

## The Human Genetics Laboratory at a glance

**Our Mission...** is to be leaders in providing exceptional genetic testing and interpretation for patients, families, and communities we serve through extraordinary patient care, premier educational programs, and innovative research.

### History:

- **1974** – Laboratory established  
Conventional cytogenetics launched
- **1994** – Fluorescence *in situ* hybridization (FISH)
- **1999** – Methylation
- **2001** – Array technology introduced
- **2006** – Sanger sequencing
- **2008** – Multiplex ligation-dependent probe amplification (MLPA)
- **2012** – Next generation sequencing (NGS)
- **2018** – Exome sequencing

### Organization:

- Accredited by the College of American Pathologists (CAP) and certified by the Clinical Laboratory Improvement Amendments (CLIA); state licensed in California, Florida, and Pennsylvania
- Reference laboratory for national protocols: Cancer and Leukemia Group B (CALGB) and Children's Oncology Group (COG)
- Approximately 60 team members, including directors board-certified by the American Board of Medical Genetics and Genomics (ABMGG), technologists certified in cytogenetic and molecular biology and a licensed and certified genetic counselor
- Four technical service sections: Cytogenetics, FISH, Microarray, Molecular
- A fully developed client services team to triage communications and specimen intake, manage insurance pre-authorizations, and oversee billing
- A dedicated marketing specialist who provides client outreach including web design, newsletters, and educational materials for patients and providers
- A highly integrated operation with the clinical genetics division

### Clinical services:

- Genetic testing for regional, national, and international health care providers
- Over 130 genetic test offerings in cytogenetics and molecular genetics
- Diagnostic tests for all areas of genetic health: Prenatal and Pregnancy Loss, Postnatal, Hereditary Cancer, Hematology/Oncology
- Complimentary specimen collection kits
- Test ordering and result reporting interface capabilities

### Educational and research activities:

- Fellowship program through the American Board of Medical Genetics and Genomics (ABMGG)
- Courses taught: MMI911, MMI912, MMI921, MMI922, PEDS727, CAHP412
- Critical rotation site in UNMC and CUMC pathology resident and fellowship programs
- International collaborations, including a long-standing partnership with colleagues in Singapore
- Hundreds of national/international presentations and peer-reviewed publications
- Field-specific tradeshow presence and community outreach