Medical Genetics
Consult and Referral Guidelines
HDVCH has developed these consult and referral guidelines as a general reference tool to assist referring physicians with the specialty referral process. Pediatric medical needs are complex and these guidelines may not apply in every case. HDVCH relies on its referring providers to exercise their own professional medical judgment with regard to the appropriate treatment and management of their patients. Referring providers are solely responsible for confirming the accuracy, timeliness, completeness, appropriateness and helpfulness of this material and making all medical, diagnostic or prescription decisions.

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<th>Suggested Workup/Initial Management</th>
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| 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. | 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 hypotonia or hypertonia.                             | 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. | 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) |

For the most updated version of the Referral Guidelines, please visit www.helendevoschildrens.org
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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.                   | 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

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| 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

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| 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

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| 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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<td>Connective tissue disorders</td>
<td>Consider referring to cardiology&lt;br&gt;Consider ordering echocardiogram (with assessment of aortic root)&lt;br&gt;Consider referring to ophthalmology</td>
<td>Concern for a connective tissue disorder in a child.</td>
<td>Medical records including:&lt;br&gt;• Work up from other pediatric specialists (if done)&lt;br&gt;• Most recent primary care visit record&lt;br&gt;• Growth charts&lt;br&gt;• Genetic laboratory studies (if done)&lt;br&gt;• Imaging reports (if done)</td>
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<td>Disorders of sexual development</td>
<td>Consider referring to endocrinology&lt;br&gt;Consider referring to gynecology or urology as appropriate&lt;br&gt;Consider pelvic imaging (Typically ultrasound)&lt;br&gt;Consider a karyotype (also known as chromosome analysis)&lt;br&gt;Consider a FISH probe for the SRY region <a href="http://spectrumhealth.testcatalog.org/show/974">http://spectrumhealth.testcatalog.org/show/974</a></td>
<td>Concern for a disorder of sexual development.</td>
<td>Medical records including:&lt;br&gt;• Work up from other pediatric specialists (if done)&lt;br&gt;• Most recent primary care visit record&lt;br&gt;• Growth charts&lt;br&gt;• Genetic laboratory studies (if done)&lt;br&gt;• Imaging reports (if done)</td>
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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: 616 391 2414, Fax: 616 391 2505)*

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