

Pediatric Specialist Guide

A referral guide for health care providers



James Kasser, MD



Children's Hospital Boston

2009-2010

CHILDRENSHOSPITAL.ORG

GENETICS

Children's Hospital Boston's Division of Genetics offers state-of-the-art diagnostic and clinical management for children, adolescents and adults with known or suspected genetic conditions, including inborn errors of metabolism. The division consists of a multidisciplinary team of physicians, nurses and counselors who specialize in genetics, chromosome abnormalities, syndrome identification, metabolic disease, molecular genetic testing and birth defects.

The division has expertise in the following areas:

- Angelman syndrome
- brain malformations
- chromosome abnormalities
- connective tissue disorders
- fatty acid oxidation defects
- fragile X syndrome
- galactosemia
- homocystinuria
- MCAD
- methylmalonic acidemia
- MSUD
- neurofibromatosis
- organic acidemia
- phenylketonuria (PKU)
- Prader-Willi syndrome
- propionic acidemia
- sensorineural hearing loss
- skeletal dysplasia
- Smith Lemli Optiz syndrome
- velo-cardio-facial syndrome

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.

Scheduling: 617-355-2711

Genetic Counselor: 617-919-4371

Fax: 617-919-2300

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 Pediatrics, Psychology, Neurology and other medical services as well as community, educational and support services.

Scheduling: 617-355-4697

Genetics Program

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

Scheduling: 617-355-6394

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.

Scheduling: 617-355-6394

Metabolism Program

Evaluates children, adolescents and adults with known or suspected metabolic disorders, acute or chronic in nature.

Scheduling: 617-355-6394

Neurofibromatosis (NF) Program

Provides comprehensive diagnostic evaluations, follow-up care and genetic counseling for individuals with known or suspected neurofibromatosis.

Scheduling: 617-355-4697

GENETICS

Children's Division of Genetics provides outpatient services at our Boston campus on the 10th floor of the Fegan Building and in the following communities throughout Greater Boston:

COMMUNITY	LOCATION	SCHEDULING
Boston	Children's Hospital Boston 300 Longwood Avenue, Fegan-10	617-355-6394
Lexington	Children's Hospital Boston at Lexington 482 Bedford Street	781-672-2100
Norwood	Children's Hospital Physicians at Caritas Norwood Hospital 800 Washington Street	781-440-9619
Waltham	Children's Hospital Boston at Waltham 9 Hope Avenue	781-216-2100
Weymouth	Children's Hospital Physicians at South Shore Hospital Nevin Bldg., 851 Main Street, Suite 6	781-331-4715

To make an appointment at any of our office locations, please call the appropriate scheduling office telephone number above or visit childrenshospital.org/genetics and select Request An Appointment.

Urgent Appointment Triage

Genetics:

857-218-4637

Metabolism:

617-355-2521

GENETICS

Walsh, Christopher A., MD, PhD

Chief, Division of Genetics

Children's Hospital Boston

☎ 617-919-2923

☎ 617-919-2010

✉ christopher.walsh@childrens.harvard.edu

Board Certifications: Neurology

Corzo, Deya, MD

Assistant in Medicine

Instructor, Harvard Medical School

Children's Hospital Boston

☎ 617-355-4697

☎ 617-730-0466

✉ deyanira.corzo@childrens.harvard.edu

Board Certifications: Pediatrics, Genetics

Specialties: dysmorphology; lysosomal storage diseases and disorders; neurogenetic disorders

Hecht, Leah E., MEd, RN, MSN

Nurse Practitioner, Metabolism Program

Clinical Coordinator

Children's Hospital Boston

☎ 617-355-6394

✉ leah.hecht@childrens.harvard.edu

Board Certifications: PNCB

Specialties: metabolic disorders; nursing

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

☎ 617-355-4697

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✉ mira.irons@childrens.harvard.edu

Other Office: Weymouth

Board Certifications: Clinical Genetics, Biochemical Genetics, Pediatrics

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

Cox, Gerald F., MD, PhD

Assistant in Medicine

Instructor, Harvard Medical School

Children's Hospital Boston

☎ 617-355-4697

☎ 617-730-0466

✉ gerald.cox@childrens.harvard.edu

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

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

Holm, Ingrid A., MD, MPH

Associate Director, Bone Health Program

Director, Phenotyping Core, Program in

Associate Physician in Medicine

Assistant Professor of Pediatrics, Harvard

School

Children's Hospital Boston

☎ Bone Health: 617-355-6021

Endocrine: 617-355-7

☎ 617-730-0245

✉ ingrid.holm@childrens.harvard.edu

Board Certifications: Pediatric Endocrinology, Medical Genetics

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

Allard, Melissa A., MS

Genetic Counselor

Cockayne Syndrome Clinical Research

Coordinator

Children's Hospital Boston

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✉ melissa.allard@childrens.harvard.edu

Specialties: Cockayne syndrome; genetic counseling; genetics

Harris, David J., MD

Attending Physician

Lecturer in Pediatrics, Harvard Medical School

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✉ david.harris2@childrens.harvard.edu

Board Certifications: Clinical Genetics, Pediatrics

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

James, Philip M., MD, MPH

Physician in Medicine

Instructor in Pediatrics, Harvard Medical School

Children's Hospital Boston

☎ 617-355-6394

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✉ philip.james@childrens.harvard.edu

