

# Pediatric Specialist Guide

*A referral guide for health care providers*



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Children's Hospital Boston

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CHILDRENSHOSPITAL.ORG

## GENETICS

### Genetics

[childrenshospital.org/genetics](http://childrenshospital.org/genetics)

## SCHEDULING

**Children's Hospital Boston**  
300 Longwood Avenue, Fegan 10  
Boston, MA 02115  
617-355-6394  
Fax: 617-730-0466

**Children's Hospital Boston at Waltham**  
9 Hope Avenue  
Waltham, MA 02453  
781-216-1600  
Fax: 617-730-0466

**Boston Children's Physicians South**  
Stetson Medical Center  
541 Main Street  
Weymouth, MA 02190  
781-331-4715  
Fax: 617-730-0466

**Children's Hospital Physicians at Good Samaritan Medical Center**  
235 North Pearl Street  
Brockton, MA 02301  
508-427-3000  
Fax: 617-730-0466

**Children's Hospital Boston at Lexington**  
482 Bedford Street  
Lexington, MA 02420  
781-672-2100  
Fax: 617-730-0466

**Boston Children's North**  
10 Centennial Drive  
Peabody, MA 01960  
978-538-3600  
Fax: 617-730-0466

**Children's Hospital Physicians at Norwood Hospital**  
800 Washington Street  
Norwood, MA 02062  
781-440-9619  
Fax: 617-730-0466

**Request an appointment:** [childrenshospital.org/genetics](http://childrenshospital.org/genetics)

**Urgent appointment triage**

Genetics: 857-218-4637

Metabolism: 617-355-2521

<p><b>AREAS OF EXPERTISE:</b></p> <ul style="list-style-type: none"><li>● Angelman syndrome</li><li>● antenatal/fetal genetics and genetic syndromes</li><li>● brain malformations</li><li>● chromosome abnormalities</li><li>● connective tissue disorders</li><li>● fatty acid oxidation defects</li><li>● fragile X syndrome</li><li>● galactosemia</li><li>● homocystinuria</li><li>● MCAD</li><li>● methylmalonic acidemia</li><li>● MSUD</li><li>● neurofibromatosis</li><li>● organic acidemia</li><li>● phenylketonuria (PKU)</li><li>● Prader-Willi syndrome</li><li>● propionic acidemia</li><li>● sensorineural hearing loss</li><li>● skeletal dysplasia</li><li>● Smith Lemli Optiz syndrome</li><li>● velo-cardio-facial syndrome</li></ul>	<p><b>HIGHLIGHTS:</b></p> <ul style="list-style-type: none"><li>● 4,700 outpatient visits per year</li><li>● 9 specialty clinics and programs</li><li>● 19 geneticists/biochemical geneticists, 5 genetic counselors and 1 nurse practitioner on staff</li><li>● 24-hour a day care and consultation available</li></ul> <p>For more information, call 1-800-355-7944 or visit <a href="http://www.childrenshospital.org">www.childrenshospital.org</a></p>
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## GENETICS

### CLINICAL PROGRAMS:

#### **Brain Development and Genetics Clinic**

Provides multidisciplinary consult care primarily for children with structural brain malformations, also known as neuronal migration disorders, usually seen on brain MRI. We also see children diagnosed with other conditions affecting brain development. Our team of professionals, with specialists in pediatric neurology, epileptology and genetics counseling, reviews imaging, diagnoses, test results and potential treatment options and discusses genetic concepts and inheritance patterns. This clinic also offers opportunities for counseling and research study enrollment for patients and their families.

617-355-2711 / Genetic Counselor: 617-919-4371 / Fax: 617-919-2300

**Connective Tissue Disease Program** Provides multidisciplinary care for individuals of all ages with connective tissue diseases, a group of disorders with complex medical needs that can impact almost every organ system. The program includes physicians from different disciplines, including general and vascular surgery, orthopedic surgery, genetics, cardiology, pain management, hematology, dermatology, and rheumatology.

617-355-4697

#### **Craniofacial Anomalies Program**

Provides a multidisciplinary approach to the evaluation, diagnosis, and treatment of children and adults with congenital or acquired facial deformities.

