Gene Analysis
- MPS6S  Mucopolysaccharidosis VI, Full Gene Analysis
- XALDS  X-Linked Adrenoleukodystrophy, Full Gene Analysis
- SUMFS  Multiple Sulfatase Deficiency, Full Gene Analysis
- ML23S  GNPTAB Gene, Full Gene Analysis
- FLGA  FLG Gene, Mutation Analysis
- HOXB1  HOXB13 Mutation Analysis (G84E)
- C9OR  C9orf72, Molecular Analysis

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### SEE FULL TEST DETAILS AT

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