

Genetic Services in St. Louis



Dr. Kathy Grange
Genetics and Genomic Medicine
Department of Pediatrics
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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
 - Shriners' Hospital for Children
- St. Louis University
 - Cardinal Glennon Children's Medical Center
 - St. Mary's Health Center
- Mercy Hospital
- St. Luke's Hospital

Children's

HOSPITAL • ST. LOUIS

BJC HealthCare™

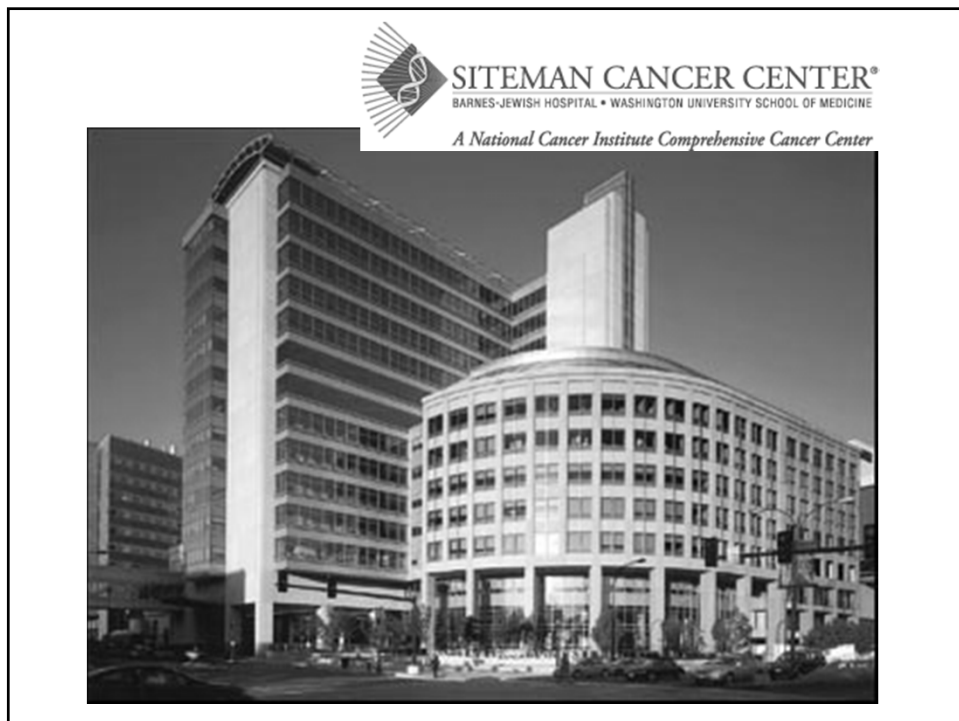


St. Louis Children's Hospital

- 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



Shriners Hospitals
for Children™



Shriner's Hospital

- **Genetics Clinic**
 - Kathy Grange, MD
 - Skeletal dysplasias
 - Arthrogyrosis
- **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

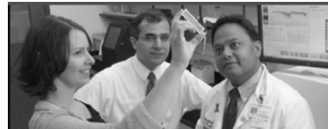
Cytogenomics and Molecular Pathology Laboratory

- 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

Gene Set Test Name <small>click on a name</small>	Included Genes
Comprehensive Cancer Set	All Genes
+ Brain Cancer Gene Set	BRAF, IDH1, IDH2, PTEN
+ Colon Cancer Gene Set	BRAF, CTNNB1, EGFR, KRAS, NRAS, PIK3CA, TP53
+ Endometrium	KRAS, PTEN
+ GI Stromal Tumor Gene Set	CSF1R, KIT (c-kit), PDGFRA
+ Lung Cancer Gene Set	ALK *, BRAF, CTNNB1, EGFR, KRAS, MAP2K2 (MEK), MAPK1 (ERK), MET, NRAS, PIK3CA, TP53
+ Melanoma Gene Set	BRAF, KRAS, MAP2K2 (MEK), MAPK1 (ERK), MET, NRAS, PIK3CA, PTEN
+ MEN2A/2B/FMTC	RET
+ Myeloid Disorder Gene Set	CHIC2, CSF1R, DNMT3A, FLT3, IDH1, IDH2, JAK2, KIT (c-KIT), MLL *, NPM1, PTPN11, RUNX1, WT1
+ Ovarian Cancer Gene Set	PTEN, TP53
+ Pancreatic Cancer Gene Set	KRAS, NRAS, PIK3CA, TP53
+ Prostate Cancer Gene Set	CTNNB1, PTEN
+ Sarcoma Gene Set	CTNNB1, KIT (c-kit), PDGFRA, PTEN, NRAS, BRAF, KRAS, MAPK1, TP53, RET
+ Testis Cancer Gene Set	PTEN, KIT (c-KIT)
+ Thyroid Cancer Gene Set	BRAF, CTNNB1, KRAS, NRAS, RET
+ TP53 Gene Sequencing	TP53
+ Wilms Tumor	WT1



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