Pediatric Medical Genetics
Consult and Referral Guidelines

25 Michigan Street NE

About Pediatric Medical Genetics

We see both pediatric and adult patients. In many cases, our initial evaluation of the patient will result in testing/evaluation of multiple family members, as genetic testing results often have a wide-reaching impact.

Most common referrals

- Known/suspected genetic conditions such as Down syndrome, Noonan syndrome, Turner syndrome, etc.
- Single/multiple congenital anomalies and dysmorphic features
- Counseling for previous genetic testing results
- Family history of a genetic condition
- Neurodevelopmental disorders, such as autism spectrum disorders, intellectual disabilities/cognitive disabilities and developmental delays
- Neurologic conditions, including cerebral palsy, hyper/hypertonia, muscular dystrophies, ataxias and seizure disorders where individuals appear syndromic or have additional health/developmental concerns
- Cardiovascular disease, including congenital heart defects, cardiomyopathy, long QT syndrome, arrhythmias
- Abnormal growth, including short stature/growth restriction, overgrowth and hemihypertrophy/hemihyperplasia
- Fetal alcohol spectrum disorders
- Preconception counseling

Pediatric Medical Genetics Appointment Priority Guide

Immediate **Urgent**	A genetics physician is on call 24/7 and can be reached by PerfectServe, Epic in basket, email or by calling our main medical genetics clinic phone number (616.391.2700) during business hours or by calling HDVCH Direct (616.391.2345) after hours or weekends.
Routine	A genetic counselor is on call during business hours and can be reached by calling our main medical genetics clinic phone number.

Diagnosis/Symptom	Suggested Workup/Initial Management	When to Refer	Information Needed
General	We typically do not recommend ordering genetic or other testing prior to referral. We will order or recommend studies pre- or post-evaluation if appropriate. If referring providers wish to begin studies or discuss this prior to the initial appointment, please contact our team for assistance.	• Family member with confirmed genetic disorder • Suspicion of a genetic disorder in patient without previously diagnosed family member	• All previous genetic testing results (for patient, or for family member if referral is for family history of condition) • Family history, including specific name of the condition of concern and which family member(s) is affected • Specific condition of concern • Clinic notes from other subspecialty evaluations not viewable in Epic. • Relevant lab and/or imaging results. • Relevant neuropsychological evaluation reports. • Growth charts

Indications we do not routinely see in our office

- Personal or family history of cancer
- Testing for an asymptomatic pediatric patient for adult-onset conditions or carrier status. Examples of this include certain types of muscular dystrophy, Huntington's disease, etc. The American Academy of Pediatrics (APP) and the American College of Medical Genetics (ACMG) do not support carrier testing in minors when it is not medically relevant. Carrier screening is appropriate for adolescents who are pregnant or are considering reproduction.
- Ehlers-Danlos Syndrome (EDS), Hypermobile Type. We do not determine eligibility for disability, prescribe medication or manage care of individuals with EDS-hypermobility type and are not scheduling patients for this indication at this time. For a detailed summary of the condition, please see the Gene Reviews article on Ehlers-Danlos, Hypermobility Type available here.
- Personal or family history of Alzheimer's disease (AD), when age of onset is greater than 50 years of age. Genetic testing is only recommended in cases of early-onset AD, prior to age 50, as there is more likely to be a single gene cause identified in these individuals. The ACMG, the National Society of Genetic Counselors and the National Institutes of Health recommend against using APOE testing as a pre-symptomatic or predictive test for AD.
- MTHFR testing or counseling of previous abnormal results, with the exception of homocystinuria caused by MTHFR mutations (characterized by abnormal blood clotting, spinal cord defects, vision and cognitive deficits; this condition typically presents in infancy and with significantly elevated homocysteine levels). Given the lack of evidence between the common MTHFR variants and disease, as well as lack of utility for determining medical management, several professional societies have released guidelines regarding MTHFR. Specifically, the ACMG, American Academy of Family Physicians (AAFP) and the American College of Obstetrics and Gynecology (ACOG) recommend against testing for the common MTHFR variants as part of routine clinical evaluation, or in the presence of a family history of MTHFR. The ACMG practice guidelines are available here.
- Testing for personal or family history of autoimmune conditions, such as multiple sclerosis, HLA-B27 testing, lupus, arthritis, etc. Due to the lack of clinical genetic testing for autoimmune conditions, referrals for autoimmune conditions are not currently being processed for genetics services. We hope to be able to provide services to these patients in the future, as our knowledge of the genetics of autoimmune conditions continues to expand.

