22q and You Center

The 22q and You Center at Children's Hospital of Philadelphia is an internationally recognized leader in the diagnosis and treatment of children with chromosome 22q11.2 deletion.

Genetic Counselors
Donna McDonald-McGinn, MS, LCGC
Bekah Wang, MS, LCGC

Beckwith Wiedemann Syndrome Clinic

The team at the Beckwith-Wiedemann Syndrome Clinic at Children's Hospital of Philadelphia (CHOP) provides support and medical guidance for children with these genetic and epigenetic disorders: Beckwith-Wiedemann syndrome, 11p Overgrowth Spectrum, hemihypertrophy/lateralized overgrowth and related disorders.

Genetic Counselor
Evan Hathaway, MS, LCGC

Biochemical Genetics

The Metabolic Disease Program at Children's Hospital of Philadelphia provides state-of-the-art diagnostic and treatment services for genetic metabolic disorders.

Genetic Counselors
Nicole Engelhardt, MS, LCGC
Kierstin Keller, MS, LCGC
Caitlin Menello, MS, LCGC
Lee Williams, MS, LCGC

Cardiology Genetics

The Familial Cardiomyopathy Program is a special program for patients with cardiomyopathies. It unites the breadth of resources from two premier institutions, Children’s Hospital of Philadelphia (CHOP) and Penn Medicine. Our team members are among the best in the country in the treatment of cardiomyopathies.

Genetic Counselor
Alyssa Rippert, MS, LCGC
**Center for Applied Genomics**

The Center aims to discover the genetic causes of prevalent childhood diseases and translate findings into medical innovations. The Center is engaged in a number of major research projects and aims to collect and analyze data from more than 100,000 children.

**Genetic Counselors**
- Margaret Horton Harr, MS, LCGC
- Priyanka Maripuri, MS
- Jasmine Purcell, MS
- Shannon Terek, MS, LCGC

**Center for Fetal Diagnosis & Treatment**

The Center provides an unmatched level of care for babies with prenatally diagnosed birth defects — since 1995, they’ve cared for more than 25,289 expectant parents from around the world and performed more than 1,847 fetal surgeries. Every day they’re searching for new ways to treat these life-threatening conditions, offering hope for a better future for all children with birth defects.

**Genetic Counselors**
- Natalie Burrill, MS, LCGC
- Haley Crane, MS, LCGC
- Renee Wright, MS, LCGC
- Lisa Pilchman, MS, LCGC
- Erica Schindewolf, MS, LCGC

**Center for Mitochondrial Medicine**

Mitochondrial Medicine at Children’s Hospital of Philadelphia (CHOP) is emerging as the premiere center in the world for multidisciplinary clinical care, advanced diagnostics and therapies, and individualized basic, translational, and clinical research programs dedicated to improving the health of patients of all ages living with mitochondrial disease. Mitochondrial Medicine is designated a Frontier Program by CHOP. Frontier Programs are unique, cutting-edge programs that will forge important new discoveries, deliver novel therapies, and help even more children and adults thrive.

**Genetic Counselors**
- Emily Bogush, MS, LCGC
- Elizabeth McCormick, MS, LCGC
- Colleen Muraresku, MS, LCGC
- James Peterson, MS, LCGC

**Clinical Genetics Center**

The Clinical Genetics section provides comprehensive diagnostic evaluations, follow-up care and genetic counseling for both inpatients and outpatients. Patients are seen by a clinical geneticist and a genetic counselor. Individuals with developmental delays, intellectual disabilities, birth defects, multisystem medical problems, unusual facial features or failure to thrive are referred for evaluation and diagnosis of a genetic syndrome or a chromosomal abnormality.

**Genetic Counselors**
- Jeshua DeJesse, MS, CGC
Sarah Donoghue, MS, LCGC
Beth Keena, MS, LCGC
Donna McDonald-McGinn, MS, LCGC
Morgan McManus, MS, LCGC

Congenital Hyperinsulinism Center

The Congenital Hyperinsulinism Center brings together a team of congenital hyperinsulinism experts who work together to provide specialized, patient-centered and seamless care for children with congenital HI and their families.

Genetic Counselor
Victoria Sanders, MS, LCGC

Division of Genomic Diagnostics

The Division of Genomic Diagnostics (DGD) provides a wide spectrum of testing for genetic conditions, cancer diagnosis and treatment, and histocompatibility and immunogenetics. Testing ranges from genome wide analyses, such as cytogenetics, chromosomal microarray analysis and exome sequencing to more targeted testing, including HLA typing, fluorescence in situ hybridization (FISH), gene panels and single gene testing.

