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
- 1974 – 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