Genetic Services in St. Louis

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St. Louis, Missouri

• Founded: 1764
• City population: 318,069 (2011)
• Racial/Ethnic
  – 49.2% African American
  – 43.9% White
  – 2.9% Asian
  – 3.5% Hispanic or Latino of any race
• Metropolitan area population: 2,817,355 (19th largest US city)
Genetic Services in St. Louis

• Washington University
  – St. Louis Children’s Hospital
  – Barnes-Jewish Hospital
  – Shriner’s Hospital for Children

• St. Louis University
  – Cardinal Glennon Children’s Medical Center
  – St. Mary’s Health Center

• Mercy Hospital

• St. Luke’s Hospital
St. Louis Children’s Hospital

- Clinical Geneticists (4 FTE)
  - Dr. Kathy Grange
  - Dr. Marcia Willing
  - Dr. Marwan Shinawi
  - Dr. Beth Kozel
- Genetic Counselors (2.2 FTE)
  - Linda Manwaring
  - Niki Armstrong
  - Marisa Vineyard
- Metabolic Dietitians (2 FTE)
  - Laurie Sprietsma
  - Liz Toolan
- Nurse Practitioner
  - Melissa Sicard

St. Louis Children’s Hospital

- General Genetics Clinic
- PKU and Metabolic Clinics
- Down Syndrome Center
- Skeletal Dysplasia Clinic
- Marfan Syndrome Clinic
- Connective Tissue Disorders Clinic
- Craniofacial Clinic
- Neurofibromatosis Clinic
Center for Advanced Medicine
Barnes-Jewish Hospital

- Maternal Fetal Medicine and Prenatal Ultrasound Services
- 12 faculty in division
- OB Geneticists
  - Dr. Diana Gray
  - Dr. Ryan Longman
  - Dr. Lisa Bernhard
- Prenatal genetic counselors
  - Erin Berry, MS
  - Melissa Fuller, MS

Siteman Cancer Center

- Adult Cancer Genetics Clinic
  - Dr. Alison Whelan
  - Jennifer Ivanovich, MS
- Gynecologic Oncology---Ovarian Cancer
  - Sherry Babb, MS
Shriner’s Hospital

• Genetics Clinic
  – Kathy Grange, MD
    • Skeletal dysplasias
    • Arthrogryposis

• Research programs
  – Michael Whyte, MD
  – Gary Gottesman, MD
    • Osteogenesis Imperfecta
    • Hypophosphatasia
    • Hypophosphatemic Rickets
    • Other rare metabolic bone disorders
Biochemical Genetics Laboratory

• Serum and urine amino acid analysis
• Urine organic acid analysis
• Acylcarnitine profile
• Total and free carnitine levels

Cytogenomics and Molecular Pathology Laboratory

• 3 Cytogeneticists
• Over 7000 cases in 2011
• Classical Cytogenetics Studies
  – Constitutional
  – Prenatal Chromosomes
  – Products of Conception
  – Cancer Chromosome Studies
• Molecular Cytogenetics
  – Fluorescent In Situ Hybridization
  – Chromosomal Microarray Analysis
Molecular Diagnostics Laboratory

- Fragile X syndrome
- Prader-Willi/Angelman syndromes
- Beckwith-Wiedemann/Russell-Silver syndromes
- Multiple endocrine neoplasia (RET)
- Cystic fibrosis
- MCAD (985A>G)
- Factor V Leiden
- Prothrombin
- Cancer diagnostic and pharmacogenetic tests

Genomics and Pathology Service (GPS)

- Collaboration between the Departments of Pathology & Immunology and Genetics at Washington University
- GPS offers genomic tests to support patient care and clinical trials
- Testing by the GPS is performed in a CLIA certified, CAP accredited laboratory focused on DNA and RNA testing
Clinical Next-Generation Sequencing