617-355-6309

#### **Fragile X Program**

Provides an integrated, multidisciplinary approach to care for people with fragile X syndrome and their families by combining the expertise of clinicians and professionals from Genetics, Developmental Medicine, Psychology, Neurology and other medical services as well as community, educational and support services.

617-355-4697 / [childrenshospital.org/fragilex](http://childrenshospital.org/fragilex)

#### **Genetics Program**

Provides diagnosis and genetic counseling for pregnant women, newborns, children, and adults with known or suspected inherited diseases, especially congenital malformations and chromosomal disorders.

617-355-6394

#### **Galactosemia Program**

Provides comprehensive, multidisciplinary care for children and adults with galactosemia caused by GALT enzyme deficiency and the related disorders of GALE and GALK deficiencies.

857-218-4636

#### **Lysosomal Storage Program**

Provides a multidisciplinary service for the diagnosis and management of lysosomal storage diseases, by initial referral to the Metabolism Program in the Division of Genetics.

857-218-4636

#### **Metabolism Program**

Provides Comprehensive evaluations and follow-up for children, adolescents, and adults, with known or suspected metabolic disorders, acute or chronic in nature. The team is comprised of clinicians, nurse practitioners, nutritionists, social workers, psychologists, and genetic counselors.

857-218-4636 / [childrenshospital.org/metabolism](http://childrenshospital.org/metabolism)

#### **Neurofibromatosis (NF) Program**

Provides comprehensive diagnostic evaluations, follow-up care, and genetic counseling for individuals with known or suspected neurofibromatosis and related disorders. The program offers specialty care from multiple disciplines, including genetics, neurology, endocrinology, neurooncology, general surgery, orthopedics, neurosurgery, otolaryngology, ophthalmology, pain management, dermatology, plastic surgery, and neuropsychology.

857-218-4018 / [childrenshospital.org/nf](http://childrenshospital.org/nf)

#### **Pediatric Cancer Risk Program**

Provides a comprehensive, multidisciplinary approach to care for individuals and their families whose diagnosis or family history suggests an increase risk of cancer. With providers at both Children's Hospital Boston and Dana Farber Cancer Institute; the program provides genetic counseling, surveillance, management, and treatment for both children and adults with genetic predisposition to various types of cancer.

617-355-4388

## GENETICS

### Walsh, Christopher A., MD, PhD

Chief, Division of Genetics

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NPI: 1821053133

**Board Certifications:** Neurology

### Irons, Mira B., MD

Associate Chief, Division of Genetics

Chief, Clinical Genetics

Director, Accreditation Council for Graduate Medical

Education (ACGME) Internal Reviews

Associate Professor, Harvard Medical School

Children's Hospital Boston

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**Other Office:** Weymouth

NPI: 1134184666

**Board Certifications:** Clinical Genetics, Biochemical Genetics, Pediatrics

**Specialties:** dysmorphism; genetic counseling; genetics; neurofibromatosis; prenatal genetics; Smith-Lemli-Opitz syndrome

### Berry, Gerard, MD

Director, Metabolism Program

Professor of Pediatrics, Harvard Medical School

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**Board Certifications:** Pediatrics, Biochemical Genetics, Pediatric Endocrinology

**Specialty:** metabolic disorders

### Corzo, Deya, MD

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**Board Certifications:** Pediatrics, Genetics

**Specialties:** dysmorphism; lysosomal storage diseases and disorders

### Cox, Gerald F., MD, PhD

Assistant in Medicine

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NPI: 1811955479

**Board Certifications:** Clinical Genetics, Biochemical Genetics, Molecular Genetics, Pediatrics

**Specialties:** Barth syndrome; cardiomyopathy; dysmorphism; genetic eye diseases and disorders; disorders; genetics; lysosomal storage diseases and disorders; velo-cardio-facial syndrome (VCFS)

### Harris, David J., MD

Attending Physician

Lecturer in Pediatrics, Harvard Medical School

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**Board Certifications:** Clinical Genetics, Pediatrics Pediatrics

**Specialties:** chromosomal abnormalities; dysmorphism; genetic eye diseases and disorders; disorders; genetics; inborn errors of metabolism; neurogenetic disorders; prenatal genetics

### Hawley, Pamela P., MS

Licensed Genetic Counselor

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☎ 617-355-4697

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✉ pamela.hawley@childrens.harvard.edu

