Pediatric Genetics Referrals: When to Refer to Genetics

- Genetic risk assessment and testing can be helpful in aiding in the care of individuals with a personal or family history of genetic conditions.
- Some genetic conditions are inherited through either **males or females**, while others can be inherited through **both males and females**.
- Those with an unknown or limited family history can still be referred to genetics for evaluation.
- Genetic test results can:
  - Impact treatment and management recommendations
  - Inform unaffected individuals of possible future risks to themselves or future children
  - Guide appropriate screenings

General Referral Guidelines for Pediatric Indications

A genetics evaluation should be considered for individuals with any of the following:

- A **known genetic pathogenic variant/mutation identified** in an individual or family member
- A known or suspected **genetic disorder, birth defect, or chromosomal abnormality**
- A **newborn** with any of the following:
  - Abnormal newborn screening test result
  - Congenital hypotonia or hypertonia
  - Unexplained intrauterine growth retardation
- A **newborn, infant, or child** with any of the following:
  - A **single major anomaly**, or **multiple major and/or minor anomalies**
  - **Dysmorphic features** that are not familial
  - **Failure to thrive**
  - A known **metabolic disorder** or symptoms of a metabolic disorder (Note: *A normal newborn screening result does not rule out all metabolic disorders)*
  - **Abnormal brain MRI findings**
  - **Unusual growth patterns**
  - Evidence of a **connective tissue disorder**
  - **Congenital eye defects** or **blindness**
  - Significant **hearing loss** or **deafness**
  - **Cardiomyopathy** not secondary to a viral infection
  - **≥ 6 café-au-lait macules** at least 0.5 cm in diameter
  - **Unusual skin findings** such as multiple types of lesions, multiple lipomas, numerous hypo– or hyperpigmented lesions, or albinism
  - Bilateral or multifocal malignancies
- A **child** with:
  - **Developmental delay**
  - **Intellectual disability**
  - **Autism Spectrum Disorders**
  - **Immunodeficiency**
  - Progressive **muscle weakness**

Full referral guidelines are referenced on the back of this handout. If you are concerned about a patient's personal or family history, a genetics professional can help determine if further risk assessment and/or genetic testing is indicated.

Take Action

If your patient meets any of these criteria:

- Talk to your patient and/or your patient's family about recommendations for a genetics referral
- Emphasize the importance of making and keeping an appointment with a genetics provider
- For assistance locating the nearest genetics service provider, in the New York-Mid-Atlantic Consortium (NYMAC) Region, please contact the **Genetic Services Referral Call Center** at **1-833-545-3218** or visit our website.

Guidelines and Recommendations (Links)

The Professional Practice and Guidelines Committee of the American College of Medical Genetics and Genomics (ACMG) Indication for genetic referral: a guide for healthcare providers

American Academy of Pediatrics—Genetics in Primary Care Institute, When to Refer

Resources (Links)

- **National Society of Genetic Counselors (NSGC) Find a Genetic Counselor**
  A tool developed by NSGC for patients and providers to locate genetic counseling services in North America (U.S. and Canada)
- **Baby's First Test**
  A website providing information about newborn screening and resources for families caring for a child with a genetic condition
- **GeneReviews through the NIH**
  Resource for providers about the diagnosis and management of patients with genetic conditions

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