<table>
<thead>
<tr>
<th>Gene Set Test Name</th>
<th>Included Genes</th>
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<tbody>
<tr>
<td><strong>Comprehensive Cancer Set</strong></td>
<td><em>All Genes</em></td>
</tr>
<tr>
<td>Brain Cancer Gene Set</td>
<td>BRAF, IDH1, IDH2, PTEN</td>
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<tr>
<td>Colon Cancer Gene Set</td>
<td>BRAF, CTNNB1, EGFR, KRAS, NRAS, PIK3CA, TP53</td>
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<tr>
<td>Endometrium</td>
<td>KRAS, PTEN</td>
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<tr>
<td>GI Stromal Tumor Gene Set</td>
<td>CSF1R, KIT (c-KIT), PDGFR-A</td>
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<tr>
<td>Lung Cancer Gene Set</td>
<td>ALK*, BRAF, CTNNB1, EGFR, KRAS, MAP2K2 (MEK), MAPK1 (ERK), MET, NRAS, PIK3CA, TP53</td>
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<tr>
<td>Melanoma Gene Set</td>
<td>BRAF, KRAS, MAP2K2 (MEK), MAPK1 (ERK), MET, NRAS, PIK3CA, PTEN</td>
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<tr>
<td>MEN2A/2B/FMTC</td>
<td>RET</td>
</tr>
<tr>
<td>Myeloid Disorder Gene Set</td>
<td>CHIC2, CSF1R, DNMT3A, FLT3, IDH1, IDH2, JAK2, KIT (c-KIT), MLL*, NPM1, PTPN11, RUNX1, WT1</td>
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<tr>
<td>Ovarian Cancer Gene Set</td>
<td>PTEN, TP53</td>
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<tr>
<td>Pancreatic Cancer Gene Set</td>
<td>KRAS, NRAS, PIK3CA, TP53</td>
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<tr>
<td>Prostate Cancer Gene Set</td>
<td>CTNNB1, PTEN</td>
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<tr>
<td>Sarcoma Gene Set</td>
<td>CTNNB1, KIT (c-KIT), PDGFR-A, PTEN, NRAS, BRAF, KRAS, MAPK1, TP53, RET</td>
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<tr>
<td>Testis Cancer Gene Set</td>
<td>PTEN, KIT (c-KIT)</td>
</tr>
<tr>
<td>Thyroid Cancer Gene Set</td>
<td>BRAF, CTNNB1, KRAS, NRAS, RET</td>
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<tr>
<td>TP53 Gene Sequencing</td>
<td>TP53</td>
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<tr>
<td>Wilms Tumor</td>
<td>WT1</td>
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</tbody>
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Genome Institute

• The Genome Institute is a leader in the constantly changing field of genomics
• One of three NIH funded large-scale sequencing centers in the United States
• High-speed, comprehensive genomics
• Since inception in 1993 the Genome Institute has received over $800 million in funding

Genetics Training Programs

• Categorical Medical Genetics Residency (Fellowship)
  – 2 year program
  – 3rd year of research
• Combined Pediatrics/Medical Genetics Residency
  – 5 year program
St. Louis University School of Medicine

Cardinal Glennon
SSM Cardinal Glennon Children's Medical Center
Cardinal Glennon Children’s Medical Center

- **Pediatric Geneticists (1.2 FTE)**
  - Dr. Stephen Braddock
  - Dr. Sandra Davenport
- **Genetic Counselors (2.8 FTE)**
  - Katherine Christensen, MS
  - Erin Torti, MS
  - Laura Waldman, MS
- **Metabolic Dietitian**
  - Beth Piper, RD
- **Nurse**
  - Deb Boylan, RN

Cardinal Glennon Children's Medical Center

- **General Genetics Clinic**
- **Metabolic Clinic**
- **Neurofibromatosis Clinic**
- **Marfan Syndrome Clinic**
- **CHARGE Syndrome Clinic**
- **Craniofacial Clinic**
- **Inpatient consultations at CGCMC and St. Mary's**
St. Louis University
Genetics Outreach Clinics

- Illinois Department of Public Health
- Grant-funded rural outreach clinics
- 28 clinics held per year
  - Fayette County
  - Jackson County
  - Jefferson County
  - Madison County

St. Louis University
Prenatal Services

- OB/GYN Geneticist
  - Kim Martin, MD
- Genetic Counselors (2.5 FTE)
  - Rachel Bradshaw
  - Meg Hefner
  - Darbey Raible
- Prenatal Clinics
  - St. Mary’s Health enter
  - SSM Cardinal Glennon Fetal Care Institute
  - SSM Outreach Maternal Fetal Medicine Clinics
St. Louis University
Other Services

- Cytogenetics Laboratory
  - Dr. Jacqueline Batanian
- Metabolic Laboratory
  - Dr. James Shoemaker
  - Organic and amino acid analysis
- Pregnancy Risk Information Services
  - Teratology
- Cancer Genetics and Risk Assessment
  Clinic for adults

Newborn Screening Follow-up

- Unless dictated by insurance, Missouri abnormal screen cases are referred to...
- St. Louis Children’s Hospital
  - First half of the alphabet
- Cardinal Glennon Children’s Med Center
  - Second half of the alphabet
- Illinois cases are referred to both hospitals as well
Mercy Hospital

- Formerly St. John’s Mercy Medical Center
- ~6000 deliveries per year
- Prenatal Genetics Services
  - Genetic Counselors (2.5 FTE)
    - Rachel Slaugh, MS
    - Kay Lechien, MS
    - Susan Jones, MS
  - Prenatal ultrasound
  - High risk pregnancy management
- NICU and other pediatric services

St. Luke’s Hospital

- Genetic Counselor
  - Heidi Beaver, MS
  - Prenatal genetic counseling
  - Cancer genetics counseling