



Valley Children's Genetics and Metabolism

Valley Children's team of board-certified or board-eligible medical geneticists and five licensed and certified genetic counselors, specialize in the care and treatment of hereditary rare diseases. Our services include comprehensive diagnostic evaluations, including facilitating genetic testing when indicated, clinical management recommendations and genetic counseling.

The accuracy of genetic testing is highly dependent on establishing a comprehensive phenotype. Valley Children's Medical Genetics and Metabolism department collaborates with many other specialties including but not limited to imaging, neurology, audiology, endocrinology, gastroenterology, cardiology, ophthalmology and orthopedics to improve phenotyping to establish diagnoses.

Just as there are thousands of different genetic diseases, there are numerous indications for referral to medical genetics and metabolism. For referred patients, please provide the following:

- Any past genetic testing laboratory results – brief description of result is insufficient, we must have the original test report
- Clinic notes, including comprehensive history summary
- Medication history
- Growth charts/curves
- Relevant lab reports
- Imaging and diagnostic reports
- Previous specialty evaluations

For urgent cases, such as when a metabolic disorder is suspected, please specify this information on the referral or speak with on-call our geneticists on call by calling our clinic at 559-353-6400.

Access Center

24/7 access for referring physicians
866-353-KIDS (5437)

Outpatient Referral

Referral forms online at valleychildrens.org/refer
FAX: 559-353-8888

Medical Genetics and Metabolism Office Numbers

Main: 559-353-6400
FAX: 559-353-7213

Referral Indication	Include with Referral
Autism and/or Intellectual Disability (ID)	<ul style="list-style-type: none"> • Documentation of autism diagnosis (preferably formal developmental evaluation) <ul style="list-style-type: none"> • Please connect patients with appropriate early intervention resources before referral • Consider referral to genetics particularly for patients with ID or autism AND one of the following: <ul style="list-style-type: none"> a. Presence of developmental regression/stagnation b. Presence of other system(s) affected c. Patients that did not undergo newborn screening testing (for example if they were born in a foreign country where NBS is not available) d. Presence of dysmorphic features
Marfan Syndrome	<ul style="list-style-type: none"> • Ophthalmology evaluation • Echocardiogram (with cardio consult as needed)
Family History of Genetic Disease in a Healthy Child	<ul style="list-style-type: none"> • Obtain written records/genetic testing report on relative with known genetic condition (this is critical to achieving a productive visit in our office)
Hemihypertrophy	<ul style="list-style-type: none"> • Growth charts • Imaging reports and/or orthopaedics consult (recommended) documenting the discrepancy • Blood AFP level (every 3 months until genetics visit) • Abdominal ultrasound (every 3 months until genetics visit)
Failure to Thrive	<ul style="list-style-type: none"> • Growth charts • Referral to gastroenterology and/or endocrinology recommended (send documentation of these consults if already performed) • Results of initial blood work up (including at least complete metabolic panel and complete blood count)
Hearing Loss	<ul style="list-style-type: none"> • Audiology and/or ENT evaluation documentation • Consider temporal imaging (CT scan)

Referral Indication	Include with Referral
Known Genetic Condition	<ul style="list-style-type: none"> • Lab report confirming diagnosis - a written description of the result (eg: "down syndrome" is NOT sufficient)
Epilepsy	<ul style="list-style-type: none"> • Neurology consultation notes • EEG results
Obesity	<ul style="list-style-type: none"> • Growth charts • Consider referral to gastroenterology and/or endocrinology before or consider referring only when genetic syndrome is suspected
Hypotonia	<ul style="list-style-type: none"> • Neurology consultation notes
Short Stature	<ul style="list-style-type: none"> • Referral to endocrinology recommended, send documentation of this consult if already performed • Skeletal survey if there is concern for skeletal dysplasia