Other Offices: Beverly; Boston

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

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

Berry, Gerard, MD

Director, Metabolism Program

Professor of Pediatrics, Harvard Medical School

Children's Hospital Boston

☎ 617-355-6394

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✉ gerard.berry@childrens.harvard.edu

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

Specialty: metabolic disorders

Hawley, Pamela P., MS

Genetic Counselor

Children's Hospital Boston

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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

Lawrence, Janette Z., MS

Genetic Counselor

Angelman Syndrome Research Study Coordinator

Children's Hospital Boston

☎ 617-355-4697

☎ 617-730-0466

✉ janette.lawrence@childrens.harvard.edu

Other Office: Lexington

Specialties: Angelman syndrome; genetic counseling; genetics

GENETICS

Levy, Harvey L., MD

Senior Associate in Medicine
Professor of Pediatrics, Harvard Medical School

Children's Hospital Boston

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✉ harvey.levy@childrens.harvard.edu

Board Certifications: Pediatrics, Medical Genetics, Clinical Biochemical Genetics

Specialty: biochemical genetics

Marsden, Deborah L., MD

Assistant in Medicine
Assistant Professor, Harvard Medical School

Children's Hospital Boston

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☎ 617-730-0466

✉ deborah.marsden@childrens.harvard.edu

Board Certifications: Biochemical Genetics, Pediatrics

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

Picker, Jonathan D., MD, PhD

Director, Fragile X Program
Assistant in Medicine

Instructor, Harvard Medical School

Children's Hospital Boston

☎ 617-355-4697

☎ 617-730-0466

✉ jonathan.picker@childrens.harvard.edu

Other Office: Weymouth

Board Certifications: Clinical Genetics

Specialties: dysmorphology; genetics; psychiatric genetics

Lincoln, Sharyn, MS

Genetic Counselor
Clinical Coordinator, Fragile X Program

Children's Hospital Boston

☎ 617-355-4697

☎ 617-730-0466

✉ sharyn.lincoln@childrens.harvard.edu

Board Certifications: Genetic Counseling

Specialties: chromosomal abnormalities; fragile X syndrome; genetic counseling; genetics; genetics of autism

McGowan, Caroline, MS

Genetic Counselor
Coordinator, Neurofibromatosis Program

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✉ caroline.mcgowan@childrens.harvard.edu

Other Office: Waltham

Board Certifications: Genetic Counseling

Specialties: genetic counseling; genetics; neurofibromatosis

Reinhard, Ann E., MS

Genetic Counselor

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✉ ann.reinhard@childrens.harvard.edu

Other Office: Weymouth

Board Certifications: Genetic Counseling

Specialties: genetic counseling; genetics

Lowe, Kathryn, MS

Genetic Counselor

Children's Hospital Boston

☎ 617-355-4697

☎ 617-730-0253

✉ kathryn.lowe@childrens.harvard.edu

Specialties: genetic counseling; genetics

Miller, David T., MD, PhD

Instructor, Harvard Medical School

Children's Hospital Boston

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✉ david.miller2@childrens.harvard.edu

Other Offices: Norwood; Waltham

Board Certifications: Clinical Genetics, Molecular Genetics, Pediatrics

Specialties: congenital malformations; DNA diagnostics; dysmorphology; genetics; genetics of autism

Roberts, Amy E., MD

Director, Cardiovascular Genetics Research Program

Associate in Cardiology

Instructor of Medicine, Harvard Medical School

Assistant Professor of Clinical Genetics, Harvard Medical School

Children's Hospital Boston

☎ 617-355-6529

☎ 617-525-4488

✉ amy.roberts@childrens.harvard.edu

Board Certifications: Pediatrics, Clinical Genetics

Specialties: cardiovascular genetics; clinical genetics; Noonan syndrome

Lyon, Helen N., MD, SM

Assistant in Medicine

Medical Director, Department of Clinical

Beth Israel Deaconess Medical Center

Instructor in Pediatrics, Harvard Medical School

Children's Hospital Boston

☎ 617-355-4697

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✉ helen.lyon@childrens.harvard.edu

Other Office: Boston

Board Certifications: Pediatrics, Clinical Genetics

Specialties: cardiovascular genetics; clinical epidemiology; clinical genetics; prenatal genetics

Neilan, Edward G., MD, PhD

Assistant in Medicine

Instructor in Pediatrics, Harvard Medical School

Children's Hospital Boston

☎ 617-355-6394

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✉ edward.neilan@childrens.harvard.edu

Board Certifications: Pediatrics

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

Smith, Sharon E., MD

Assistant in Medicine

Instructor, Harvard Medical School

Children's Hospital Boston

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✉ sharon.smith@childrens.harvard.edu

Board Certifications: Pediatrics, Medical Genetics

Specialty: genetics

GENETICS

Stoler, Joan M., MD

*Clinical Geneticist
Assistant Professor of Pediatrics, Harvard
School*

Children's Hospital Boston

☎ 617-355-4697

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✉ joan.stoler@childrens.harvard.edu

Other Offices: Lexington; Weymouth

Board Certifications: Clinical Genetics,
Pediatrics

Specialties: craniofacial anomalies or
disorders; craniosynostosis; genetics;
plagiocephaly

Tan, Wen-Hann, MD

*Attending
Instructor in Pediatrics, Harvard Medical School*

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Board Certifications: Clinical Genetics,
General Pediatrics