Genetic Counselors
Colleen Campbell, MS, LCGC
Sarah Charles, MS, LCGC
Jiani Chen, MS, LCGC
Elizabeth DeChene, MS, LCGC
Elizabeth Hopkins Denenberg, MS, LCGC
Elizabeth Fanning, MS, LCGC
Daniel Gallo, MS, LCGC
Tammy Luke, MS
Lauren Lulis, MS, LCGC
Sara L. Reichert, MS, LCGC
Nicholas Staropoli, MS, LCGC
Morgan Thomas, MS, LCGC

Friedreich’s Ataxia Program

Children and young adults with Friedreich's ataxia (FA) find dedicated, expert care at CHOP’s Friedreich's Ataxia Program. The program is part of the Friedreich’s Ataxia Center of Excellence, a first-of-its-kind collaboration of leading FA experts committed to promoting FA research and clinical care. From initial diagnosis and testing through long-term disease management, we provide comprehensive care for children and adults from all over the world with this rare, progressive neurogenetic condition.

Genetic Counselors
McKenzie Wells, MS, LCGC
Hereditary Cancer Predisposition Clinic

CHOP’s program uses the latest advances in technology to provide genetic testing, genetic counseling and cancer surveillance, and thus improve the outlook for children with a genetic predisposition to develop cancer.

Genetic Counselors
Mary Egan Clark, MS, LCGC
Sarah Jennings, MS, LCGC
Kristin Zelley, MS, LCGC

Jill and Mark Fishman Center for Lymphatic Disorders at Children's Hospital of Philadelphia

The Center provides highly specialized care for children and adults with lymphatic leaks and lymphatic flow disorders. Experts in the Center are developing advanced imaging and interventional technologies to map out the anatomy and flow of the lymphatic system. This allows our team to more accurately identify and manage several chronic and debilitating conditions using a variety of minimally invasive treatment options.

Genetic Counselor
Allison Britt, MS, LCGC

Leukodystrophy Center of Excellence

The Leukodystrophy Center of Excellence is a diagnostic, clinical care, and research initiative that addresses the unique needs of infants, children and adolescents with inherited white matter diseases. Formed in response to legislation in Pennsylvania and New Jersey to add several leukodystrophies to the newborn screening panel, the Center of Excellence meets the need for families to obtain state-of-the-art laboratory and imaging diagnostics and integrated multidisciplinary care from top specialists across CHOP.

Genetic Counselor
Kaley Arnold, MS
Carlos Dominguez Gonzalez, MS

Epilepsy Neurogenetics Initiative (ENGIN) Frontier Program

The Epilepsy Neurogenetics Initiative (ENGIN) Frontier Program at Children’s Hospital of Philadelphia (CHOP) integrates genetic testing into the diagnosis and treatment of children with difficult-to-treat or unexplained epilepsies, and provides access to expert care for children with genetic epilepsy syndromes and other genetic neurodevelopmental disorders. We combine cutting-edge clinical care and advanced genetic testing with innovative research to identify the genetic variants causing a child’s epilepsy and develop an individualized approach to treatment and management.

Genetic Counselors
Amanda Back, MS, LCGC
Brianna Berlin, MS
Stacey Cohen, MS, LCGC
Holly Dubbs, MS, LCGC
Neuromuscular Genetics

CHOP’s Neuromuscular Program is staffed by experts in the diagnosis and treatment of children with neuromuscular diseases. These diseases require timely evaluations and expert diagnosis and testing, and the team is committed to providing the most current, comprehensive and specialized care possible. The program has been recognized by Parent Project Muscular Dystrophy, the Spinal Muscular Atrophy Foundation and the Muscular Dystrophy Association.

Genetic Counselor
Brianna Gross, MS, LCGC

Roberts Individualized Medical Genetics Center

The mission of the Roberts IMGC, the first pediatric individualized genomic program in the country, remains the same: to facilitate access to state-of-the-art individualized genetic testing and management for children, families and clinicians, and to promote integration of clinical and genomic information into the diagnostic and research efforts at Children’s Hospital of Philadelphia (CHOP).

Genetic Counselors
Emma Bedoukian, MS, LCGC
Victoria Dortenzio, MS, LCGC
Christopher Gray, MS, LCGC
Emily Krauss, MS, LCGC
Jackie Leonard, MSc, MS, LCGC
Michelle Marchese, MS
Sarah Raible, MS, LCGC
Jacob Squicciarini, MS, LCGC
Lauren Voss, MS, LCGC