

Clinical Genomics Laboratory Featured Tests

CYTOGENETIC TESTING

Prenatal Testing

- Prenatal Aneuploidy FISH Screen for Chromosomes 13, 18, 21, X, Y
- Amniotic Fluid Chromosome Analysis, Standard
- Amniotic Fluid 5-cell Chromosome Analysis
- Prenatal Microarray with Parental Testing
- Prenatal Microarray without Parental Testing
- Percutaneous Umbilical Blood, Chromosome Analysis
- Amniotic Fluid Culture Only

Products of Conception (POC) Testing

- POC Microarray Analysis
- POC Microarray with 5-cell Chromosome Analysis
- Tissue Chromosome Analysis
- Tissue Culture Only

Postnatal Testing

- Chromosome Analysis (STAT reporting available): Peripheral Blood or Cord Blood
- Fibroblast Culture: Biochemical/Genetic Studies, Cryopreservation of Cultured Cells
- FISH Analysis
- SNP Microarray Analysis

MOLECULAR GENETIC TESTING

- Bone Marrow Transplant (BMT) Engraftment Analysis, Pre-Transplant/Post-Transplant
- Cystic Fibrosis Common Mutation Analysis (60 Variants)
- Familial Variant Targeted Sequencing: Targeted Variant Testing available for known variant(s)
- Fragile X Syndrome Repeat Number Analysis with Reflex to Methylation
- Krabbe Disease: *GALC* Gene Common 30-kb Deletion Detection, *GALC* Gene Sequencing
- Mucopolysaccharidosis Type I (MPS I): *IDUA* Gene Sequencing
- Prader-Willi Syndrome/Angelman Syndrome Methylation Analysis
- Reanalysis Genome Sequencing: Reanalysis of Genome or Rapid Genome Sequencing data of patients tested at Nationwide Children's
- Spinal Muscular Atrophy (SMA) Dosage Analysis



When your child needs a hospital, everything matters.

ONCOLOGY GENETIC TESTING

CNS Malignancies

- Somatic Disease/Germline Comparator Exome
- Solid Tumor Fusion Analysis (151 Fusion Partner Genes)
- CNS Tumor Classification by Methylation Array
- Medulloblastoma: *MYCN (N-MYC)* and *MYC (C-MYC)* Amplification Detection by FISH

Soft Tissue Sarcoma

- Somatic Disease/Germline Comparator Exome
- *EWSR1* FISH Tumor Analysis
- Alveolar Rhabdomyosarcoma: *FOXO1 (FKHR)* Rearrangement Detection by FISH

Neuroblastoma

- Somatic Disease/Germline Comparator Exome
- Targeted Oncology Microarray (DNA Gain/Loss and LOH Detection)
- NGS Neuroblastoma Panel
- *MYCN (N-MYC)* Amplification Detection by FISH
- *ALK* Amplification Detection by FISH

Wilms Tumor

- Somatic Disease/Germline Comparator Exome
- Targeted Oncology Microarray (DNA Gain/Loss and LOH Detection)

Other Solid Tumors and Vascular Anomalies

- Somatic Disease/Germline Comparator Exome
- Solid Tumor Fusion Analysis (151 Fusion Partner Genes)

Hematologic Cancers

- Somatic Disease/Germline Comparator Exome
- Hematologic Cancer Fusion Analysis (Fusion Detection for 148 Gene Partners)

Custom Confirmation Tests:

- Targeted Tumor Variant Analysis – Confirmatory test for a known somatic variant
- Targeted Tumor (RT-PCR) Fusion Analysis – Confirmatory test for a known gene fusion

To learn more about the tests offered, sample(s) required, and to order testing, please visit [NationwideChildrens.org/Specialties/Institute-for-Genomic-Medicine](https://www.NationwideChildrens.org/Specialties/Institute-for-Genomic-Medicine) or call (614) 722-5321.



When your child needs a hospital, everything matters.