

Genetics Referral Guidelines

The information below is needed for each type of referral.

Please send the documentation listed for a referral. All information for a patient should be sent together. If you are unable to send the requested documentation, forward the referral to Genetics for review.

All Referrals

*Diagnosis: **All Diagnoses***

- Clinic notes with the reason for referral
- Copy of any laboratory and/or radiology tests that support the reason for referral
- Copy of any genetic testing reports

*Diagnosis: **Family History of a Genetic Disorder***

- Clinic notes with the reason for referral
- Copy of any laboratory and/or radiology tests that support the reason for referral
- Copy of any genetic testing reports
- Copy of any reports if a family member is affected and has had genetic testing. If not, clinic notes that support his/her diagnosis.

For Specific Diagnoses (most frequent referrals)

*Diagnosis: **Autism***

- Clinic notes with the reason for referral
- Developmental notes and/or any tool used for the diagnosis and/or DSM-5 criteria
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Developmental Delay***

- Clinic notes with the reason for referral
- Developmental notes and/or speech evaluation and/or clear description of milestones
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Down Syndrome***

- Clinic notes with the reason for referral
- Copy of the genetic testing report (karyotype/microarray)

*Diagnosis: **Ehlers-Danlos Syndrome/Hypermobility***

- Clinic notes with the reason for referral
- Echocardiogram, or request to have one scheduled
- Dilated eye exam, or request to have one scheduled
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Failure to Thrive***

- Clinic notes with the reason for referral
- If available, endocrinology notes
- Copy of growth charts (height and weight)
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Klinefelter Syndrome***

- Clinic notes with the reason for referral
- Copy of the genetic testing report (karyotype/microarray)

*Diagnosis: **Macrocephaly/Microcephaly***

- Clinic notes with the reason for referral
- Copy of growth charts (height, weight and head circumference)
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Marfan Syndrome***

- Clinic notes with the reason for referral
- Echocardiogram, or request to have one scheduled
- Dilated eye exam, or request to have one scheduled
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Neurofibromatosis/Café-Au-Lait Spots***

- Clinic notes with the reason for referral
- If patient referred is ONE YEAR or OLDER: dilated eye exam, or request to have one scheduled
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Prader-Willi/Obesity***

- Clinic notes with the reason for referral
- Copy of growth charts (height and weight)
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Short Stature***

- Clinic notes with the reason for referral
- Copy of growth charts (height and weight)
- Copy of the genetic testing report if patient has had genetic testing

*Diagnosis: **Turner Syndrome***

- Clinic notes with the reason for referral
- Copy of the genetic testing report (karyotype/microarray)
- If patient has had an echocardiogram and/or kidney ultrasound, a copy of those reports

*Diagnosis: **Speech Delay***

- Clinic notes with the reason for referral
- Speech evaluation notes and/or clear description of milestones
- Copy of hearing evaluation
- Copy of the genetic testing report if patient has had genetic testing