Medical Genetics

Consult and Referral Guidelines

Diagnosis/Symptom	Suggested Workup/Initial Management	When to Refer	Additional Information Needed
Cognitive Impairment/ Developmental Delay/Autism	Consider referring to Early On (<3 years old) Consider having the patient evaluated to determine extent of delays/impairment.	Diagnosis of cognitive impairment. Persistent unexplained developmental delays. Family history of developmental delay/cognitive impairment.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies reports (if done) • Imaging reports (if done)
Hypotonia/Hypertonia	Consider referring to neurology	Unexplained persistent hypertonia or hypotonia.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done)
Major or minor physical anomalies or birth defects	Refer to appropriate system specialist. E.g.; cardiology, ophthalmology, plastic surgery, nephrology, ENT, etc.	Presence of any major anomaly or birth defect. If there is a pattern of anomalies that appears syndromic. For multiple minor anomalies Associated with cognitive impairment or developmental delay. Family history of similar anomalies.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done)

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Fetal Alcohol Syndrome	Consider referring for psychological evaluation or neuropsychological evaluation.	Known or suspected prenatal exposure to alcohol.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done) • Any available IEP reports or psychology records or neuropsychology evaluations.
Deafness	Consider referring to ENT Consider referring to audiology Consider temporal imaging (CT scan)	Unexplained hearing loss. Family history of hearing loss or deafness.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done) • Audiology reports
Failure to thrive or significant overgrowth	Consider referring to endocrinology Consider referring to gastroenterology	Unexplained overgrowth or undergrowth	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done)

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Hemihypertrophy	Blood AFP level (every 3 months until genetics visit) Abdominal ultrasound (every 3 months until genetics visit).	Asymmetry of the face, body, limbs or digits.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done) • Blood AFP levels • Abdominal ultrasound reports
Known Genetic Condition		Known genetic condition (Such as Duchenne Muscular Dystrophy, Turner syndrome, Down syndrome, 22q11.2 deletion syndrome, other chromosome anomalies)	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done) • Records confirming the clinical or laboratory diagnosis
Family history of a genetic condition	Obtain written records on relative with known genetic condition. This is critical to achieving a productive visit in our office)	Concern for a child to inherit a familial genetic condition.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done)

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Connective tissue disorders	Consider referring to cardiology Consider ordering echocardiogram (with assessment of aortic root) Consider referring to ophthalmology	Concern for a connective tissue disorder in a child.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done)
Disorders of sexual development	Consider referring to endocrinology Consider referring to gynecology or urology as appropriate Consider pelvic imaging (Typically ultrasound) Consider a karyotype (also known as chromosome analysis) Consider a FISH probe for the SRY region http://spectrumhealth.testcatalog.org/show/974	Concern for a disorder of sexual development.	Medical records including: • Work up from other pediatric specialists (if done) • Most recent primary care visit record • Growth charts • Genetic laboratory studies (if done) • Imaging reports (if done)

Please note:
We do not see patients for the following indications:

- **Pregnant patients for counseling regarding the pregnancy** *(Instead refer to Spectrum Health Maternal Fetal Medicine Ph: 616 391 3681 Fax: 616 391 8670 or Dr. Russel Jelsema at West Michigan Ob/Gyn Ph: 616 774 7035 fax:616 774 4057)*
- **Patients with suspected or known inherited cancer syndromes** *(Instead refer to Spectrum Health Cancer Genetics Ph: 616 486 6218, Fax 616 486 6110)*
- **Patients with abnormal newborn screening** *(Follow instead the recommendations on the information received from the newborn screen protocol) We will see these patients following the confirmation of the diagnosis as recommended by the State of Michigan NBS if needed.*
- **Patients with confirmed inborn errors of metabolism** *(Refer to Children's Hospital of Michigan Metabolic Clinic Ph:866 442 4662, Fax: 313 745 8030)*
- **Patients with suspected or confirmed neurofibromatosis type 1** *(Instead refer to the HDVCH NF Clinic Ph: 616 391 2414, Fax: 616 391 2505)*
- **Patients with suspected or confirmed cystic fibrosis** *(Instead refer to the HDVCH pulmonology clinic/ CF care center Ph: Fax:*