Spectrum Health and Helen DeVos Children's Hospital have other specialty clinics for some genetic disorders and certain patient types and indications. If there is any uncertainty about where to refer a patient, please contact our main medical genetics clinic for assistance at 616.391.2700.

For pregnancy-related genetic concerns (e.g., family history of genetic disorder, abnormal prenatal screening) refer to Spectrum Health Maternal Fetal Medicine clinic: 616.391.3681.

For pediatric patients **affected** with cancer or other features of a potential hereditary cancer condition (e.g., familial adenomatous polyposis, Cowden syndrome, Gorlin syndrome, Li-Fraumeni syndrome, juvenile polyposis syndrome, retinoblastoma, Peutz-Jeghers syndrome, MEN1, MEN2, hereditary paraganglioma-pheochromocytoma syndrome), refer to Spectrum Health Cancer Genetics clinic: 616.486.6218.

For **unaffected** pediatric patients with a known family history of an adult-onset hereditary cancer condition (e.g., BRCA-related hereditary breast and ovarian cancer syndrome, Lynch syndrome), genetic testing is often deferred until the patient is 18 or older, as cancer screenings would not begin during childhood. However, if there are uncharacteristically young cancers in the family (diagnosed at age 28 or younger), referral to Spectrum Health Cancer Genetics clinic and consideration of genetic testing for these (usually) adult-onset conditions is appropriate.

For **unaffected** pediatric patients with a family history of cancer, referral of parent(s) to Spectrum Health Cancer Genetics clinic is recommended to identify or rule out hereditary cancer risk to their child.

For metabolic, biochemical and mitochondrial genetic conditions or inborn errors of metabolism refer to Helen DeVos Children's Hospital Biochemical Genetics clinic: 616.486.9830

For abnormal newborn screen results, the patient's results report will indicate the appropriate contact specific to the abnormality. For questions pertaining to newborn screening, please contact the Helen DeVos Children's Hospital Biochemical Genetics clinic: 616.486.9830

Other specialty clinics

- For adult patients with hypertrophic cardiomyopathy (HCM) to consider genetic testing or for first degree relatives of someone with HCM for cardiac surveillance and/or genetic testing recommendations, refer to Spectrum Health Cardiovascular Medicine HCM Clinic: 616.885.5192
- For patients with suspected or confirmed in utero alcohol exposure or suspected Fetal alcohol spectrum disorder (FASD), refer to HDVCH Medical Genetics FASD Clinic: 616.391.2700
- For patients with suspected or confirmed spinal muscular atrophy, Charcot Marie Tooth Disease or Duchenne muscular dystrophy, refer to the HDVCH Neuromuscular Clinic: 616.267.2500
- For patients with suspected or confirmed genetic epilepsy not requiring a dysmorphology exam, refer to HDVCH Epilepsy Genetics Clinic: 616.267.2500
- For patients with suspected or confirmed cystic fibrosis, refer to HDVCH pulmonology clinic and cystic fibrosis care center: 616.267.2200
- For pediatric patients with suspected or confirmed cardiomyopathy or arrhythmia or a family history of cardiomyopathy or arrhythmia, refer to HDVCH cardiogenetics clinic: 616.267.9150
- For patients with oral clefts, refer to the HDVCH oral cleft clinic coordinated by Pediatric Plastic Surgery: 616.486.5885
- For patients with suspected or confirmed Huntington's disease, refer to Medical Genetics clinic: 616.391.9007
- For adult patients with suspected or confirmed Neurofibromatosis type 1, refer to HDVCH Neurofibromatosis Clinic: 616.391.2414
- For pediatric patients with suspected or confirmed Neurofibromatosis type 1, refer to HDVCH Pediatric Neurology clinic: 616.267.2500
- For patients with suspected or confirmed Tuberous Sclerosis, refer to HDVCH Pediatric Neurology clinic: 616.267.2500