**Board Certifications:** Genetic Counseling

**Specialties:** cystic fibrosis; genetic counseling; genetics; hearing disorders and loss; hereditary hemorrhagic telangiectasia; muscular dystrophy

### Hecht, Leah E., MEd, RN, MSN

Nurse Practitioner, Metabolism Program

Clinical Coordinator

Children's Hospital Boston

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✉ leah.hecht@childrens.harvard.edu

**Board Certifications:** PNCB

**Specialties:** metabolic disorders; nursing

### Holm, Ingrid A., MD, MPH

Member, Bone Health Program

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NPI: 1356306914

**Board Certifications:** Pediatric Endocrinology, Medical Genetics

**Specialties:** bone health; genetics; genetics of endocrine disorders; sex chromosome abnormalities; abnormalities; skeletal dysplasias

### James, Philip M., MD, MPH

Physician in Medicine

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**Other Offices:** Beverly; Boston

NPI: 1982674123

**Board Certifications:** Pediatrics, Neonatal-Perinatal Medicine, Medical Genetics

**Specialties:** biochemical genetics; inborn errors of metabolism; medical genetics; newborn medicine medicine

### Levy, Harvey L., MD

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**Specialty:** biochemical genetics

### Lincoln, Sharyn, MS

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**Board Certifications:** Genetic Counseling

**Specialties:** chromosomal abnormalities; fragile X syndrome; genetic counseling; genetics

## GENETICS

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**Board Certifications:** Biochemical Genetics, Pediatrics

**Specialties:** fatty acid oxidation defects; inborn errors of metabolism; lysosomal storage diseases and disorders; organic acidemia

### **Neilan, Edward G., MD, PhD**

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**NPI:** 1245297480

**Board Certifications:** Pediatrics

**Specialties:** Cockayne syndrome; inborn errors of metabolism; mitochondrial disorders

### **See, Tricia M., ScM**

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**Board Certifications:** Genetic Counseling

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### **Martinez Ojeda, Mayra L., MD**

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**Board Certifications:** Pediatrics

**Specialties:** genetics; neurofibromatosis

### **Newton, Stephanie, MS, CGC**

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**Board Certifications:** Genetic Counseling

**Specialties:** genetic counseling; genetics; metabolic disorders

### **Seman, Ann R., MS**

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**Board Certifications:** Genetic Counseling

**Specialties:** genetic counseling; genetics

### **McGowan, Caroline, MS**

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**Board Certifications:** Genetic Counseling

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### **Picker, Jonathan D., MD, PhD**

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**Other Offices:** Lexington; Weymouth

**NPI:** 1225093552

**Board Certifications:** Clinical Genetics

**Specialties:** dysmorphism; fragile X syndrome; genetics; psychiatric genetics

### **Smith, Sharon E., MD**

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**NPI:** 1760472211

**Board Certifications:** Pediatrics, Medical Genetics Genetics

**Specialty:** genetics

### **Miller, David T., MD, PhD**

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**Other Offices:** Norwood; Waltham

**NPI:** 1710942909

**Board Certifications:** Clinical Genetics, Molecular Molecular Genetics, Pediatrics

**Specialties:** DNA diagnostics; genetics; genetics of autism; neurofibromatosis

### **Roberts, Amy E., MD**

*Director, Cardiovascular Genetics Research Program  
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Children's Hospital Boston

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**Other Office:** Lexington

**NPI:** 1306836515

**Board Certifications:** Pediatrics, Medical Genetics Genetics

**Specialties:** cardiovascular genetics; clinical genetics; Noonan syndrome

### **Stoler, Joan M., MD**

*Clinical Geneticist  
Assistant Professor of Pediatrics, Harvard Medical School*

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**Other Offices:** Lexington; Weymouth

**NPI:** 1740270107

**Board Certifications:** Clinical Genetics, Pediatrics Pediatrics

**Specialties:** connective tissue disorders; craniofacial anomalies or disorders; craniosynostosis; Ehlers-Danlos syndrome; genetics; genetics; Marfan syndrome

## GENETICS

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**Board Certifications:** Clinical Genetics, General Pediatrics

### **Woerner, Audrey C., MD, MPH**

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